Informed Decision-Making for Genetic Testing: Public Discourse and Individual Choice

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"I want to encourage every woman, especially if you have a family history of breast or ovarian cancer, to seek out the information and medical experts who can help you through this aspect of your life, and to make your own informed choices."

Angelina Jolie, The New York Times, 14 May 2013

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Summary

It can be challenging for individuals to make an informed and reflected decision on whether to take a predictive genetic test, which provides a disease risk assessment in asymptomatic individuals. Since too much information might lead to information overload, adequate information is necessary for an informed decision. However, economic interests and the increasing availability of predictive genetic testing due to scientific advancements and direct-to-consumer services lead to exaggerated expectations and an information bottleneck. Thus, this dissertation aims to examine what information about genetic testing should be given to at-risk individuals and the public to foster informed decision making and to assess how such information is best transmitted.

To investigate these complex issues, this dissertation employed a mixed methodology and focused on the situation in Switzerland. Quantitative and qualitative newspaper content analysis was applied to assess what information is transmitted to the public and compared reporting between the UK and Switzerland. In parallel, Swiss individuals receiving genetic counselling for cancer predisposition genetic testing were interviewed about their reasons, attitudes and information strategies during the genetic testing decision-making process. In line with previous literature, this qualitative study revealed the individuality of the decision-making process and the influence of life philosophy on the decision. Individuals also expressed differing information needs. We identified motivators of and barriers to information-seeking behaviour, for instance, prior knowledge, the doctor-patient relationship, the decision-making style, and the individual life philosophy. The newspaper content analyses showed that coverage about predictive genetic testing was substantially higher and more positively evaluated in the UK compared to Switzerland. Moreover, UK newspapers only mentioned ethical issues on the individual, but rarely on the societal level.

In conclusion, societal issues should be the main focus of mass media coverage regarding predictive genetic testing because of their arena function. Mass media should not be the primary public source of information for predictive genetic testing, as they inherently tend to exaggerate claims and might not be interested in providing general background knowledge. Additionally, I suggest stratifying individuals according to their information needs and set up online

information platforms that provide holistic and unbiased information and decision support. In certain circumstances, individuals with low information needs and high prior knowledge could waive genetic counselling and still make an informed decision regarding predictive genetic testing. This might become necessary if genetic counselling resources become scarce due to the increasing demand.

As a secondary aim, this dissertation assessed how media content analyses could be useful for bioethics. They can serve as pilot studies for bioethical inquiries, to make recommendations on how to improve science communication from an ethical perspective, as an indicator for public engagement, and to investigate social phenomena relevant to bioethics.

Zusammenfassung

Prädiktive genetische Untersuchungen, welche das genetische Risiko für eine Krankheit (beispielsweise Krebs) bestimmen, gehen mit der herausfordernden Entscheidung einher ob ein solcher Test überhaupt erwünscht ist. Da zu viele Informationen zu Überforderung führen können, sind adäquate und individuell abgestimmte Informationen angezeigt. Die steigende Verfügbarkeit solcher Tests dank wissenschaftlicher Fortschritte und das Aufkommen von Firmen, die solche Tests online zum Verkauf anbieten führen jedoch zu übertriebenen Erwartungen der Bevölkerung und zu einem Informationsengpass. Deswegen hat diese Dissertation zum Ziel, zu untersuchen welche Informationen an Betroffenen und an die Allgemeinbevölkerung abgegeben werden sollen, um eine informierte Entscheidungsfindung bezüglich prädiktiver genetischer Untersuchungen zu fördern; und zu evaluieren wie solche Informationen am besten verbreitet werden.

Um diese komplexen Fragestellungen zu untersuchen, wurden zwei empirische Projekte durchgeführt, die sich auf die Situation in der Schweiz fokussierten. Erstens wurden Zeitungsinhaltsanalysen durchgeführt um die Art der durch Massenmedien verbreiteten Informationen zu evaluieren. Dazu wurde die Deutschschweizer Berichterstattung mit derjenigen von Grossbritannien verglichen. Zweitens wurden Interviews mit Erwachsenen durchgeführt, die eine genetische Beratung für vererbbare Krebserkrankungen in Anspruch genommen und sich mit der Entscheidung für oder gegen eine prädiktive genetische Untersuchung auseinander gesetzt haben. Diese qualitative Studie zeigte, dass die Entscheidung durch zahlreiche individuelle interne und externe Faktoren, vor allem von der eigenen Lebensphilosophie, beeinflusst wird. Der Informationsbedarf war sehr unterschiedlich, und wir identifizierten Faktoren die die Informationsbeschaffung förderten oder behinderten. Dazu gehörten das Vorwissen der Teilnehmenden, das Arzt-Patienten-Verhältnis, der Entscheidungsfindungsstil, und die Lebensphilosophie. Die Zeitungsanalyse zeigte, dass britische Tageszeitungen deutlich häufiger und positiver über prädiktive genetische Untersuchungen berichteten als Schweizer Tageszeitungen. Ausserdem wurden ethische Probleme nur auf der individuellen, nicht aber auf der gesellschaftlichen Ebene diskutiert.

Aufgrund ihrer Arenafunktion sollten Tageszeitungen sich jedoch mehr auf die gesellschaftlichen Fragestellungen bezüglich prädiktiver genetischer Untersuchungen konzentrieren und sollten nicht die Hauptinformationsquelle sein für Menschen, die sich überlegen einen prädiktiven Gentest zu machen. Dies deswegen weil Massenmedien inhärent zu Übertreibungen und Sensationalisierung neigen und aufgrund ihrer Ausrichtung an Nachrichtenwerten wenig daran interessiert sind, differenzierte Hintergrundinformationen zu prädiktiven Gentests zu liefern.

Daneben schlage ich vor, die Menschen nach ihrem Informationsbedarf einzuteilen und die Informationsbeschaffung auf weitere Kanäle auszuweiten. Eine elektronische Informationsplattform mit unabhängigen und ausgewogenen Hintergrundinformationen und Entscheidungshilfen ist dabei ein grundlegender Bestandteil. Unter bestimmten Umständen könnten manche dann auf eine persönliche genetische Beratung verzichten und dennoch eine informierte Entscheidung treffen. Dies könnte notwendig werden, sollte es aufgrund der gestiegenen Nachfrage zu einem Engpass im genetischen Beratungsangebot kommen.

Zusätzlich hatte diese Dissertation zum Ziel, die Nützlichkeit von Medieninhaltsanalysen für die Bioethik zu evaluieren. Medieninhaltsanalysen können als Pilotstudien für bioethische empirische Untersuchungen fungieren. Sie können zu Empfehlungen führen, wie die Wissenschaftskommunikation aus ethischer Perspektive verbessert werden könnte. Ausserdem können Medieninhaltsanalysen Indikatoren sein für die Einbindung der Öffentlichkeit in wissenschaftsrelevante Debatten, und können genutzt werden um bioethisch relevante soziale Phänomene zu untersuchen.

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List of Abbreviations

ACR	Article Count Ratio
BD	Big Data in Medical Genetics and Health
DTC-GT	Direct-to-consumer genetic testing
GEd	Genome editing
GRe	Genetic research
GTBH	Genetic Testing on Born Humans
GTh	Gene therapy
HBOC	Hereditary Breast and Ovarian Cancer
HGD	Human genetic diseases
HGTA	Human Genetic Testing Act
PEST	Public Engagement with Science and Technology
PGD	Preimplantation genetic diagnosis
PGT	Predictive Genetic Testing
PM	Personalized medicine
PPGT	Preimplantation and prenatal genetic testing
PUS	Public Understanding of Science
SMA	Spinal muscular atrophy

Chapter 1: Introduction

1.1 Dissertation structure

Genetic testing is becoming widespread and concerns an increasing number of people. Especially predictive genetic testing can have far-reaching medical, psychological and social consequences. Because receiving relevant information is vital for informed genetic testing decision-making, this dissertation analyses how and to what extent information about predictive genetic testing should be provided on a public and an individual level.

Chapter 1 includes a general introduction to this dissertation. Chapter 1.2 introduces genetic testing applications, genetic counselling and direct-to-consumer genetic testing and defines predictive genetic testing. In chapter 1.3, the theoretical background from philosophy, bioethics and the media and communication sciences is presented. Chapter 1.4 describes societal challenges related to genetic testing and illustrate the relevance of the dissertation. The objectives of this dissertation are presented in chapter 1.5.

Chapter 2 explains the methodology and entails a critical reflection on the strengths and weaknesses of the methodologies chosen for this dissertation and the connection between empirical and normative work. It also provides an overview of the contributions of the researchers and students involved in generating the results of this dissertation.

Chapters 3-5 show the results of an interview study with people undergoing genetic counselling for inherited cancer predisposition. These results explain the role of life philosophy in the genetic testing decision-making process (chapter 3), examine what motivators and barriers people face when collecting information during the decision-making process (chapter 4) and focus on the role of the social network (chapter 5).

Chapters 6-8 present results from the newspaper content analyses. Chapter 6 entails a screening of Swiss print media about medical genetics in general, indicating what topics caused most media attention between 2013 and 2016. In chapter 7, a country comparison on broadsheet newspaper coverage of predictive genetic testing is revealed, showing that UK coverage is more extensive and more positive than Swiss coverage. Chapter 8 includes a semi-quantitative analysis of the ethical aspects covered by UK broadsheets.

Chapter 9 includes a theoretical discussion on the extent the public should be informed about genetic testing. Finally, chapter 10 presents the discussion of this dissertation. It includes practical recommendations for fostering informed genetic testing decision-making through public education and for genetic counselling in the context of scarce resources (chapter 10.2). Chapter 10.3 elaborates on the applications of newspaper content analyses for bioethics research. The

limitations of this dissertation and ideas for future research are presented in chapter 10.4, and the dissertation closes with a conclusion in chapter 10.5.

1.2 Genetic testing

Genetic testing has a variety of medical applications that can be distinguished based on six dimensions (Table 1-1). The next paragraphs introduce these dimensions. The first dimension concerns the aim of genetic testing. While asymptomatic individuals might seek genetic testing to prevent or postpone disease outbreak through predictive genetic testing, genetic testing is also used in patients to diagnose genetic diseases, provide prognoses of disease progression or inform disease treatment.[1] The inheritance pattern is a second dimension to distinct genetic testing applications. While some diseases have a dominant inheritance pattern, meaning one disrupted allele is sufficient for disease development, recessive disorders need two disrupted alleles to develop, requiring both parents to be carriers (Vallance and Ford, Jason 2003). Third, genetic testing can be performed on somatic cells, for instance, tumour cells, or analyse germline mutations (Kamps et al. 2017).[2] Only germline genetic variants are inheritable. Fourth, genetic testing can be applied in embryos, fetuses, newborns, children and adults.

Table 1-1: Dimensions of genetic testing applications.

1	**Aim of testing**	treatment (affected)	-	prognosis (affected)	-	diagnosis (affected)	-	prevention (asymptomatic)	
2	**Inheritance pattern**	dominant			-	recessive			
3	**Inherited or not?**	somatic			-	germline			
4	**Moment of testing**	prenatal	-	newborn	-	child	-	adult	
5	**Level of specificity**	variant-specific	-	gene-specific	-	panel-specific	-	exome-wide	- genome-wide
6	**Level of penetrance**	complete	-	high	-	medium	-	low	

1 For example, spinal muscular atrophy (SMA) is a rare genetic disorder causing muscle degeneration and early death and is diagnosed by identifying loss-of-function variants in the *SMN1* gene (Arnold, Kassar, and Kissel 2015). Pharmacogenomic testing is used to optimise drug dose and response (Relling and Evans, W. 2015). And certain genetic tests on tumour tissue allow prognostic evaluations regarding disease progression and life expectancy (Ntavatzikos et al. 2017; Alix-Panabières and Pantel 2016).

2 For example, ovarian cancer patients are recommended to have a germline test for pathogenic BRCA variants. If this test does not reveal any pathogenic genetic variant, a somatic test for BRCA is recommended for treatment purposes (Vergote et al. 2016).

A fifth dimension concerns the specificity of genetic analyses. Variant-specific genetic testing is possible if a relative has already received the diagnosis of a disease-causing genetic variant. For monogenetic diseases, genetic testing in one specific gene is applicable.[3] Panel testing refers to a collection of genes tested simultaneously.[4] Exome-wide and genome-wide analyses are usually applied to undiagnosed conditions with an expected genetic cause (Henggeler and Matyas 2017). Exome-wide analyses include all gene-coding regions of the genome. Genome-wide analyses are even broader, including the whole genome (Meienberg et al. 2016).

Finally, pathogenic genetic variants might possess different levels of penetrance.[5] While Huntington's disease is fully penetrant[6] (McColgan and Tabrizi 2018), most other genetic variants detected in predictive genetic testing are not. For example, numerous genetic variants with variable penetrance have been associated with breast cancer risk. If more than 50% of carriers develop breast cancer, variants count as highly penetrant. Moderate penetrance means 20-50% lifetime risk. Below 20%, genetic variants have low penetrance (Ghoussaini, Pharoah, and Easton 2013). Carriers of moderate- or low-risk variants usually are not considered for preventive surgeries or treatment but are under more frequent cancer surveillance than non-carriers (Eliade et al. 2017). Thus, genetic testing decision-making is complex, and results are challenging to interpret. They require professional guidance.

1.2.1 Genetic counselling

Genetic counselling supports individuals in their decision-making process and result interpretation. As it requires additional training for healthcare professionals, specialised genetic counsellors provide this service in a growing number of countries, for instance, the United Kingdom, the United States or the Netherlands (Cordier et al. 2018). Table 1-2 compares the content of genetic counselling as recommended by the European genetic counselling guidelines for predictive genetic testing and required by the Swiss Human Genetic Testing Act (HGTA).[7] The European guidelines emphasise the importance of adequate information provision "based on

3 Monogenetic diseases are caused by one single gene that is disrupted or overexpressed. For example, genetic testing for Huntington's disease specifically tests for disease-causing CAG-repeats in the *HTT* gene.

4 Such panels are used, for instance, to test for Hereditary Breast and Ovarian Cancer syndrome (HBOC) (Desmond et al. 2015).

5 Penetrance is the percentage of people that develop a disease from a pathogenic genetic variant.

6 Huntington's disease is caused by a high number of CAG repeats in the *HTT* gene. 39 or more CAG repeats are fully penetrant, but 36-39 repeats are pathogenic with reduced penetrance (McColgan and Tabrizi 2018).

7 The HGTA specifies the content of genetic counselling for prenatal and predictive genetic testing.

the patient's needs and expectations" (Skirton et al. 2013, 258). Swiss law requires genetic counselling before and after predictive genetic testing (Swiss Federal Council 2004). While the European guidelines are more comprehensive, the legal requirements in Switzerland will become more detailed in the revised version of the HGTA (Swiss Federal Council 2017). While this indicates that genetic counselling is standard practice in the clinical setting, direct-to-consumer genetic testing does not have such a policy.

Table 1-2: Genetic counselling for predictive genetic testing in European guidelines and Swiss law.

Information	**Europ. Guidelines (Skirton et al. 2013)**	**HGTA**
Scope, purpose and process of test	x	x
Storage of sample and genetic data	x	x
Support options after the test	x[d]	x
Options after testing (surveillance/screening/treatment)	x	x
Disease-specific information	x	x
Meaning and implications of a test result	x	x
Consent	x	x[a]
Implications for family members	x[b]	x[a]
Psychological and social risks and issues	x[c]	x[a]
Patient's wishes and expectations	x	
Plan of disclosure to family members	x	
Waiting time	x	
Alternative options	x	
Way of result disclosure and plan after disclosure	x	
Possibility of secondary or incidental findings	(x)	x
Insurance coverage		x
Right not to knowe		x[a]
Legal conditions regarding disclosuref		x[a]

Notes: HGTA = Swiss Human Genetic Testing Act.

[a] To be implemented in the revised version of the HGTA (Swiss Federal Council 2017).

[b] Special emphasis on offspring.

[c] Specified: coping with test results, uncertainty, dealing with changed genetic status (psychological issues), impact on employment, insurance, personal and family relationships, privacy, confidentiality (social issues).

[d] Also during the testing process.

[e] Of patients and family members.

[f] Swiss law allows insurances to ask for genetic information in some circumstances (HGTA Art 27/2).

1.2.2 Direct-to-consumer genetic testing

Direct-to-consumer genetic testing makes genetic testing available to members of the public, as they can access genetic testing without a referral from a healthcare professional. Such tests can be purchased online from anywhere in the world by mailing a saliva sample and sometimes cost less than 100 USD. It usually does not include genetic counselling.

Even though the demand for direct-to-consumer genetic testing is limited, users and supporters see significant benefits in the availability of this service. They appreciate the autonomy people gain over their health and the social and personal benefits (Lee, S. and Crawley 2009; Covolo et al. 2015; Loi 2016; Turrini and Prainsack 2016) as they are interested in proactive medical care, particularly for existing conditions (Meisel et al. 2015). Some buy a test out of curiosity about their genetic predisposition (Wasson et al. 2013). Additionally, some users are interested in donating their genetic data to research by uploading them to data sharing platforms such as openSNP (Haeusermann et al. 2017; Vayena et al. 2014). However, as some studies conclude, most members of the public have limited interest in direct-to-consumer genetic testing (Charbonneau et al. 2019; Schaper, Wöhlke, and Schicktanz 2019). Moreover, increased genetic literacy is associated with lower interest in direct-to-consumer genetic testing (Stewart et al. 2018a; Gray, S. et al. 2009).

Additionally, direct-to-consumer genetic testing faces substantial criticism. First, its clinical validity and utility are limited: Risk predictions are often inaccurate because of incomplete scientific knowledge. Moreover, false-positive and false-negative test results occur frequently (Huml et al. 2019; Tandy-Connor et al. 2018). A second problematic aspect is the persuasive wording on direct-to-consumer genetic testing companies' websites. Being based on marketing communication strategies, they emphasise benefits over risks (Bunnik, Schermer, and Janssens 2014; Schaper and Schicktanz 2018; Singleton et al. 2012). Third, direct-to-consumer genetic testing companies based in the United States fall short on privacy policies because of the lack of legal requirements (Hazel and Slobogin 2018; Laestadius, Rich, and Auer 2017). Fourth, direct-to-consumer genetic testing might increase healthcare costs if users ask for their healthcare professionals' advice (Plothner et al. 2017). Even though an extensive survey study from the United States revealed that only a few users used clinical genetic counselling afterwards (Koeller et al. 2017), holistic analyses assessing the overall healthcare costs caused by direct-to-consumer genetic testing are missing. A fifth criticism of healthcare professionals comprises the concern that genetic risk predictions without genetic counselling could cause anxiety and misperceptions of risk (Moscarello et al. 2019; Lippi, Favaloro, and Plebani 2011). While meta-analyses of

empirical studies with direct-to-consumer genetic testing users so far did not confirm this worry (Covolo et al. 2015; Stewart et al. 2018b), methodological shortcomings and misconceptions on the concept of anxiety leave the necessity for further investigations (Oliveri et al. 2016). Thus, while some criticism is substantiated, other concerns have not been empirically confirmed.

Legislations regarding direct-to-consumer genetic testing differ between countries. While the United Kingdom has no legal restrictions, the U.S. Food and Drug Administration has legally restricted applications for medical risk predictions. Recently, the reporting of some genetic variants with high prevalence, actionability and scientific evidence have been approved (U.S. Food and Drug Administration 2017, 2018). Other countries, such as France, have banned most direct-to-consumer genetic testing applications (Kalokairinou et al. 2018). The drafted revision of the Swiss Human Genetic Testing Act also intends to ban most uses of direct-to-consumer genetic testing (Swiss Federal Council 2017). These differing legislations reflect different cultures and the differing perceptions of direct-to-consumer genetic testing, emphasising either the benefits of empowerment or the risks of harm. While the availability of direct-to-consumer genetic testing increases autonomy by providing a variety of choice to people (Vayena 2015), it has been questioned whether the decisions of users to buy such a test are truly informed (Bunnik, Janssens, and Schermer 2014; Jackson, Goldsmith, and Skirton 2014). Thus, direct-to-consumer genetic testing makes informing the public about genetic testing necessary, as they imply accessibility to genetic testing to everybody (Bunnik, Schermer, and Janssens 2014, see also chapter 1-4).

1.2.3 Predictive genetic testing

In summary, this dissertation assesses predictive genetic testing in asymptomatic individuals. Predictive genetic testing in the context here only concerns dominant conditions and always affects the germline (dimensions two and three, Table 1-1). Thus, it has implications for relatives. The scope of this dissertation excludes prenatal genetic testing and mainly focuses on adults, as guidelines and healthcare professionals discourage predictive genetic testing for late-onset diseases in minors (Swiss Academy of Medical Sciences 2011). Swiss law also restricts such applications in minors (HGTA Art 10/2). Reasons are the risk of psychological harm, adverse effects on families, discrimination and missing respect for the child's autonomy (Ross et al. 2013; Borry et al. 2008).[8]

8 Conversely, ethicists and social scientists call for allowing such applications in minors because of, for instance, few reported psychologically adverse effects, positive effects for the tested individuals and their families and avoidance of professional paternalism (Mand et al. 2012; Biesecker, B. 2016).

By contrast, the scope of this dissertation covers the whole spectra of dimensions five and six. While the main focus is on targeted genetic testing, which is variant- or gene-specific, broader genetic analyses are within the scope if they lead to predictive results. The less specific the analysis, the more probable are unintended findings with predictive components and unknown clinical relevance. It is challenging for healthcare professionals to decide whether to disclose such findings to the tested individual (Green, R. et al. 2013; Saelaert et al. 2018). Test results could also reveal variants of uncertain significance, for which the pathogenicity is unknown (Richards, S. et al. 2015). Moreover, predictive genetic testing includes all levels of penetrance, but most variants have incomplete penetrance, leading to the inherent uncertainty in predictive genetic testing and complicating result interpretation.

Moreover, it is essential to distinguish between predictive genetic testing in a clinical setting, which includes genetic counselling and direct-to-consumer genetic testing. Regulations and clinical utility differ importantly between these two settings. Only direct-to-consumer genetic testing applications that include disease-specific predictive outcomes are within the scope of this dissertation.[9] Having defined the scope of this dissertation regarding genetic testing, the following chapter introduces the theoretical background, focussing on informed decision-making and mass media communication.

1.3 Theoretical background

This chapter explains the conditions and requirements for informed decision-making in the context of predictive genetic testing. Individuals usually use both private and public information channels. Mass media communication, an example of public information channels, is introduced in chapter 1.3.2.

1.3.1 Autonomy and informed decision-making

This dissertation focuses on informed genetic testing decision-making. The decision-makers in this context are patients and at-risk individuals considering genetic testing for medical purposes. The next sections introduce what constitutes informed decision-making from a theoretical perspective, examining the conditions of autonomous decision-making and the requirements for informed decision-making.

9 This only applies to the newspaper content analyses (chapters 6-8). The interview study is limited to the clinical setting and excludes direct-to-consumer genetic testing (see chapter 2.1 for the methodology).

Conditions for autonomous decision-making

In light of abusive research practices and medical paternalism, autonomy is a central concept for contemporary bioethics (O'Neill 2002, 34–36; Beauchamp 2010). Faden and Beauchamp (1986) define three conditions for autonomous action in the context of informed consent:[10]

> *"X acts autonomously only if X acts*
>
> *1. intentionally,*
>
> *2. with understanding, and*
>
> *3. without controlling influences." (Faden and Beauchamp 1986, 238)*

First, an *intentional* decision is made willingly and according to a plan. Decisions are still intentional in the case of unwanted side-effects resulting from it, as long as they were anticipated (Beauchamp 2010, 84). While intentionality is either present or absent, the other two conditions are a matter of degree. Thus, substantial but not full understanding and a certain level of controlling influences are acceptable to call an action substantially autonomous (Faden and Beauchamp 1986, 239). As actions made with understanding are usually intentional, the first condition is met if the second is, too (Faden and Beauchamp 1986, 248).

Second, the condition of *understanding* is conceptualised as an understanding of the consequences of one's action (Faden and Beauchamp 1986, 250–54). Competence is necessary to gain such understanding because information must be processed and interpreted accurately and rationally. Competence requires a sufficiently healthy mental and cognitive state to understand and critically assess relevant information as well as the capability to express decisions based on a free-will (Welie, J. and Welie, S. 2001).

Third, regarding *controlling influences*, Beauchamp and Childress refer to coercion, manipulation or mental illness (Beauchamp and Childress 2013, 104–5). Relatedly, Huibers and van `t Spijker (1998) distinguish between strong voluntariness of choice without any controlling influences and weak voluntariness, which is fulfilled if at least two options are available. Accordingly, the decision-making person must perceive to have at least two realistic options to choose from (Huibers and van `t Spijker 1998; White 1998).[11] While these concepts acknowledge the presence of external controlling influences, relational autonomy sees autonomy and the social

10 In *Principles for Biomedical Ethics,* Beauchamp and Childress (2013, 104–15) adopt the same conditions.

11 In the case of genetic testing decision-making, these two options are either to take the test or not to take it. While this seems obvious, some at-risk individuals perceive not taking genetic testing as no real option (d'Agincourt-Canning 2006; Hallowell et al. 2006). These findings thus question the voluntariness of genetic testing decision-making for these individuals.

environment as an intertwined entity. Some relational autonomy theories involve causal accounts, meaning relationships *cause* autonomy. Others, however, use a constitutive conception where relationships *define* autonomy (Stoljar 2018).[12] Thus, while most theoretical approaches accept external controlling influences as long as they are not coercive or manipulative, their theoretical conception differs fundamentally.

In bioethical discussions, autonomous decision-making is often linked to informed consent, which is the right to accept or refuse medical treatment. Patients, however, might provide or withhold consent without fulfilling the conditions of autonomous decision-making. The emphasis on informed consent might even diminish autonomy if it is reduced to the legal procedure, for instance, signing the consent form (O'Neill 2002, 37–38). Accordingly, Beauchamp and Childress (2013: 122-123) distinguish between two meanings of informed consent. One concerns the informed decision-making process, the other the legal requirement for medical doctors to obtain consent. This dissertation focuses on the first meaning only.

While conditions for autonomous decision-making are required to make an informed decision, informed decision-making emphasises the level of information processed before the decision.

Informed decisions

The complexity of predictive genetic testing requires a realistic approach regarding the extent of information because excessive or unstructured information might cause confusion and information overload (Huibers and van `t Spijker 1998). Accordingly, Manson and O'Neill (2007) explicate the standards for successful information transmission. Information must be relevant for the particular person, "adequately accurate" and needs to be transmitted in an appropriate, understandable language. These standards include simplifying complex information while staying adequately correct and truthful (Manson and O'Neill 2007, 83–90).

> *"Good communication takes account of what others already know, and of what they want and need to know at that time, in that context. Epistemically adequate communication is relevant communication, and has to be limited to what is appropriate to the actual context. Good communicative practice therefore always involves withholding information – comprehensible, true, grounded information – that could have been conveyed." (Manson and O'Neill 2007, 63)*

However, the act of withholding certain information deemed inadequate bears the risk of paternalism. Remaining correct and truthful while assessing what nature and amount of information are adequate for each individual is a challenging task for healthcare professionals. Beauchamp

12 Chapter 5.2 introduces relational autonomy in more detail.

and Childress sensibly suggest to use as a standard of disclosure what a reasonable average person would want to know, and to then adjust information to the individual information needs of patients (Beauchamp and Childress 2013, 125–27). Many authors acknowledge that a certain degree of paternalism is justified if it benefits the patient and part of the medical practice (Shaw and Elger 2013; Beauchamp and Childress 2013, 216–19). Nevertheless, it remains a practical challenge to assess how much information is needed and necessary to make an informed decision.

In summary, autonomous decisions must be made intentionally, with a sufficient level of understanding and without extensive controlling influences. That requires adequate information, as excessive or unstructured information might cause information overload and misconceptions. Such information can be transmitted through private and public information channels, which are introduced in the following section.

1.3.2 Information channels

People can obtain information through private or public channels (Figure 1-1). Private information channels, such as personal communication or closed meetings, have a well-defined audience and usually include the option to engage in a conversation or ask questions. By contrast, public information, such as mass media communication or public lectures, reaches an undefined audience. In this dissertation, genetic counselling serves as an example of private information channels and mass media communication for public information channels.[13] The following chapter introduces the theoretical basis of mass media communication.

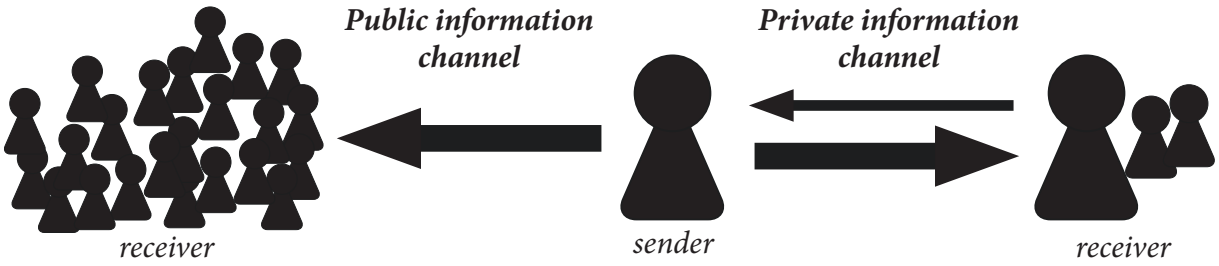

Figure 1-1: Public and private information channels.

13 A detailed introduction and literature reviews on genetic counselling and newspaper coverage about genetic testing are presented in the corresponding articles (see chapters 3 and 4 for genetic counselling; and chapters 6 and 7 for newspaper coverage).

1.3.3 Mass media communication

The functions of mass media in society

Functions of mass media are manifold and dependent on cultures and perspectives.[14] Mass media functions for Western societies include (1) socialisation of society members (including culture, social orientation and integration); (2) entertainment and recreation; (3) education and information provision, including explanations and interpretations of such information; (4) review and control function of other social systems, especially politics (McQuail 1987, 71–73; Ronneberger 2002). However, diverse stakeholders might attribute different roles to mass media (Ronneberger 2002).

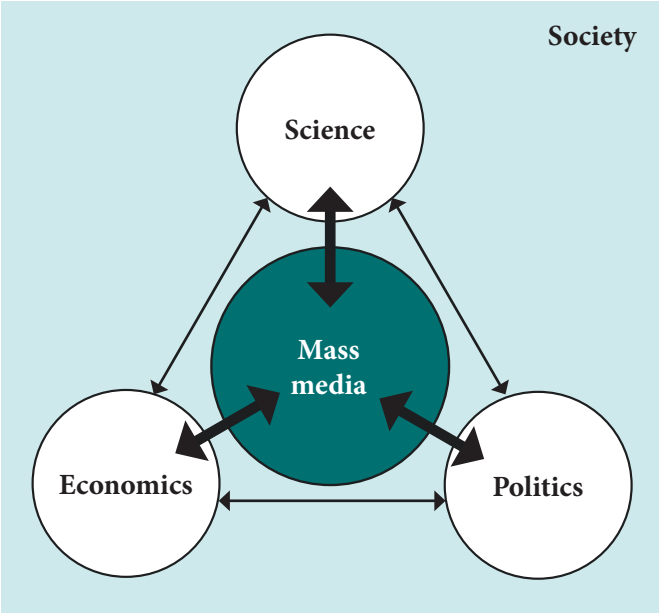

Figure 1-2: Conceptualising mass media and science as system of society that interact with each other and with other social systems.

Luhmann (1987) conceptualises mass media and science as systems of society. They interact with each other and with other societal systems, such as politics and economy (Figure 1-2). Each system has its own rules and functioning. According to Luhmann (1987: 555), society is communication. Therefore, the mass media have an additional role in society, as they provide society and its systems a platform for self-reflection (Luhmann 2009, 118–28) also called "arena function" (Kamber and Imhof 2011; Peters, H. 1994). For the context of this dissertation, the information, education and arena functions of mass media are most relevant. Apart from this, Luhmann's system approach illustrates the relationship and connections between mass media, science and the public, which are explained in the following two sections.[15]

14 Therefore, comparing media systems and media coverage from different countries can be meaningful. For instance, Western media systems are comparable regarding the connection between the mass media and politics, structures and developments of national media markets, the level and culture of journalistic professionalism or level of state control over mass media (Blum 2014; Hallin and Mancini 2011; see also chapter 7).

15 See also chapter 7.2.2.

Relationship between science and mass media

Science and mass media as two social systems share many differences, but also similarities. Both gain factual knowledge through systematic research, aiming for objectivity and credibility. Both systems use print and online media to disseminate their message. By contrast, while science is interested in finding regularities and focuses on longterm problems, mass media portray predominantly extraordinary and current issues. Only scientific publications gain credibility and relevance through peer-review processes and citation indices (Kolb and Burkhardt 2008; Weingart 2012).

The theory of Medialisation assumes that the mass media have become increasingly vital for legitimising and establishing the political influence of science (Peters, H. et al. 2008; Weingart, Rödder, and Franzen 2012). Accordingly, scientific institutions make substantial efforts to foster the public understanding of and engagement with science (Weingart 2012). Their aims are manifold and include (besides the legitimation of funding, controversial policy discussions and the increase in public willingness to fund scientific research), the public participation to science to improve public and private decision-making (Franzen, Weingart, and Rödder 2012).

Relationship between science and the public

There are opposing theoretical concepts regarding the relationship between science and the public. The "deficit model" (Condit 2010) claims that scientific experts have the correct understanding of science and thus need to educate the unknowing and entirely passive, non-specialist public about scientific facts. By contrast, the constructionist model conceptualises the public as experts, as they represent the "real" interests of society (Bonney et al. 2016; Prior 2003). Condit (2010) suggests a middle-ground, where both actors are equally relevant, and expert and public views complement each other.

Two science communication approaches build on these concepts. The Public Understanding of Science (PUS) approach is based on the deficit model. It is a top-down approach conceptualising scientists educating the public about science. By contrast, the more recently emerging approach of Public Engagement with Science and Technology (PEST) conceptualises scientists, the public and other societal stakeholders as equals engaging in public discourse about science (e.g., Jones 2011; Stilgoe, Locke, and Wilsdon 2014).[16] While the previous sections have focussed on the relationship between mass media, science and society, it follows an introduction to established theories regarding the generation of media coverage and the effects on recipients and society.

16 A more detailed introduction to these approaches is presented in chapter 7.2.

Theories for the generation of media content

There are various theories about the factors influencing mass media content and effects of media coverage. Here, the most relevant theories for this dissertation are outlined (see Figure 1-3 for a schematic overview).

Figure 1-3: Overview over factors influencing media content and the effects reporting has on recipients, society and other stakeholders.

First, the Gatekeeping Theory addresses the process of news selection, focussing on three interconnected levels. The individual level investigates the journalist as a gatekeeper, the institutional level focuses on editors and publishers, and the cybernetic level includes the socio-cultural context in general. As such, the Gatekeeping theory has gradually expanded its attention from the microlevel (the individual journalist) to the macrolevel (sociocultural context) (Shoemaker 2002).

Second, the Theory of New Values aims to identify selection criteria of topics covered (or not covered) by media. While the empirically determined news values are manifold, Schulz (1990) established a core list of six news factors, being time, closeness, status, dynamics, valence and identification. More recently, Badenschier and Wormer (2012) suggested different news values for science journalism. Scientific news values seem to depend on timing and circumstances since political news will always trump science news. Scientific communication also has more value for mass media if they connect to other news, if well-known people talk about it and whether the story has a high scientific impact.[17] Factors to consider are importance (actuality, political relevance, economic relevance, relevance to recipients/society, scientific relevance, intention, personalisation, range), surprise (astonishment, controversy, unexpectedness) and appearance (graphical material) (Badenschier and Wormer 2012).

Third, the Issue Attention Cycle concept suggests looking at the quantity of media coverage over time and investigating peaks in coverage more narrowly, taking into account the theories

17 The scientific impact is considered higher if the story is associated with a large scientific conference or published in a high-impact journal.

described above (Kolb and Burkhardt 2008; Kolb 2005). An issue attention cycle consists of five phases. In the first phase, the issue remains a scientific problem. Mass media rarely report on it. In the second phase, one or several key events arouse the mass media's interest. The increasing media coverage focuses predominantly on the societal and political relevance of the issue at stake. Mass media interest peaks once a societal solution is in place. Afterwards, media coverage declines quickly.

Media effects

In addition to these theoretical considerations regarding the generation of mass media coverage, manifold media effect models and theories alternately attributed strong or weak effects of mass media coverage on recipients over the past 90 years (Neuman and Guggenheim 2011). The following sections introduce the agenda-setting and the framing model.

The agenda-setting model claims that media influence priorities of issues by choosing some over others. Therefore, mass media emphasise certain aspects through the topic selection and thus make recipients more aware of them (Maurer 2010). Attempts to investigate this claim include priorities of some topics over others versus the salience of one particular issue. These attempts are analysable on a societal or an individual level (Maurer 2010, 40–55).

The framing model claims that mass media influence the public by selecting certain aspects and perspectives and making them more salient than others (Matthes 2014; Chong and Druckman 2007). Entman (1993) formulated the widely used definition of frames:

> *"Frames, then, define problems - determine what a causal agent is doing with what costs and benefits, usually measured in terms of common cultural values; diagnose causes - identify the forces creating the problem; make moral judgments - evaluate causal agents and their effects; and suggest remedies - offer and justify treatments for the problems and predict their likely effects." (Entman 1993, 52)*

In summary, these two models suggest that the mass media influence the public discourse by choosing some topics over others and by selecting certain aspects of those topics and making them more salient. However, the public's opinion is not directly deducible from such effects. On the individual level, effects might be different than on the societal level (Maurer 2010). Despite these limitations, they demonstrate a substantial impact of mass media on the public discourse and, thus, on society (Condit 2001; Caulfield 2005). Following these theoretical considerations regarding autonomous decision-making and information dissemination through mass media communication, the next chapter introduces contemporary challenges for society regarding predictive genetic testing.

1.4 Societal challenges of predictive genetic testing

Genetic testing, and especially predictive genetic testing, is associated with societal challenges that make investigations on informed decision-making relevant. As indicated in Figure 1-4, increasing public awareness and breakthroughs in genetic research are increasing the availability and accessibility of genetic testing services. Raising awareness among the general population (Lebo et al. 2015), the American actress Angelina Jolie caused extensive media coverage on predictive genetic testing and preventive surgeries (Kamenova, Reshef, and Caulfield 2014; chapter 7). In a New York Times article, she revealed that she had a preventive double mastectomy because she carried a pathogenic variant in the *BRCA2* gene that increased her risk of developing breast and ovarian cancer (Jolie, May 13, 2013).

Scientific breakthroughs concern DNA sequencing and genome editing. Next-generation sequencing technologies made DNA sequencing faster and cheaper, leading to the generation of high amounts of genetic and genetic-associated data (Bahassi and Stambrook 2014). International consortia, such as HapMap (International HapMap Consortium 2003) or the 1000 Genomes Project (Auton et al. 2015), contribute to the identification of new pathogenic genetic variants and more clinical genetic testing applications (de Vries, P. et al. 2017). Next-generation sequencing also made whole-exome and whole-genome analyses affordable. Moreover, Crispr-Cas9 genome editing (Jinek et al. 2013) made it faster and cheaper for research laboratories to study specific genetic variants *in vitro* and *in vivo*, contributing to a better understanding of links between genetic variants and disease.

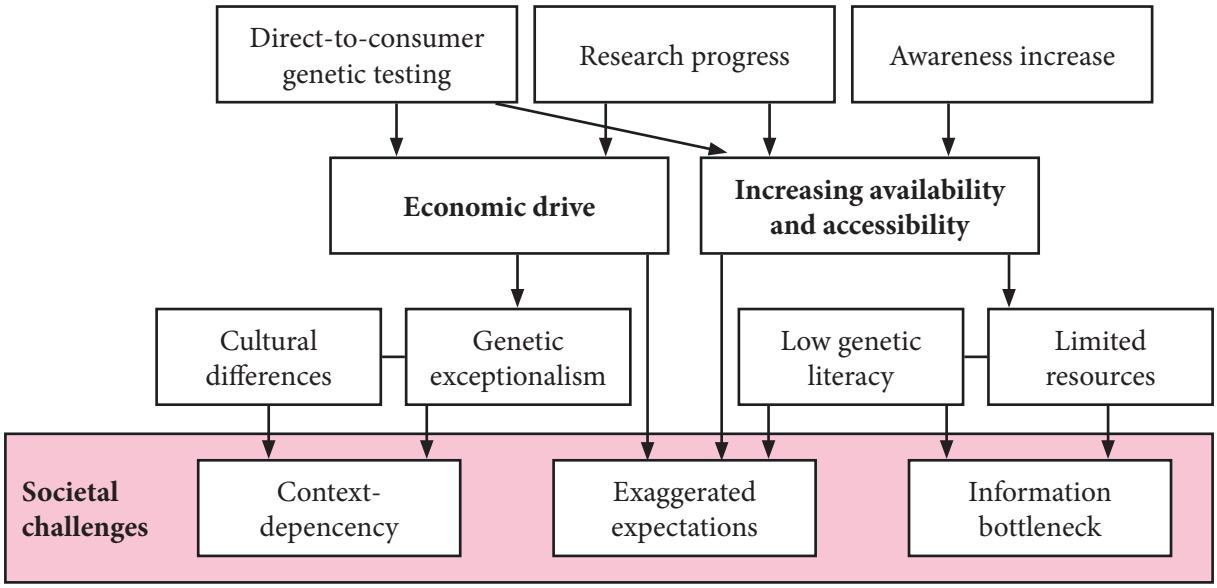

Figure 1-4: Schematic overview over the factors connected to current societal challenges in predictive genetic testing.

Such scientific progress, as well as direct-to-consumer genetic testing (see chapter 1.2.2), promote the economisation of genetic testing (Conrad 2005). Consequently, the biotech and pharmaceutical industries develop high financial interests in genetic testing. These developments lead to societal challenges introduced in the following sections.

1.4.1 Information bottleneck

The first societal challenge regarding predictive genetic testing is the risk of an information bottleneck. The more widespread genetic testing becomes, the more people will need adequate information to make informed decisions. Low genetic literacy among the general population and limited personal resources for genetic counselling are risk factors that might lead to such an information bottleneck.

Low genetic literacy

Abrams et al. (2015) suggest operationalising genetic literacy on three levels: awareness, practical knowledge and factual knowledge. Factual knowledge about genetics is low, even in well-educated populations (Chapman et al. 2019; Etchegary et al. 2015; Goltz, Bergman, and Goodson 2016; Hoffman et al. 2010). Younger age and higher education positively influenced genetic literacy (Haga et al. 2013). Genetic literacy tends to be higher in the United States than in Europe (Haga et al. 2013; de Vries, H. et al. 2005), but there is only limited data from European countries. Low health literacy is associated with a feeling of information overload (Khaleel et al. 2020).

Higher genetic literacy is associated with more enthusiastic and more sceptical attitudes towards genetics. Low genetic literacy leads to difficulties in forming an attitude at all (Jallinoja and Aro 2000). Cultural frames and social experiences shape beliefs and conceptions of genetics more than factual knowledge (Goltz, Bergman, and Goodson 2016; Condit 2010). Misconceptions impair genetic literacy (Rose, A. et al. 2005) and people notably have problems in understanding genetic risk (Condit and Parrott 2004; Condit and Shen 2010). There is consensus that genetic literacy among the general population should be improved (Marzuillo et al. 2013; Corbellini 2004; Martin, D., Greenwood, and Nisker 2010).

Limited personal resources for genetic counselling

Additionally, the demand for professional genetic counselling rises with the increasing availability of genetic testing applications. At the same time, personal and financial resources for genetic counselling are limited, particularly in Switzerland. Compared to other countries, Switzerland has few adequately trained personnel (Abacan et al. 2019), as geneticists and other medical

doctors with specialised training are responsible for genetic counselling. The genetic counsellor profession is, to date, not officially accredited in Switzerland. Instating genetic counsellors trained in France is becoming more common in the French-speaking part of Switzerland. A Master program in Italy, which started in 2017 (Abacan et al. 2019), makes Italian-speaking personnel available for the Italian-speaking part of Switzerland. However, since no training program is available for genetic counselling in the German language, few additional personnel will be available in the German-speaking region of Switzerland. In the medium-term, this situation might lead to an information bottleneck, considering the increasing availability of genetic testing.

1.4.2 Exaggerated expectations

A second societal challenge concerns exaggerated expectations regarding predictive genetic testing. The promises regarding genetic testing for personalised healthcare and disease prevention are creating high expectations among experts and the general population and are transmitted through mass media (Kamenova et al. 2016; Marcon, Bieber, and Caulfield 2018). However, many conditions known to have a heritable component, such as diabetes, obesity or hypertonia, are not linkable to distinct genetic variants (Lyssenko and Laakso 2013; Caskey et al. 2014). Instead, environmental factors often are more influential for disease outbreak and disease progress than genetic predisposition. Thus, the predictive value of genetic testing remains low for many common diseases. The expectations regarding predictive genetic testing might thus be exaggerated.

Mass media reporting, however, is not the only reasons for exaggerated expectations towards genetics among the general public (Caulfield 2004). The commercialisation of genetics, as well as the scientists themselves, also drive this "hype", as industry, science and mass media profit from exaggerating news about genetics (Caulfield 2005; Caulfield and Condit 2012). Such exaggerations are problematic as it might weaken public trust in genetics (Bubela 2006). As members of the public cannot distinguish between different genetic applications, a lack of trust in genetic research might also affect acceptance of clinically meaningful and well-established genetic analyses.

1.4.3 Context-dependency

Context-dependency is the third challenge related to predictive genetic testing. Policy-making and regulations need to be adapted according to the cultural setting and the specific testing applications. Thus, policy-relevant research must be context-specific, too.

Genetic exceptionalism and contextualism

Advocates of genetic exceptionalism request special treatment of genetic tests and genetic information because they differ from other medical tests and information (Sulmasy 2015; McGuire et al. 2008). That leads, for instance, to special regulations and clinical processes, like genetic-specific legislations and the institutionalisation of genetic counselling.

As such, the view of genetic exceptionalism has been influential, despite numerous contemporary objections (Rothstein 2005). Some authors consider genetic information as no different from other medical information (Evans, J. and Burke 2008; Green, M. and Botkin 2003; Zimmern and Khoury 2012). Complexities attributed to genetic testing, such as family involvement, predictive information, distress or risk of re-identification are also applicable to other medical information. Different applications of genetic testing have different implications, some being more exceptional than others. Moreover, it might be impossible to separate genetic from non-genetic information in medical records (Manson and O'Neill 2007, 136-138). Only a minority of people consider genetic information as different from other medical information (Lenk et al. 2019; Ruiz-Canela, Valle-Mansilla, and Sulmasy 2011).

In a critique of this binary discussion for or against genetic exceptionalism, Garrison, N.' et al. (2019) offer a middle-ground solution. They consider "genetic contextualism" more fruitful than genetic exceptionalism because this framework acknowledges that genetic information unifies several features that separately are not unique to genetic information. The combination of features, however, makes genetics a relevant topic for public discussions, which, in turn, also need to be contextualised (Garrison, N.' et al. 2019). Contextualism, however, is more challenging to implement in policies and regulations, as they require context-specific knowledge.

Cultural differences

The second layer of context lies within countries and cultures. Because they differ regarding people's attitudes towards genetic risk information, regulations and policies should adapt to these different setups. This dissertation focuses on Western culture with particular emphasis on Switzerland. Studies from the United States, Canada, Australia and the United Kingdom dominate research on attitudes, knowledge and decision-making.[18] Recently, research studies assessed the culture-specific situation in the Netherlands (Vermeulen et al. 2014), Spain (Rivera-Navarro, Cubo, and Mariscal 2015) and Italy (Godino et al. 2018). However, no empirical studies about attitudes, knowledge and decision-making of people considering genetic testing in the medical context have focused on Switzerland.

18 See chapter 4.2 for a literature review.

1.4.4 Summary: Relevance of the research project

In sum, technological advancements, direct-to-consumer genetic testing and increasing public awareness lead to increased availability and accessibility of genetic testing for both patients and the general population, further driven by economic interests. These circumstances lead to new and ongoing societal challenges. First, low genetic literacy among the general population and limited resources for personal genetic counselling might eventually lead to an information bottleneck. Thus, not all people accessing and using genetic testing might receive the support necessary for informed decision-making. Second, the public debate tends to exaggerate the possibilities and potential of genetic testing. Third, it is context- and culture-dependent to what extent societies want to introduce predictive genetic testing. While some genetic testing applications might become standard clinical practice, others might indeed need special regulation. Additionally, technological means might not everywhere surpass ethical or economic evaluations due to cultural differences. The legislation of the German-speaking region differs from the well-studied areas (Kalokairinou et al. 2018), but public debates and decision-making regarding genetic testing have, so far, not been studied in this setting.

1.5 Objectives

The objectives of this dissertation ground on the aforementioned societal challenges related to genetic testing (chapter 1.4). The information bottleneck and exaggerated expectations make informed decision-making for those considering genetic testing challenging. Thus, the primary aim of this doctoral dissertation is to examine what information about genetic testing should be given to at-risk individuals and the public to foster informed decision-making. Another objective is to investigate how such information is best transmitted. To address context-dependency, this dissertation has a specific focus on predictive genetic testing for actionable and non-actionable variants in the cultural context of the German-speaking part of Switzerland (using the United Kingdom as a comparison). More specifically, the following research questions are investigated:

1. How do people at risk for a genetic cancer predisposition inform themselves regarding genetic testing? What do they need to make an informed decision?
2. What are appropriate solutions if genetic counselling resources become scarce?
3. What do mass media report about genetic testing, especially on predictive genetic testing?
4. What role should mass media play in informing the public about genetic testing?

5. What and how much information to the public (through mass media or other channels) is appropriate when it comes to genetic testing?

Based on these assessments, this dissertation further aims to evaluate how newspaper content analyses can inform bioethics research and to explore the usefulness and limits of connecting these two fields.

1.6 References

Abacan, MaryAnn, Lamia Alsubaie, Kristine Barlow-Stewart, Beppy Caanen, Christophe Cordier, Eliza Courtney, Emeline Davoine et al. 2019. "The Global State of the Genetic Counseling Profession." Eur J Hum Genet 27 (2): 183–97. doi:10.1038/s41431-018-0252-x.

Abrams, Leah R., Colleen M. McBride, Gillian W. Hooker, Joseph N. Cappella, and Laura M. Koehly. 2015. "The Many Facets of Genetic Literacy: Assessing the Scalability of Multiple Measures for Broad Use in Survey Research." PLoS One 10 (10): e0141532. doi:10.1371/journal.pone.0141532.

Alix-Panabières, Catherine, and Klaus Pantel. 2016. "Clinical Applications of Circulating Tumor Cells and Circulating Tumor DNA as Liquid Biopsy." Cancer Discovery 6 (5): 479–91. doi:10.1158/2159-8290.CD-15-1483.

Arnold, W. D., Darine Kassar, and John T. Kissel. 2015. "Spinal Muscular Atrophy: Diagnosis and Management in a New Therapeutic Era." Muscle Nerve 51 (2): 157–67. doi:10.1002/mus.24497.

Auton, Adam, Lisa D. Brooks, Richard M. Durbin, Erik P. Garrison, Hyun M. Kang, Jan O. Korbel, Jonathan L. Marchini, Shane McCarthy, Gil A. McVean, and Gonçalo R. Abecasis. 2015. "A Global Reference for Human Genetic Variation." Nature 526 (7571): 68–74. doi:10.1038/nature15393.

Badenschier, Franziska, and Holger Wormer. 2012. "Issue Selection in Science Journalism: Towards a Special Theory of News Values for Science News?" In Rödder, Franzen, and Weingart 2012, 59–86.

Bahassi, El M., and Peter J. Stambrook. 2014. "Next-Generation Sequencing Technologies: Breaking the Sound Barrier of Human Genetics." Mutagenesis 29 (5): 303–10. doi:10.1093/mutage/geu031.

Beauchamp, Tom L. 2010. Standing on Principles: Collected Essays. New York: Oxford University Press.

Beauchamp, Tom L., and James F. Childress. 2013. Principles of Biomedical Ethics. 7th ed. New York: Oxford University Press.

Biesecker, Barbara B. 2016. "Predictive Genetic Testing of Minors: Evidence and Experience with Families." Genet Med 18 (8): 763–64. doi:10.1038/gim.2015.191.

Blum, Roger. 2014. Lautsprecher Und Widersprecher: Ein Ansatz Zum Vergleich Der Mediensysteme. Köln: Herbert von Halem.

Bonney, Rick, Tina B. Phillips, Heidi L. Ballard, and Jody W. Enck. 2016. "Can Citizen Science Enhance Public Understanding of Science?" Public Underst Sci 25 (1): 2–16. doi:10.1177/0963662515607406.

Borry, Pascal, Tom Goffin, Herman Nys, and Kris Dierickx. 2008. "Attitudes Regarding Predictive Genetic Testing in Minors: A Survey of European Clinical Geneticists." Am J Med Genet C 148C (1): 78–83. doi:10.1002/ajmg.c.30165.

Bubela, Tania M. 2006. "Science Communication in Transition: Genomics Hype, Public Engagement, Education and Commercialization Pressures." Clin Genet 70 (5): 445–50. doi:10.1111/j.1399-0004.2006.00693.x.

Bunnik, Eline M., A. C. J. W. Janssens, and Maartje H. N. Schermer. 2014. "Informed Consent in Direct-to-Consumer Personal Genome Testing: The Outline of a Model Between Specific and Generic Consent." Bioethics 28 (7): 343–51. doi:10.1111/bioe.12004.

Bunnik, Eline M., Maartje H. N. Schermer, and A. C. J. W. Janssens. 2014. "Naming and Framing in Genomic Testing." Trends Mol Med 20 (2): 63–65. doi:10.1016/j.molmed.2013.11.001.

Caskey, C. T., Manuel L. Gonzalez-Garay, Stacey Pereira, and Amy McGuire. 2014. "Adult Genetic Risk Screening." Annu Rev Med 65:1–17. doi:10.1146/annurev-med-111212-144716.

Caulfield, Timothy A. 2004. "Biotechnology and the Popular Press: Hype and the Selling of Science." Trends Biotechnol 22 (7): 337–39. doi:10.1016/j.tibtech.2004.03.014.

———. 2005. "Popular Media, Biotechnology, and the Cycle of Hype the Mass Media's Influence on Health Law and Policy Symposium." Houston J Health Law Policy 5: 213-233.

Caulfield, Timothy A., and Celeste M. Condit. 2012. "Science and the Sources of Hype." Public Health Genom 15 (3-4): 209–17. doi:10.1159/000336533.

Chapman, Robert, Maxim Likhanov, Fatos Selita, Ilya Zakharov, Emily Smith-Woolley, and Yulia Kovas. 2019. "New Literacy Challenge for the Twenty-First Century: Genetic Knowledge Is Poor Even Among Well Educated." J Community Genet 10 (1): 73–84. doi:10.1007/s12687-018-0363-7.

Charbonneau, Jan, Dianne Nicol, Don Chalmers, Kazuto Kato, Natsuko Yamamoto, Jarrod Walshe, and Christine Critchley. 2019. "Public Reactions to Direct-to-Consumer Genetic Health Tests: A Comparison Across the US, UK, Japan and Australia." Eur J Hum Genet. doi:10.1038/s41431-019-0529-8.

Chong, Dennis, and James N. Druckman. 2007. "Framing Theory." Annu Rev Polit Sci 10 (1): 103–26. doi:10.1146/annurev.polisci.10.072805.103054.

Condit, Celeste M. 2001. "What Is 'Public Opinion' About Genetics?" Nat Rev Genet 2 (10): 811–15. doi:10.1038/35093580.

———. 2010. "Public Understandings of Genetics and Health." Clin Genet 77 (1): 1–9. doi:10.1111/j.1399-0004.2009.01316.x.

Condit, Celeste M., and Roxanne Parrott. 2004. "Perceived Levels of Health Risk Associated with Linguistic Descriptors and Type of Disease." Sci Commun 26 (2): 152–61. doi:10.1177/1075547004270078.

Condit, Celeste M., and Lijiang Shen. 2010. "Public Understanding of Risks from Gene-Environment Interaction in Common Diseases: Implications for Public Communications." Public Health Genom 14 (2): 115–24. doi:10.1159/000314915.

Conrad, Peter. 2005. "The Shifting Engines of Medicalization." J Health Soc Behav 46 (1): 3–14. doi:10.1177/002214650504600102.

Corbellini, Gilberto. 2004. "Genetic Risk, Medical Education, Public Understanding of Genetics and Evolutionary Medicine: The Challenges of Genetic Counselling for Complex Disorders." Topoi 23 (2): 187–93. doi:10.1007/s11245-004-5379-4.

Cordier, Christophe, Marion McAllister, Clara Serra-Juhe, Joana Bengoa, Sara Pasalodos, Inga Bjornevoll, Irène Feroce, Ramona Moldovan, Milena Paneque, and Deborah Lambert. 2018. "The Recognition of the Profession of Genetic Counsellors in Europe." Eur J Hum Genet 26 (12): 1719–20. doi:10.1038/s41431-018-0260-x.

Covolo, Loredana, Sara Rubinelli, Elisabetta Ceretti, and Umberto Gelatti. 2015. "Internet-Based Direct-to-Consumer Genetic Testing: A Systematic Review." J Med Internet Res 17 (12): e279. doi:10.2196/jmir.4378.

d'Agincourt-Canning, Lori. 2006. "Genetic Testing for Hereditary Breast and Ovarian Cancer: Responsibility and Choice." Qual Health Res 16 (1): 97–118. doi:10.1177/1049732305284002.

de Vries, Hein, Ilse Mesters, Hermanna van de Steeg, and Cora Honing. 2005. "The General Public's Information Needs and Perceptions Regarding Hereditary Cancer: An Application of the Integrated Change Model." Patient Educ Couns 56 (2): 154–65. doi:10.1016/j.pec.2004.01.002.

de Vries, Paul S., Maria Sabater-Lleal, Daniel I. Chasman, Stella Trompet, Tarunveer S. Ahluwalia, Alexander Teumer, Marcus E. Kleber et al. 2017. "Comparison of HapMap and 1000 Genomes Reference Panels in a Large-Scale Genome-Wide Association Study." PLoS One 12 (1): e0167742. doi:10.1371/journal.pone.0167742.

Desmond, Andrea, Allison W. Kurian, Michele Gabree, Meredith A. Mills, Michael J. Anderson, Yuya Kobayashi, Nora Horick et al. 2015. "Clinical Actionability of Multigene Panel Testing for Hereditary Breast and Ovarian Cancer Risk Assessment." JAMA Oncol 1 (7): 943–51. doi:10.1001/jamaoncol.2015.2690.

Eliade, Marie, Jeremy Skrzypski, Amandine Baurand, Caroline Jacquot, Geoffrey Bertolone, Catherine Loustalot, Charles Coutant et al. 2017. "The Transfer of Multigene Panel Testing for Hereditary Breast and Ovarian Cancer to Healthcare: What Are the Implications for the Management of Patients and Families?" Oncotarget 8 (2): 1957–71. doi:10.18632/oncotarget.12699.

Entman, Robert M. 1993. "Framing: Toward Clarification of a Fractured Paradigm." J Commun 43 (4): 51–58.

Etchegary, Holly, Jane Green, Patrick Parfrey, Catherine Street, and Daryl Pullman. 2015. "Community Engagement with Genetics: Public Perceptions and Expectations About Genetics Research." Health Expect 18 (5): 1413–25. doi:10.1111/hex.12122.

Evans, James P., and Wylie Burke. 2008. "Genetic Exceptionalism. Too Much of a Good Thing?" Genet Med 10 (7): 500–501. doi:10.1097/GIM.0b013e31817f280a.

Faden, Ruth R., and Tom L. Beauchamp. 1986. A History and Theory of Informed Consent. With the assistance of N. M. P. King. New York: Oxford University Press.

Franzen, Martina, Peter Weingart, and Simone Rödder. 2012. "Exploring the Impact of Science Communication on Scientific Knowledge Production: An Introduction." In Rödder, Franzen, and Weingart 2012, 3–16.

Garrison, Nanibaa' A., Kyle B. Brothers, Aaron J. Goldenberg, and John Lynch. 2019. "Genomic Contextualism: Shifting the Rhetoric of Genetic Exceptionalism." Am J Bioeth 19 (1): 51–63. doi:10.1080/15265161.2018.1544304.

Ghoussaini, Maya, Paul D. P. Pharoah, and Douglas F. Easton. 2013. "Inherited Genetic Susceptibility to Breast Cancer: The Beginning of the End or the End of the Beginning?" Am J Pathol 183 (4): 1038–51. doi:10.1016/j.ajpath.2013.07.003.

Godino, Lea, Leigh Jackson, Daniela Turchetti, Catherine Hennessy, and Heather Skirton. 2018. "Decision Making and Experiences of Young Adults Undergoing Presymptomatic Genetic Testing for Familial Cancer: A Longitudinal Grounded Theory Study." Eur J Hum Genet 26:44–53. doi:10.1038/s41431-017-0030-1.

Goltz, Heather H., Margo Bergman, and Patricia Goodson. 2016. "Explanatory Models of Genetics and Genetic Risk Among a Selected Group of Students." Front Public Health 4:111. doi:10.3389/fpubh.2016.00111.

Gray, Stacy W., Cristin O'Grady, Lauren Karp, Daniel Smith, J. S. Schwartz, Robert C. Hornik, and Katrina Armstrong. 2009. "Risk Information Exposure and Direct-to-Consumer Genetic Testing for BRCA Mutations Among Women with a Personal or Family History of Breast or Ovarian Cancer." Cancer Epidemiol Biomarkers Prev 18 (4): 1303–11. doi:10.1158/1055-9965.EPI-08-0825.

Green, Michael J., and Jeffrey R. Botkin. 2003. "Genetic Exceptionalism in Medicine: Clarifying the Differences Between Genetic and Nongenetic Tests." Public Health Eth-UK 138 (7): 571–75.

Green, Robert C., Jonathan S. Berg, Wayne W. Grody, Sarah S. Kalia, Bruce R. Korf, Christa L. Martin, Amy McGuire et al. 2013. "ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing." Genet Med 15 (7): 565–74. doi:10.1038/gim.2013.73.

Haeusermann, Tobias, Bastian Greshake, Alessandro Blasimme, Darja Irdam, Martin Richards, and Effy Vayena. 2017. "Open Sharing of Genomic Data: Who Does It and Why?" PLoS One 12 (5): e0177158. doi:10.1371/journal.pone.0177158.

Haga, Susanne B., William T. Barry, Rachel Mills, Geoffrey S. Ginsburg, Laura Svetkey, Jennifer Sullivan, and Huntington F. Willard. 2013. "Public Knowledge of and Attitudes Toward Genetics and Genetic Testing." Genet Test Mol Biomarkers 17 (4): 327–35. doi:10.1089/gtmb.2012.0350.

Hallin, Daniel C., and Paolo Mancini. 2011. Comparing Media Systems Beyond the Western World: Cambridge University Press.

Hallowell, Nina, Audrey Ardern-Jones, Ros Eeles, Claire Foster, Anneke Lucassen, Clare Moynihan, and Maggie Watson. 2006. "Guilt, Blame and Responsibility: Men's Understanding of Their Role in the Transmission of BRCA1/2 Mutations Within Their Family." Sociol Health Illn 28 (7): 969–88. doi:10.1111/j.1467-9566.2006.00515.x.

Hazel, James W., and Christopher Slobogin. 2018. "Who Knows What, and When? A Survey of the Privacy Policies Proffered by U.S. Direct-to-Consumer Genetic Testing Companies." Cornell J Law Public Policy 28 (1): 35–66.

Henggeler, Caroline, and Gabor Matyas. 2017. "Gentests in Der Medizin – Ein Update." Hausarzt Praxis 12 (9): 14–18.

Hoffman, Richard M., Carmen L. Lewis, Michael P. Pignone, Mick P. Couper, Michael J. Barry, Joann G. Elmore, Carrie A. Levin, John van Hoewyk, and Brian J. Zikmund-Fisher. 2010. "Decision-Making Processes for Breast, Colorectal, and Prostate Cancer Screening: The DECISIONS Survey." Med Decis Making 30 (5_suppl): 53–64. doi:10.1177/0272989X10378701.

Huibers, Alex K., and Adriaan van 't Spijker. 1998. "The Autonomy Paradox: Predictive Genetic Testing and Autonomy: Three Essential Problems." Patient Educ Couns 35 (1): 53–62. doi:10.1016/S0738-3991(98)00083-4.

Huml, Anne M., Catherine Sullivan, Maria Figueroa, Karen Scott, and Ashwini R. Sehgal. 2019. "Consistency of Direct-to-Consumer Genetic Testing Results Among Identical Twins." Am J Med 133 (1): 143-146.e2. doi:10.1016/j.amjmed.2019.04.052.

International HapMap Consortium. 2003. "The International HapMap Project." Nature 426 (6968): 789–96. doi:10.1038/nature02168.

Jackson, Leigh, Lesley Goldsmith, and Heather Skirton. 2014. "Guidance for Patients Considering Direct-to-Consumer Genetic Testing and Health Professionals Involved in Their Care: Development of a Practical Decision Tool." Fam Pract 31 (3): 341–48. doi:10.1093/fampra/cmt087.

Jallinoja, P., and Arja R. Aro. 2000. "Does Knowledge Make a Difference? The Association Between Knowledge About Genes and Attitudes Toward Gene Tests." J Health Commun 5 (1): 29–39. doi:10.1080/10810730050019546.

Jinek, Martin, Alexandra East, Aaron Cheng, Steven Lin, Enbo Ma, and Jennifer A. Doudna. 2013. "RNA-Programmed Genome Editing in Human Cells." eLife 2:e00471. doi:10.7554/eLife.00471.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jones, Richard A. L. 2011. "Introduction: Public Engagement in an Evolving Science Policy Landscape." In Successful Science Communication: Telling It Like It Is, edited by David J. Bennet and Richard C. Jennings, 1–14. Cambridge: Cambridge University Press.

Kalokairinou, Louiza, Heidi C. Howard, Santa Slokenberga, Eva Fisher, Magdalena Flatscher-Thöni, Mette Hartlev, Rachel E. van Hellemondt et al. 2018. "Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape." J Community Genet 9 (2): 117–32. doi:10.1007/s12687-017-0344-2.

Kamber, Esther, and Kurt Imhof. 2011. "Qualitätsmedien Sind Wissensvermittler." In Krise Der Leuchttürme Öffentlicher Kommunikation: Vergangenheit Und Zukunft Der Qualitätsmedien, edited by Roger Blum, Heinz Bonfadelli, Kurt Imhof, and Otfried Jarren, 131–50. Wiesbaden: Verlag für Sozialwissenschaften.

Kamenova, Kalina, Vardit Ravitsky, Spencer McMullin, and Timothy A. Caulfield. 2016. "Media Portrayal of Non-Invasive Prenatal Testing: A Missing Ethical Dimension." J Sci Comm 15 (02): A03. doi:10.22323/2.15020203.

Kamenova, Kalina, Amir Reshef, and Timothy A. Caulfield. 2014. "Angelina Jolie's Faulty Gene: Newspaper Coverage of a Celebrity's Preventive Bilateral Mastectomy in Canada, the United States, and the United Kingdom." Genet Med 16 (7): 522–28. doi:10.1038/gim.2013.199.

Kamps, Rick, Rita D. Brandão, Bianca J. van den Bosch, Aimee D. C. Paulussen, Sofia Xanthoulea, Marinus J. Blok, and Andrea Romano. 2017. "Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification." Int J Mol Sci 18 (2): 308. doi:10.3390/ijms18020308.

Khaleel, Israa, Barbara C. Wimmer, Gregory M. Peterson, Syed T. R. Zaidi, Erin Roehrer, Elizabeth Cummings, and Kenneth Lee. 2020. "Health Information Overload Among Health Consumers: A Scoping Review." Patient Educ Couns 103 (1): 15–32. doi:10.1016/j.pec.2019.08.008.

Koeller, Diane R., Wendy R. Uhlmann, Deanna A. Carere, Robert C. Green, and J. S. Roberts. 2017. "Utilization of Genetic Counseling After Direct-to-Consumer Genetic Testing: Findings from the Impact of Personal Genomics (PGen) Study." J Genet Couns 26 (6): 1270–79. doi:10.1007/s10897-017-0106-7.

Kolb, Steffen. 2005. Mediale Thematisierung in Zyklen: Theoretischer Entwurf und empirische Anwendung. Köln: Herbert von Halem.

Kolb, Steffen, and Steffen Burkhardt. 2008. "Mission Impossible? Media Coverage of Scientific Findings." In Environmental Crises, edited by Hans von Storch, Richard S. J. Tol, and Götz Flöser, 65–81. Berlin, Heidelberg: Springer.

Laestadius, Linnea I., Jennifer R. Rich, and Paul L. Auer. 2017. "All Your Data (Effectively) Belong to Us: Data Practices Among Direct-to-Consumer Genetic Testing Firms." Genet Med 19 (5): 513–20. doi:10.1038/gim.2016.136.

Lebo, Patricia B., Franz Quehenberger, Lars-Peter Kamolz, and David B. Lumenta. 2015. "The Angelina Effect Revisited: Exploring a Media-Related Impact on Public Awareness." Cancer 121 (22): 3959–64. doi:10.1002/cncr.29461.

Lee, Sandra S.-J., and LaVera Crawley. 2009. "Research 2.0: Social Networking and Direct-to-Consumer (DTC) Genomics." Am J Bioeth 9 (6-7): 35–44. doi:10.1080/15265160902874452.

Lenk, Christian, Gunnar Duttge, Laura Flatau, Debora Frommeld, Wolfgang Poser, Markus Reitt, Thomas Schulze, Alexandra Weber, and Barbara Zoll. 2019. "A Look into the Future? Patients' and Health Care Staff's Perception and Evaluation of Genetic Information and the Right Not to Know." Am J Med Genet B 180 (8): 576–88. doi:10.1002/ajmg.b.32751.

Lippi, G., E. J. Favaloro, and M. Plebani. 2011. "Direct-to-Consumer Testing: More Risks Than Opportunities." Int J Clin Pract 65 (12): 1221–29. doi:10.1111/j.1742-1241.2011.02774.x.

Loi, Michele. 2016. "Direct to Consumer Genetic Testing and the Libertarian Right to Test." J Med Ethics 42 (9): 574–77. doi:10.1136/medethics-2015-102827.

Luhmann, Niklas. 1987. Soziale Systeme: Grundriss einer allgemeinen Theorie. Suhrkamp Taschenbuch. Wissenschaft. Frankfurt am Main: Suhrkamp.

———. 2009. Die Realität der Massenmedien. 4th ed. Neue Bibliothek der Sozialwissenschaften. Wiesbaden: Verlag für Sozialwissenschaften.

Lyssenko, Valeriya, and Markku Laakso. 2013. "Genetic Screening for the Risk of Type 2 Diabetes: Worthless or Valuable?" Diabetes Care 36 Suppl 2:S120-6. doi:10.2337/dcS13-2009.

Mand, Cara, Lynn Gillam, Martin B. Delatycki, and Rony E. Duncan. 2012. "Predictive Genetic Testing in Minors for Late-Onset Conditions: A Chronological and Analytical Review of the Ethical Arguments." J Med Ethics 38 (9): 519–24. doi:10.1136/medethics-2011-100055.

Manson, Neil, and Onora O'Neill. 2007. Rethinking Informed Consent in Bioethics. Cambridge: Cambridge University Press.

Marcon, Alessandro, Mark Bieber, and Timothy A. Caulfield. 2018. "Representing a "Revolution": How the Popular Press Has Portrayed Personalized Medicine." Genet Med 20 (9): 950–56. doi:10.1038/gim.2017.217.

Martin, Douglas K., Heather L. Greenwood, and Jeff Nisker. 2010. "Public Perceptions of Ethical Issues Regarding Adult Predictive Genetic Testing." Health Care Anal 18 (2): 103–12. doi:10.1007/s10728-009-0113-4.

Marzuillo, Carolina, Corrado de Vito, E. D'Andrea, Annalisa Rosso, and Paolo Villari. 2013. "Predictive Genetic Testing for Complex Diseases: A Public Health Perspective." An International Journal of Medicine 107 (2): 93–97. doi:10.1093/qjmed/hct190.

Matthes, Jörg. 2014. Framing. Konzepte 10. Baden-Baden: Nomos.

Maurer, Marcus. 2010. Agenda-Setting. Konzepte: Ansätze der Medien- und Kommunikationswissenschaft. Baden-Baden: Nomos.

McColgan, Peter, and Sarah J. Tabrizi. 2018. "Huntington's Disease: A Clinical Review." Eur J Neurol 25 (1): 24–34. doi:10.1111/ene.13413.

McGuire, Amy, Rebecca Fisher, Paul Cusenza, Kathy Hudson, Mark A. Rothstein, Deven McGraw, Stephen Matteson, John Glaser, and Douglas E. Henley. 2008. "Confidentiality, Privacy, and Security of Genetic and Genomic Test Information in Electronic Health Records: Points to Consider." Genet Med 10 (7): 495–99. doi:10.1097/GIM.0b013e31817a8aaa.

McQuail, Denis. 1987. Mass Communication Theory: An Introduction. 2nd ed. London: SAGE Publications.

Meienberg, Janine, Rémy Bruggmann, Konrad Oexle, and Gabor Matyas. 2016. "Clinical Sequencing: Is WGS the Better WES?" Hum Genet 135 (3): 359–62. doi:10.1007/s00439-015-1631-9.

Meisel, Susanne F., Deanna A. Carere, Jane Wardle, Sarah S. Kalia, Tanya A. Moreno, Joanna L. Mountain, J. S. Roberts, and Robert C. Green. 2015. "Explaining, Not Just Predicting, Drives Interest in Personal Genomics." Genome Med 7 (1): 74. doi:10.1186/s13073-015-0188-5.

Moscarello, Tia, Brittney Murray, Chloe M. Reuter, and Erin Demo. 2019. "Direct-to-Consumer Raw Genetic Data and Third-Party Interpretation Services: More Burden Than Bargain?" Genet Med 21 (3): 539–41. doi:10.1038/s41436-018-0097-2.

Neuman, W. Russell, and Lauren Guggenheim. 2011. "The Evolution of Media Effects Theory: A Six-Stage Model of Cumulative Research." Commun Theor 21 (2): 169–96. doi:10.1111/j.1468-2885.2011.01381.x.

Ntavatzikos, Anastasios, Aris Spathis, Paul Patapis, Nikolaos Machairas, George Peros, Stefanos Konstantoudakis, Danai Leventakou, Ioannis G. Panayiotides, Petros Karakitsos, and Anna Koumarianou. 2017. "Integrating TYMS, KRAS and BRAF Testing in Patients with Metastatic Colorectal Cancer." World J Gastroenterol 23 (32): 5913–24. doi:10.3748/wjg.v23.i32.5913.

Oliveri, Serena, Heidi C. Howard, Chiara Renzi, Mats G. Hansson, and Gabriella Pravettoni. 2016. "Anxiety Delivered Direct-to-Consumer: Are We Asking the Right Questions About the Impacts of DTC Genetic Testing?" J Med Genet 53 (12): 798–99. doi:10.1136/jmedgenet-2016-104184.

O'Neill, Onora. 2002. Autonomy and Trust in Bioethics. Gifford lectures 2001. Cambridge, New York: Cambridge University Press.

Peters, Hans P. 1994. "Mass Media as an Information Channel and Public Arena." Risk 5: 241.

Peters, Hans P., Harald Heinrichs, Arlena Jung, Monika Kallfass, and Imme Petersen. 2008. "Medialisierung der Wissenschaft als Voraussetzung ihrer Legitimierung und politischen Relevanz." In Wissensproduktion und Wissenstransfer: Wissen im Spannungsfeld von Wissenschaft, Politik und Öffentlichkeit, edited by Renate Mayntz, Friedhelm Neidhardt, Peter Weingart, and Ulrich Wengenroth, 269–92. Science studies. Bielefeld: transcript Verlag.

Plothner, Marika, Mike Klora, Daniel Rudolph, and Graf von der Schulenburg, Johann-Matthias. 2017. "Health-Related Genetic Direct-to-Consumer Tests in the German Setting: The Available Offer and the Potential Implications for a Solidarily Financed Health-Care System." Public Health Genom 20 (4): 203–17. doi:10.1159/000477559.

Prior, Lindsay. 2003. "Belief, Knowledge and Expertise: The Emergence of the Lay Expert in Medical Sociology." Sociol Health Illn 25:41–57. doi:10.1111/1467-9566.00339.

Relling, Mary V., and William E. Evans. 2015. "Pharmacogenomics in the Clinic." Nature 526 (7573): 343–50. doi:10.1038/nature15817.

Richards, Sue, Nazneen Aziz, Sherri Bale, David Bick, Soma Das, Julie Gastier-Foster, Wayne W. Grody et al. 2015. "Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology." Genet Med 17 (5): 405–24. doi:10.1038/gim.2015.30.

Rivera-Navarro, Jesús, Esther Cubo, and Natividad Mariscal. 2015. "Analysis of the Reasons for Non-Uptake of Predictive Testing for Huntington's Disease in Spain: A Qualitative Study." J Genet Couns 24 (6): 1011–21. doi:10.1007/s10897-015-9840-x.

Rödder, Simone, Martina Franzen, and Peter Weingart, eds. 2012. The Sciences' Media Connection - Public Communication and Its Repercussions. Sociology of the sciences A yearbook. Dordrecht: Springer.

Ronneberger, Franz. 2002. "Funktionen des Systems Massenmedien." In Mediensysteme im Wandel : Struktur, Organisation und Funktionen der Massenmedien, edited by Hannes Haas and Otfried Jarren. 3rd rev. ed., 61–68. Studienbücher zur Publizistik- und Kommunikationswissenschaft. . Edited by Hannes Haas. Wien: Braumüller.

Rose, Abigail L., Nikki Peters, Judy A. Shea, and Katrina Armstrong. 2005. "Attitudes and Misconceptions About Predictive Genetic Testing for Cancer Risk." Community Genet 8 (3): 145–51. doi:10.1159/000086757.

Ross, Laine F., Howard M. Saal, Karen L. David, and Rebecca R. Anderson. 2013. "Technical Report: Ethical and Policy Issues in Genetic Testing and Screening of Children." Genet Med 15 (3): 234–45. doi:10.1038/gim.2012.176.

Rothstein, Mark A. 2005. "Genetic Exceptionalism & Legislative Pragmatism." Hastings Cent Rep 35 (4): 27–33. doi:10.1353/hcr.2005.0046.

Ruiz-Canela, Miguel, J. I. Valle-Mansilla, and Daniel P. Sulmasy. 2011. "What Research Participants Want to Know About Genetic Research Results: The Impact of "Genetic Exceptionalism"." J Empir Res Hum Res Ethics 6 (3): 39–46. doi:10.1525/jer.2011.6.3.39.

Saelaert, Marlies, Heidi Mertes, Elfride de Baere, and Ignaas Devisch. 2018. "Incidental or Secondary Findings: An Integrative and Patient-Inclusive Approach to the Current Debate." Eur J Hum Genet 26 (10): 1424–31. doi:10.1038/s41431-018-0200-9.

Schaper, Manuel, and Silke Schicktanz. 2018. "Medicine, Market and Communication: Ethical Considerations in Regard to Persuasive Communication in Direct-to-Consumer Genetic Testing Services." BMC Med Ethics 19 (1): 56. doi:10.1186/s12910-018-0292-3.

Schaper, Manuel, Sabine Wöhlke, and Silke Schicktanz. 2019. ""I Would Rather Have It Done by a Doctor"-Laypeople's Perceptions of Direct-to-Consumer Genetic Testing (DTC GT) and Its Ethical Implications." Med Health Care Philos 22 (1): 31–40. doi:10.1007/s11019-018-9837-y.

Schulz, Winfried. 1990. Die Konstruktion von Realität in den Nachrichtenmedien : Analyse der aktuellen Berichterstattung. 2nd ed. Alber-Broschur Kommunikation. Freiburg i. Br., München: Verlag Karl Alber.

Shaw, David, and Bernice Elger. 2013. "Evidence-Based Persuasion: An Ethical Imperative." JAMA-J Am Med Assoc 309 (16): 1689–90. doi:10.1001/jama.2013.2179.

Shoemaker, Pamela J. 2002. "A New Gatekeeping Model." In McQuail's Reader in Mass Communication Theory, edited by Denis McQuail, 253–59. London: SAGE Publications.

Singleton, Amanda, Lori H. Erby, Kathryn V. Foisie, and Kimberly A. Kaphingst. 2012. "Informed Choice in Direct-to-Consumer Genetic Testing (DTCGT) Websites: A Content Analysis of Benefits, Risks, and Limitations." J Genet Couns 21 (3): 433–39. doi:10.1007/s10897-011-9474-6.

Skirton, Heather, Lesley Goldsmith, Leigh Jackson, and Aad Tibben. 2013. "Quality in Genetic Counselling for Presymptomatic Testing — Clinical Guidelines for Practice Across the Range of Genetic Conditions." Eur J Hum Genet 21 (3): 256–60. doi:10.1038/ejhg.2012.174.

Stewart, Kelly F. J., Dasa Kokole, Anke Wesselius, Schols, Annemie M W J, Maurice P. Zeegers, Hein de Vries, and Liesbeth A. D. M. van Osch. 2018a. "Factors Associated with Acceptability, Consideration and Intention of Uptake of Direct-to-Consumer Genetic Testing: A Survey Study." Public Health Genom 21 (1-2): 45–52. doi:10.1159/000492960.

Stewart, Kelly F. J., Anke Wesselius, Maartje A. C. Schreurs, Schols, Annemie M W J, and Maurice P. Zeegers. 2018b. "Behavioural Changes, Sharing Behaviour and Psychological Responses After Receiving Direct-to-Consumer Genetic Test Results: A Systematic Review and Meta-Analysis." J Community Genet 9 (1): 1–18. doi:10.1007/s12687-017-0310-z.

Stilgoe, Jack, Simon Locke, and James Wilsdon. 2014. "Why Should We Promote Public Engagement with Science?" Public Underst Sci 23 (1): 4–15. doi:10.1177/0963662513518154.

Stoljar, Natalie. 2018. "Feminist Perspectives on Autonomy." In the Stanford Encyclopedia of Philosophy, edited by Edward N. Zalta. Winter 2018: Metaphysics Research Lab, Stanford University. https://plato.stanford.edu/entries/feminism-autonomy/. Accessed October 14, 2019.

Sulmasy, Daniel P. 2015. "Naked Bodies, Naked Genomes: The Special (but Not Exceptional) Nature of Genomic Information." Genet Med 17 (5): 331–36. doi:10.1038/gim.2014.111.

Swiss Academy of Medical Sciences. 2011. Genetik Im Medizinischen Alltag: Ein Leitfaden Für Die Praxis. 2nd rev. ed. Accessed January 29, 2020. https://www.samw.ch/dam/jcr:e7c2f6b4-73dc-47ba-8afc-32373fb30171/leitfaden_samw_genetik.pdf.

Swiss Federal Council. 2004. "CC 810.12 Federal Act of 8 October 2004 on Human Genetic Testing (HGTA)." Accessed January 23, 2020. https://www.admin.ch/opc/en/classified-compilation/20011087/index.html.

———. 2017. "Draft of the Revised Human Genetic Testing Act." Accessed January 23, 2020. https://www.bag.admin.ch/dam/bag/de/dokumente/biomed/genetischeuntersuchung/aktuelle-rechtsetzungsprojekte/entwurf-gumg-2017.pdf.download.pdf/entwurf-gumg-2017.pdf.

Tandy-Connor, Stephany, Jenna Guiltinan, Kate Krempely, Holly LaDuca, Patrick Reineke, Stephanie Gutierrez, Phillip Gray, and Brigette Tippin Davis. 2018. "False-Positive Results Released by Direct-to-Consumer Genetic Tests Highlight the Importance of Clinical Confirmation Testing for Appropriate Patient Care." Genet Med 20 (12): 1515–21. doi:10.1038/gim.2018.38.

Turrini, Mauro, and Barbara Prainsack. 2016. "Beyond Clinical Utility: The Multiple Values of DTC Genetics." Applied & Translational Genomics 8:4–8. doi:10.1016/j.atg.2016.01.008.

U.S. Food and Drug Administration. 2017. "FDA Allows Marketing of First Direct-to-Consumer Tests That Provide Genetic Risk Information for Certain Conditions." Accessed January 23, 2020. https://www.fda.gov/news-events/press-announcements/fda-allows-marketing-first-direct-consumer-tests-provide-genetic-risk-information-certain-conditions.

———. 2018. "FDA Authorizes, with Special Controls, Direct-to-Consumer Test That Reports Three Mutations in the BRCA Breast Cancer Genes." Accessed January 23, 2020. https://www.fda.gov/news-events/press-announcements/fda-authorizes-special-controls-direct-consumer-test-reports-three-mutations-brca-breast-cancer.

Vallance, Hilary, and Jason Ford. 2003. "Carrier Testing for Autosomal-Recessive Disorders." Crit Rev Clin Lab Sci 40 (4): 473–97. doi:10.1080/10408360390247832.

Vayena, Effy. 2015. "Direct-to-Consumer Genomics on the Scales of Autonomy." J Med Ethics 41 (4): 310–14. doi:10.1136/medethics-2014-102026.

Vayena, Effy, Christian Ineichen, Elia Stoupka, and Ernst Hafen. 2014. "Playing a Part in Research? University Students' Attitudes to Direct-to-Consumer Genomics." Public Health Genom 17 (3): 158–68. doi:10.1159/000360257.

Vergote, Ignace, Susana Banerjee, Anne-Marie Gerdes, Christi van Asperen, Christian Marth, Fatima Vaz, Isabelle Ray-Coquard et al. 2016. "Current Perspectives on Recommendations for BRCA Genetic Testing in Ovarian Cancer Patients." Eur J Cancer 69:127–34. doi:10.1016/j.ejca.2016.10.006.

Vermeulen, Eric, Lidewij Henneman, Carla G. van El, and Martina C. Cornel. 2014. "Public Attitudes Towards Preventive Genomics and Personal Interest in Genetic Testing to Prevent Disease: A Survey Study." Eur J Public Health 24 (5): 768–75. doi:10.1093/eurpub/ckt143.

Wasson, Katherine, Tonya N. Sanders, Nancy S. Hogan, Sara Cherny, and Kathy J. Helzlsouer. 2013. "Primary Care Patients' Views and Decisions About, Experience of and Reactions to Direct-to-Consumer Genetic Testing: A Longitudinal Study." J Community Genet 4 (4): 495–505. doi:10.1007/s12687-013-0156-y.

Weingart, Peter. 2012. "The Lure of the Mass Media and Its Repercussions on Science." In Rödder, Franzen, and Weingart 2012, 17–34.

Weingart, Peter, Simone Rödder, and Martina Franzen. 2012. "Dimensions of Medialization: Concluding Remarks." In Rödder, Franzen, and Weingart 2012, 363–74.

Welie, J. V., and S. P. Welie. 2001. "Patient Decision Making Competence: Outlines of a Conceptual Analysis." Med Health Care Philos 4 (2): 127–38. doi:10.1023/a:1011441816143.

White, Mary T. 1998. "Decision-Making Through Dialogue: Reconfiguring Autonomy in Genetic Counseling." Theor Med Bioeth 19 (1): 5–19. doi:10.1023/a:1009976119490.

Zimmern, R. L., and Muin J. Khoury. 2012. "The Impact of Genomics on Public Health Practice: The Case for Change." Public Health Genom 15 (3-4): 118–24. doi:10.1159/000334840.

Chapter 2: Methodology

To achieve the objectives presented in chapter 1.5, this dissertation implements an interdisciplinary mixed-methods approach. Two projects were conducted within this dissertation. First, an interview study with people undergoing counselling for cancer predisposition genetic testing was performed to get insights into the individuals' perspective (see also chapters 3-5). Methodologically, this project based on a grounded theory approach (Strauss and Corbin 1990), which originates from sociology.[1] Besides, aspects of information-seeking behaviour were analysed using thematic analysis (chapter 4).

In the second project, the public's perspective was assessed with a media content analysis (see also chapters 6-8), following the research tradition of the media and communication sciences.[2] Print media represented mass media in this project, which consisted of three parts: The project started with a quantitative screening study focussing on Swiss coverage, to determine what topics about medical genetics Swiss print media covered and what language journalists used as a pilot for the other studies. In the second part, broadsheet newspaper coverage in the UK and Switzerland were compared regarding coverage about predictive genetic testing. The third part included a semi-quantitative content analysis to investigate how UK broadsheet newspapers report on ethical issues in predictive genetic testing.

The approach to combine media content analyses with a qualitative interview study as an empirical basis to draw ethically relevant conclusions was explorative. The following chapters provide a critical reflection on the methodological challenges of the methodologies as an addition to the description in the methods sections in chapters 3-8. Chapter 2.3 then explains how the empirical work connected to normative research, which is a critical and often discussed aspect of empirical bioethics. Finally, the collaborators involved in these projects are presented in chapter 2.4.

2.1 Interview study

The interview study intended to consult people already in touch with genetic testing as a subsample of the general population and to get insights into the individual's perspective on informed decision-making. The aim was the qualitative assessment of information strategies, reasons and attitudes of people undergoing genetic counselling for testing of inherited cancer risk. This qualitative study was exploratory: No such study had been conducted in Switzerland before.

1 See chapter 3.3 for more details on the methodology.

2 See chapters 6.4 and 7.3 for more details on the methodology.

2.1.1 Recruitment and data collection

The grounded theory approach was the framework for data collection and analysis. This method aims to create a theory that can be generally applied to similar contexts as the study (Strauss and Corbin 1990). As a pilot study, the interviewer passively observed one genetic counselling sessions and one support group with carriers of a risk-increasing genetic variant for breast and ovarian cancer.

Medical doctors who provided genetic counselling recruited participants. They either handed over the study information leaflet[3] to eligible participants during a counselling session or sent them a letter. We collaborated with four medical doctors at the Inselspital Bern or the University Hospital Basel. People who went to at least one genetic counselling session, spoke either German, French or English, were 18-75 years old, not pregnant and in a good psychological state were eligible for the study. For ethical reasons, the recruiting doctors were instructed not to include particularly vulnerable or psychologically unstable patients. In line with the grounded theory approach, the inclusion criteria were refined during the study, aiming to reach theoretical saturation: At a later stage, we specifically searched for men and individuals refusing genetic testing.

Those interested in participating either contacted the interviewer directly or permitted their medical doctor to share their contact details with the interviewer. The interviewer additionally explained the study implications and details on the phone. All participants gave written informed consent before the interview and received no compensation for their participation. At the end of the interview, all participants were asked if they agreed to be contacted again to give feedback on results.

For data triangulation, we performed two interviews with two medical doctors providing genetic counselling after the completion of the patient interviews. Medical doctors were asked about their points of view and impressions, guided by the preliminary analysis of the patient interviews. Since no additional aspects came up, we decided to stop data collection after two expert interviews and assumed that theoretical saturation had been reached.

The semi-structured interviews were face to face[4] and took place either at the Institute for Biomedical Ethics, the University Hospital Bern or, upon their request, in the participants' home. Interviews were recorded, transcribed ad verbatim and encrypted upon transcription. The regional ethics committees of Northwest and Central Switzerland, as well as Bern, approved the study (No 2017-00316).

3 See Appendix A-3.

4 Interview guides are provided in Appendix A.

2.1.2 Data analysis

One basic principle of data analysis according to grounded theory is the iterative, cyclic character and the constant comparison in the analysis: data collection and analysis are in constant interplay during the study, and during data analysis, the different interviews and different participants and contents are constantly compared to each other (Strauss and Corbin 1990).

Accordingly, we first applied line-by-line coding to each interview before focusing on larger data elements, forming categories that reflected the content of the interview. After six interviews, we sightly adapted the interview guide to prompt for aspects that we found interesting when analysing them. The defined codes were then reorganised, related to each other and put into a hierarchy by constant comparison of the codes within one interview and among different interviews. Text passages with similar codes from different interviews were compared to find similarities and differences. Categories were then rearranged to identify and review the central categories, laying the basis for theory building. Each finding was critically challenged, consulting the data for supporting and opposing statements. We wrote memos during the whole study period, and the data analysis was discussed in the research team. The MaxQDA data analysis software supported the data analysis. Chapters 3 and 5 present the results of the analysis.

Statements regarding information strategies did not fit in the grounded theory, as they were mainly prompted by the interviewer and did not come up from participants themselves. Therefore, an additional thematic analysis was applied, investigating aspects of information-seeking behaviour. Chapter 4 presents the results and provides more details regarding the methodology applied to this analysis.

2.1.3 Challenges

The main obstacle for this study was recruitment. Four medical doctors providing genetic counselling distributed the study information. However, there was no control over the frequency they provided the study information. Retrospectively, adding the option of snowballing or purposive sampling and including another hospital from the French-speaking part of Switzerland might have improved the outcome. The grounded theory approach (Strauss and Corbin 1990) was chosen because the aim was to generate hypotheses that can be quantitatively validated.[5]

5 This validation was not part of this dissertation but remains an ambition for future research projects.

2.2 Newspaper content analyses

Content analyses can be quantitative, qualitative or a mix of both. The selection of the most appropriate approach depends on the research question. For comparative studies, such as the comparison of newspaper coverage between Swiss language regions (chapter 6) or between the German-speaking part of Switzerland and the UK (chapter 7), quantitative approaches are useful. However, quantitative content analyses require simplifications to standardise data collection, which limits the content depth. Thus, qualitative approaches are more suitable for investigations concerning the detailed content of newspaper coverage. The next sections will justify why newspapers were analysed and provide insights into the methodological challenges faced during the newspaper content analyses, as these aspects were not extensively covered in the publications (chapters 6-8) due to lack of space.

2.2.1 Justification for using newspaper content analyses

Mass media include print and online media, radio and television. In this dissertation, print media, especially broadsheet newspapers (chapters 7-8), represent mass media communication, excluding tabloids and regional newspapers, radio, television and online news platforms. This section will justify why broadsheet newspapers provide a suitable sample of mass media for this investigation, despite their economic difficulties and a steady decrease in readership (Meier, Bonfadelli, and Trappel Josef 2012; Blum et al. 2011).[6]

Broadsheet newspapers influence society through their impact on societal elites, other journalists and the reporting of other media (Kamber and Imhof 2011; Jarren and Vogel 2011; Jandura and Brosius 2011).[7] Thus, even though tabloids, online news platforms or television often reach a higher number of people than broadsheet newspapers, their influence on society is amplified because elites and journalists mainly read broadsheets (Jandura and Brosius 2011). Opinion-leading broadsheet newspapers have a comparatively high readership, are of national or international relevance, cover a variety of topics and have specialised sections, for instance, a science section (Wilke 2009).

Additionally, content analyses of newspapers have methodological advantages compared to other mass media. First, newspaper articles are published in established entities and are thus not subject to constant change. Databases, such as Factiva and LexisNexis, collect newspaper articles

6 Free-of-charge news available online or through the cost-free press, for instance 20 minutes in Switzerland, pressure traditional newspapers economically.

7 Accordingly, the inter-media agenda-setting theory was built upon the observation that few media influence mass media coverage worldwide (Golan 2006; Mathes and Pfetsch 1991).

systematically and provide reliable and transparent search algorithms for data selection. By contrast, the content of the World Wide Web is continuously changing. The algorithms of online search engines are intransparent and biased (McMillan 2009). Moreover, due to the variety and size of the mass media system, some media selection is unavoidable for media content analyses. Otherwise, extensive computational knowledge would be required to meet challenges regarding data volume and variability.

In summary, newspaper content analyses provide a stable, reliable and thus quantifiable option to analyse a public debate retrospectively. Still, this does not imply that investigating other mass media is not possible or less critical. For instance, online news might differ to print media regarding news coverage (Cacciatore et al. 2012; Anderson, Brossard, and Scheufele 2012) and they have a high readership. Social media are relevant as well, as they increasingly influence the public's knowledge and opinion (Weeks, Ardèvol-Abreu, and Gil de Zúñiga 2017). These media, however, goes beyond the scope of this dissertation.

2.2.2 Challenges in quantitative newspaper content analyses

In the process of the quantitative newspaper analysis (presented in chapters 6 and 7), we were facing several methodological challenges. The first challenge concerned the systematic and exhaustive collection of newspaper articles about predictive genetic testing. Therefore, the systematic screening study for medical genetics topics covered in Swiss print media[8] served as a pilot to determine the terms journalists use to explain or describe predictive genetic testing. It allowed the development of an exhaustive search algorithm regarding predictive genetic testing.[9] We found that the expressions used in newspaper articles to describe or explain predictive genetic testing were often unspecific. Thus, the search algorithm included general search terms, such as "genetic testing", and we had to exclude the majority of search hits manually.[10]

Second, we generated a codebook for quantitative data collection, which needed to meet the quality criteria of reliability and validity.[11] The validity was addressed by adapting codebooks from relevant previous studies. Inter- and intra-coder-reliability was measured quantitatively.[12] During the process of reliability testing, the codebook was specified and simplified until reliability scores were sufficiently high. This process was especially challenging for the part of the

8 See chapter 6 and Appendix B for the corresponding publication and methodology.
9 The detailed search algorithms are provided in Appendix B-1 and Appendix C-1.
10 Please refer to chapter 7.3 and Appendix C-2 for the inclusion and exclusion criteria.
11 The full codebook is provided in Appendix C-2.
12 See chapter 7.3 for more details.

codebook that captured the topic-specific content. To obtain the coding list, we performed a summarising qualitative content analysis (Mayring 2015). Following a protocol from Potthoff (2012), the initial aim was to maintain a medium level of abstraction to preserve the specific content of the articles. The output of this qualitative pre-study was a list of 89 "content statements" in seven categories (see Appendix C-4). The initial strategy was to apply this list to the articles to quantitatively collect content-specific data, and then to investigate how these statements connect to evaluations and stakeholders through statistical analysis (Potthoff 2012).[13] However, no acceptable reliability scores were obtained, despite multiple rounds of training. Therefore, the content statements were further condensed to a more abstract level, resulting in a 14-item list of topics for which we reached good inter- and intra-personal reliability scores.

The third challenge was choosing a suitable data collection software. Because we collected data on three levels, a simple excel spreadsheet was not sufficient for data entry. First, some variables, such as the article title or the date of publication, were coded on the article level. Additionally, we assigned one or several topics to each article. Second, on the topic level, we coded one or several stakeholders commenting on the topic. On the third level, stakeholder-specific evaluations were collected.[14] Microsoft Access 2010 allowed for such multi-layered data entry and provided three spreadsheets on the different levels used for statistical data analysis with IBM SPSS 24 software.

The fourth challenge concerned language issues: English and German articles required different search algorithms. Besides, no collaborator was speaking German, French and English. Thus, the French-speaking part of Switzerland was excluded from the analysis after the pilot study. In summary, we had to address challenges related to search algorithms, reliability of the analysis, and administrative challenges concerning software and collaborators for the quantitative content analysis (chapter 7). Additional challenges arose in the analysis of ethical issues.

2.2.3 Challenges in the newspaper content analysis of ethical issues

The third part of the newspaper content analysis investigated how media portrayed ethical issues about predictive genetic testing (chapter 8). We chose a deductive approach to quantify how many times newspapers mentioned a particular ethical issue, and the coding list was derived from reviewing scientific literature. However, newspapers framed some issues differently than scientific literature. Thus, the original coding tree was revised, which required additional inductive steps.

13 Matthes and Kohring (2008) had proposed that this approach was more objective than other approaches.
14 See Appendix C-2 for the codebook.

Additionally, it was challenging to reach acceptable inter-coder reliability scores, as newspapers often mentioned ethical issues implicitly. Consequently, the coders (BZ and DS) frequently detected ethical issues in differing text passages. Therefore, we included the relevant text passages upon discussion and refrained from quantitative analysis, as it would not have proved reliable. However, once the text passages were selected, ethical principles were reliably allocated, which allowed for a limited quantitative comparison (see chapter 8.4). We also emphasised qualitative aspects by providing detailed quotes and examples.

In conclusion, it proved challenging to reliably analyse ethical issues in newspaper coverage, as such aspects were often implicitly mentioned. Qualitative methodologies might be a suitable alternative to assess these aspects, but we were also successfully applying a coding scheme derived from ethical issues discussed among the scientific community. This study is one example to use empirical methodologies in bioethics, which will be discussed in the following section.

2.3 Empirical bioethics: extrapolating normative conclusions

The design of this dissertation project began with a strong emphasis on empirical research. While the justification and the relevance of the research topic based on ethical grounds,[15] the research questions were initially mainly empirical. During the process of data analysis, normative aspects of the work became more salient. Normative research questions arose during data analysis: First, we performed a follow-up study about newspaper coverage of ethical issues of predictive genetic testing (chapter 8), using the British part of the data material from the quantitative media content analysis. Second, normatively relevant aspects of individual and relational aspects of decision-making were identified in the interview data and connected to theories of relational autonomy (chapter 5). At the last stages of the dissertation, these normative insights gathered through the empirical work were generalised and summarised (chapters 9 and 10).

In summary, the dissertation project included two phases regarding the connection between empirical and normative parts. It started with an exploratory phase to identify current practices and assess the current situation.[16] From a bioethical perspective, this first phase remained on a descriptive level. However, the study's strong foundations in other disciplines provided a framework for interpretation that was meaningful for the later steps. Referring to these disciplines

15 Issues of informed decision-making in the context of the progress and increasing availability of genetic testing.

16 Chapters 3, 4, 6 and 7.

guided methodological rigour and quality assessment, factors that have been criticised in empirical bioethics research (Adler and Zlotnik Shaul 2012). In the second phase of the dissertation, normative and empirical considerations and findings influenced each other.[17] Others have referred to this approach as "critical applied ethics" (Leget, Borry, and de Vries 2009; Molewijk et al. 2004).

2.4 Collaborators

For the quantitative newspaper content analyses, Prof. Dr Steffen Kolb provided supervision and practical guidance. PhD, Master and Bachelor students were involved for reliability testing and qualitative analyses.[18] Dr David Shaw was involved in interpreting the results, revising the manuscripts and contributed to the empirical work for the qualitative analysis on ethical issues mentioned in newspaper coverage (chapter 8). Prof. Dr Bernice Elger discussed the results, commented on the study design and read and revised all articles.

Dr med. Insa Koné (chapters 3 and 5) and Julia Fanderl (chapter 4) supported the interview analysis. Besides, four medical doctors providing genetic counselling (Prof. Dr Karl Heinimann, Dr. med. Laura Knabben, Dr. med. Nicole Bürki, Dr. med. Manuela Rabaglio) recruited participants, were involved in the study design and revised the manuscripts. Dr David Shaw and Prof. Dr Bernice Elger provided advice and support for the study design and the ethics approval procedure, were involved in result interpretation and the revision and proofreading of the manuscripts. The theoretical considerations in published articles resulted from in-depth discussions with Prof. Dr Bernice Elger and Dr David Shaw, based on the empirical data gathered.

2.5 References

Adler, Daniel, and Randi Zlotnik Shaul. 2012. "Disciplining Bioethics: Towards a Standard of Methodological Rigor in Bioethics Research." Account Res 19 (3): 187–207. doi:10.1080/08989621.2012.692971.

Anderson, Ashley A., Dominique Brossard, and Dietram A. Scheufele. 2012. "News Coverage of Controversial Emerging Technologies. Evidence for the Issue Attention Cycle in Print and Online Media." Politics Life Sci 31 (1-2): 87–96. doi:10.2990/31_1-2_87.

Blum, Roger, Heinz Bonfadelli, Kurt Imhof, and Otfried Jarren, eds. 2011. Krise Der Leuchttürme Öffentlicher Kommunikation: Vergangenheit Und Zukunft Der Qualitätsmedien. Wiesbaden: Verlag für Sozialwissenschaften.

17 Chapters 5 and 8.

18 Fabian Zimmermann, Noah Aebi and Jana Möller.

Cacciatore, Michael A., Ashley A. Anderson, Doo-Hun Choi, Dominique Brossard, Dietram A. Scheufele, Xuan Liang, Peter J. Ladwig, Michael Xenos, and Anthony Dudo. 2012. "Coverage of Emerging Technologies: A Comparison Between Print and Online Media." New Media Soc 14 (6): 1039–59. doi:10.1177/1461444812439061.

Golan, Guy. 2006. "Inter-Media Agenda Setting and Global News Coverage." Journalism Stud 7 (2): 323–33. doi:10.1080/14616700500533643.

Jandura, Olaf, and Hans-Bernd Brosius. 2011. "Wer Liest Sie (Noch)? Das Publikum Der Qualitätszeitungen." In Blum et al. 2011, 195-206.

Jarren, Otfried, and Martina Vogel. 2011. "„Leitmedien" Als Qualitätsmedien: Theoretisches Konzept Und Indikatoren." In Blum et al. 2011, 17–30.

Kamber, Esther, and Kurt Imhof. 2011. "Qualitätsmedien Sind Wissensvermittler." In Blum et al. 2011, 131–50.

Leget, Carlo, Pascal Borry, and Raymond de Vries. 2009. "'nobody Tosses a Dwarf!' the Relation Between the Empirical and the Normative Reexamined." Bioethics 23 (4): 226–35. doi:10.1111/j.1467-8519.2009.01711.x.

Mathes, Rainer, and Barbara Pfetsch. 1991. "The Role of the Alternative Press in the Agenda-Building Process: Spill-over Effects and Media Opinion Leadership." Eur J Comm 6 (1): 33–62. doi:10.1177/026732319100600103.

Matthes, Jörg, and Matthias Kohring. 2008. "The Content Analysis of Media Frames: Toward Improving Reliability and Validity." J Commun 58 (2): 258–79. doi:10.1111/j.1460-2466.2008.00384.x.

Mayring, Philipp. 2015. Qualitative Inhaltsanalyse: Grundlagen Und Techniken. 12 rev. ed. Weinheim: Beltz.

McMillan, Sally J. 2009. "The Challenge of Applying Content Analysis to the World Wide Web." In the Content Analysis Reader, edited by Klaus Krippendorff and Mary A. Block, 60–67. Thousand Oaks, Calif: SAGE Publications.

Meier, Werner A., Heinz Bonfadelli, and Trappel Josef, eds. 2012. Gehen in den Leuchttürmen die Lichter aus? Was aus den Schweizer Leitmedien wird. Swiss: Forschung und Wissenschaft. Edited by Werner A. Meier. Münster: LIT Verlag.

Molewijk, Bert, Anne M. Stiggelbout, Wilma Otten, Heleen M. Dupuis, and Job Kievit. 2004. "Scientific Contribution. Empirical Data and Moral Theory. A Plea for Integrated Empirical Ethics." Med Health Care Philos 7 (1): 55–69. doi:10.1023/B:MHEP.0000021848.75590.b0.

Potthoff, Matthias. 2012. Medien-Frames Und Ihre Entstehung. Wiesbaden: Verlag für Sozialwissenschaften.

Strauss, Anselm, and Juliet M. Corbin. 1990. Basics of Qualitative Research: Grounded Theory Procedures and Techniques. Newbury Park, Calif: SAGE Publications.

Weeks, Brian E., Alberto Ardèvol-Abreu, and Homero Gil de Zúñiga. 2017. "Online Influence? Social Media Use, Opinion Leadership, and Political Persuasion." Int J Public Opin Res 29 (2): 214-239. doi:10.1093/ijpor/edv050.

Wilke, Jürgen. 2009. "Historische und intermediale Entwicklungen von Leitmedien: Journalistische Leitmedien in Konkurrenz zu anderen." In Leitmedien: Konzepte - Relevanz - Geschichte. Vol. 1, edited by Daniel Müller, Annemone Ligensa, and Peter Gendolla. 2 vols, 29–52. Medienumbrüche Vol 1. . Edited by Daniel Müller. Bielefeld: transcript Verlag.

Chapter 3: How the "Control-Fate- Continuum" Helps Explain the Genetic Testing Decision-making Process: A Grounded Theory Study

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3.1 Abstract

Genetic testing decision-making for cancer predisposition is inherently complex. Understanding the mechanisms and influencing factors of the decision-making process is essential for genetic counselling and has not yet been investigated in Switzerland. This study's aim is thus to provide a theory about the individual's decision-making process regarding genetic testing for cancer predispositions in order to provide medical geneticists and genetic counsellors with insights into the needs and expectations of counsellees. We interviewed at-risk individuals who underwent genetic counselling in a clinical setting in Switzerland, using a grounded theory approach. Based on the interview data, we propose that a control-fate-continuum, which is part of the individuals' life philosophy, importantly influences the decision-making process. Those in need for control decide differently compared to those leaving their future to fate. Several psychosocial factors influence the position on the control-fate-continuum: *"Looking for Certainty"*; *"Anticipating Consequences"*; *"Being Socially Influenced"*; *"Deciding Intuitively vs Reflectively"*; and *"Simplifying Risks"*. The control-fate-continuum theory gives insights into the possible reasons behind decision-making regarding genetic testing for cancer predispositions. It includes both acceptors and decliners of genetic testing. Our theory helps healthcare professionals offering genetic counselling to anticipate problems within at-risk families and adapting their services to people's needs.

3.2 Introduction

Genetic testing for cancer predispositions was used for the first time in 1993/1994 when testing for hereditary breast and ovarian cancer (HBOC) became possible (Castilla et al., 1994; Miki et al., 1994). Pre-test genetic counselling aims to enable individuals to make an informed decision regarding the genetic test and facilitates self-management. Besides disease-specific information, genetic counselling should provide information regarding possible psychological, social and legal consequences of genetic testing. Healthcare professionals offering genetic counselling should tailor the content of the consultation to the individual needs and expectations of their counsellees (Skirton et al., 2013). Thus, it is essential for them to anticipate which needs and expectations counsellees have in the decision-making process for or against genetic testing.

The main reasons for undergoing genetic testing for cancer predisposition are the early detection of cancer and a feeling of responsibility towards family members (Claes et al., 2004; Etchegary et al., 2009; Foster et al., 2002; Hallowell et al., 2005). Many at-risk individuals make their decision intuitively and have already decided before genetic counselling to undergo genetic

testing (Battistuzzi et al., 2019; Godino et al., 2018). Reasons declining genetic testing for cancer predisposition include lack of knowledge, anxiety, perception of non-relevance, objections from relatives and privacy concerns (Kanga-Parabia et al., 2018; Keogh et al., 2017; Schlich-Bakker et al., 2007). While people perceive themselves as well informed, they perform poorly on knowledge surveys (Hoffman et al., 2010a).

Most studies about the decision-making process for genetic testing for cancer predisposition stem from English-speaking countries, and there is a particular lack of data from the German-speaking area. While recent studies have investigated this phenomenon in Southern Europe, particularly in Italy (Battistuzzi et al., 2019; Godino et al., 2018), there are considerable differences between these countries and other European countries regarding legislation (Harper et al., 2014; Kalokairinou et al., 2018; Soini, 2012), public discourse (Zimmermann et al., 2019a) and culture (Binetti et al., 2006; Godino et al., 2018; Press et al., 2000). This study aims to fill the existing research gap by interviewing at-risk individuals from the German-speaking part of Switzerland.

In Switzerland, Article 14/1 of the Federal Act on Human Genetic Testing (HGTA) requires professional genetic counselling before and after predictive genetic testing (Swiss Federal Council, 2004). The decision-making process starts when a person first realises that he or she might be genetically predisposed to cancer. After that, the first genetic consultation takes place, which is mandatory in order to get a test referral. The healthcare professional offering genetic counselling discloses and discusses information about testing procedures and the meaning of test results with the counselee (Skirton et al., 2013; Swiss Federal Council, 2004). That includes implications for the patient and relatives, the meaning of positive and negative test results, and the medical consequences of the test. There is a mandatory reflection period (usually several weeks) after the pre-test genetic consultation (Art 14/4 HGTA) (Swiss Federal Council, 2004).

In order to help people through the inherently complex decision-making process of genetic testing for cancer predispositions, it is vital to assess how they make decisions about whether or not to take the genetic test and identify influencing factors. This article aims to explore how at-risk individuals in Switzerland experience the decision-making process of genetic testing for cancer predisposition.

3.3 Materials and methods

3.3.1 Strategy

In this qualitative interview study, we used a grounded theory approach for study design, sampling and data analysis (Charmaz, 2006; Strauss and Corbin, 1990). This well-established qualitative method aims to construct a theory grounded on data. We chose this method because we intended to theorise our findings and build new hypotheses, applying a post-positivist research paradigm (Annells, 1996). One researcher (BZ) has a background in genetics and a positive attitude towards genetic testing. Another team member (IK) is a medical doctor working in general practice and somewhat sceptical towards genetic testing. The different attitudes allowed for a critical and constant reflection on personal views and opinions. We also asked participants for feedback regarding the results of this study and performed two expert interviews with clinicians after the patient interviews to triangulate data and verify, support and complement our findings. The regional ethics committees of Northwest and Central Switzerland, as well as Bern, approved the study (No 2017-00316). All at-risk individuals signed an informed consent form before the interview.[1] Experts gave their informed consent verbally, in line with the ethics committee recommendations and Swiss law.

3.3.2 Recruitment

We recruited at-risk individuals through four medical doctors responsible for genetic counselling at Basel and Bern University Hospitals. They informed eligible counsellees about the study and handed over the study information leaflet. Those interested in participation contacted the interviewer proactively, except for four at-risk individuals who permitted the medical doctor to forward their contact details and to be contacted directly by the interviewer. The recruitment procedure varied slightly according to doctors' preferences for recruiting. However, all used the same inclusion criteria. Eligible for the study were men and women aged 18-70 who underwent at least one genetic counselling session for genetic testing for any cancer predisposition, were psychologically stable and not pregnant. Both cancer patients and people opting for presymptomatic genetic testing for cancer predisposition were included. The recruiting medical doctors controlled for these criteria before handing over the study information leaflet. Selection criteria were refined during the study to allow for theoretical saturation: to compensate for the higher percentage of women and those in favour of testing among participating at-risk individuals, we

1 See Appendix A-3 for the study information leaflet and the informed consent form.

specifically recruited male at-risk individuals and those who decided against testing. For the expert interviews, we recruited healthcare professionals providing genetic counselling.

3.3.3 Data collection

We collected all data between August 2017 and February 2019. The first author (BZ) held all interviews face to face at the university hospital or the participants' home, according to the participants' preferences. Using a semi-structured interview guide, we asked the at-risk individuals to talk about their experiences regarding genetic counselling and genetic testing. They also answered questions regarding other kinds of testing to assess their more general attitudes, such as predictive genetic testing for nonactionable genetic variants, and predictive genetic testing for diseases with environmental influence (such as diabetes) in the form of hypothetical vignettes. The experts were asked about themes and observations that were identified during the analysis of the patient interviews.[2]

3.3.4 Data analysis

All patient and expert interviews were held, recorded and transcribed by the first author. One interview was in French, and three in High-German. They were transcribed *ad verbatim*. The spoken language of the remaining 16 interviews was Swiss-German dialect. The first author translated them to High-German during transcription, and all transcripts were anonymised. Following grounded theory, we started our inductive analysis during data collection. We used MaxQDA 2018 (VERBI GmbH) for data analysis. We first analysed all interviews by open coding and discussed them. Reflection on interpretative patterns of the researchers, constant comparison of the content of the different interviews and identification of new concepts and aspects were central for this first analytical step. We built concepts and categories by grouping and connecting initial codes constantly. At the same time, we added abductive steps, asking questions to the data, collecting information on interesting findings in further interviews, and creating and testing hypotheses on our data. We then started connecting different concepts and categories, using the coding paradigm proposed by Strauss and Corbin (Strauss and Corbin, 1990). Finally, we performed several selective coding steps, where we critically examined and discussed hypotheses and questions raised during data analysis. In all steps, we wrote memos and discussed the analysis.

2 See Appendix A-1 and A-2 for the interview guides.

For this publication, we aimed for an English translation of illustrating quotations that reflects their meaning and context. Three researchers independently checked the quotations' translation for appropriateness. The two expert interviews took place before the selective coding and were used to inform and confirm the analysis. Since these two expert interviews did not add any new aspect to our analysis, we assumed theoretical saturation. At the end stage of the project, we sent the participating at-risk individuals our preliminary results, asking them to comment on how well they could identify themselves with them. Eleven out of eighteen (61%) replied. Two of them indicated a slightly different position than we would have anticipated from the interview, and we included their additional comments in our analysis.

3.4 Results

We interviewed eighteen at-risk individuals (Table 3-1) and two medical doctors specialised in genetics. Most at-risk individuals felt well informed and were happy with the quality and content of the genetic consultation. Criticism focused on clinical procedures and psychological components such as unexpected emotional distress while waiting for the test result. Most at-risk individuals had sustainably consolidated decisions regarding genetic testing and showed little uncertainty in this regard.

Table 3-1: Sample characteristics of interviewed counsellees (N=18).

Gender	Female	14 (78%)
	Male	4 (22%)
Age	20-29	1 (6%)
	30-39	5 (28%)
	40-49	3 (17%)
	50-59	5 (28%)
	60+	4 (22%)
Outcome	Mutation-positive test result	7 (39%)
	Mutation-negative test result	8 (44%)
	Decided against genetic testing	3 (17%)
Cancer predisposition that was tested for	Lynch syndrome	2 (11%)
	Hereditary breast/ovarian cancer	16 (89%)
Participants that had blood-related children		12 (67%)
Participants that were affected by cancer at the moment of genetic counselling		6 (33%)
Participants that had a known mutation in the family at the moment of genetic counselling		5 (28%)
Mean interview duration (min)		59 (27-101)

3.4.1 The theory

How at-risk individuals decided regarding genetic testing for cancer predispositions was a reflection of their general life philosophy. Those who preferred to control their life tended to have a positive attitude towards genetic testing. For them, knowledge created certainty and thus gave them a sense of control over their present and future lives. Hence, testing was important and powerful to them. Some of these individuals found it difficult to understand when others decided against testing (see Table 3-2 for quotation Q1). By contrast, other at-risk individuals preferred to leave their future life to fate. They felt more comfortable with living in the present rather than dealing with possible future scenarios and did not wish to gain certainty through genetic knowledge. They tended to reject genetic testing (Q2). We theorise that these two positions are the two extremes of what we term the control-fate-continuum. There might be numerous positions within this continuum and we do not claim that we can represent all possible positions. Instead, we provide seven examples that illustrate positions that we identified from our interview data (Box 3-1). We also identified psychosocial factors influencing the position on the control-fate-continuum, and thus the genetic testing decision-making process (Figure 3-1).

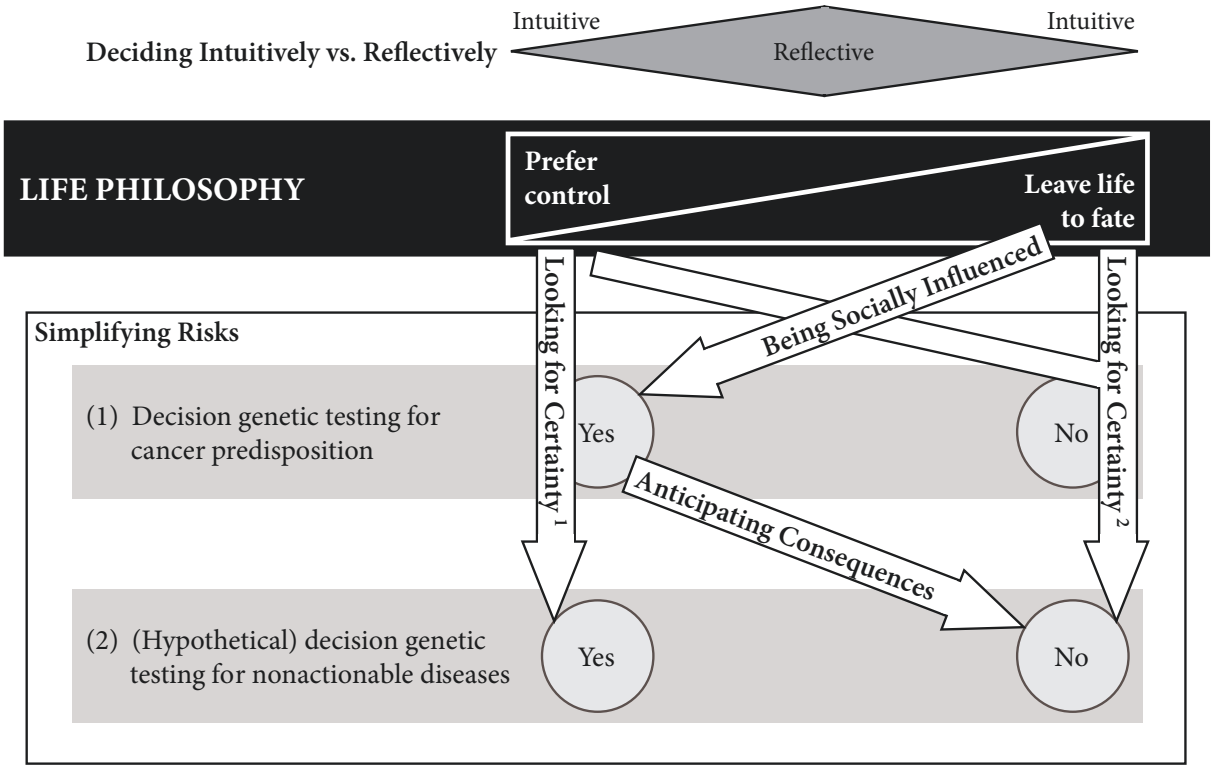
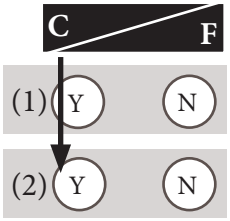


Figure 3-1: Model for decision-making regarding genetic testing for cancer predisposition and nonactionable diseases in relation to life philosophy and psychosocial factors.
[1] Perception that genetic testing reduces uncertainty; [2] Perception that genetic testing increases uncertainty.

Box 3-1: Examples of positions in the Control-Fate-Continuum. The numbers of the examples correspond to the numbers in Figure 3-1.

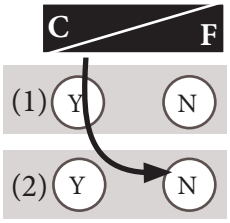
Example 1: Genetic testing provides me with a unique chance to take control of my life

Facts are very important to me. They give me a sense of certainty, a sense of control over my life. Therefore, genetic testing for inheritable cancer risk was self-evident for me. If I am at risk for a disease, I really want to know, I need to know, even if there were no preventive measures available. To me, knowledge is useful as such, even if there is no way to act. I often find it difficult to understand why others in my family do not want to be tested. I think this is irresponsible, especially when they have children. I also strongly believe in scientific progress. It provides me with a lot of hope to learn about recent advances, and I am optimistic that there will be much more progress within the next ten years.



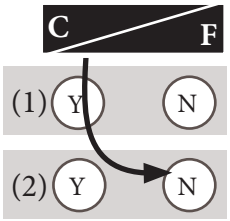
Example 2: Genetic testing is useful, but only if there is some preventive action I could take

Deciding for genetic testing for inheritable cancer risk has been easy for me. I know that cancer runs in my family, and because there are well-established preventive measures, such as screenings or surgeries, I think it is a logical consequence that I perform this test. It might also help my family members. By knowing my genetic cancer risk to be elevated, I am sure that I can prevent myself from getting sick as my mother or father did. However, I would not want to know my genetic fate if there is nothing I can do to prevent it. I do not see the benefits in that case; why should I want to know that? It would only haunt me and make me all nervous and anxious. I do not want a genetic test determining my life in such a way.



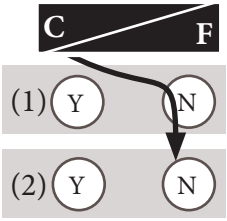
Example 3: I want genetic testing exclusively if there are immediate consequences for my health strategy

Even though I think genetic testing is useful and a necessity for my health, I will wait to get tested until preventive measures are relevant for me. Doing genetic testing too early, before preventive interventions are recommended, would only worry me unnecessarily. Likewise, genetic testing for nonactionable diseases is not an option for me. If there are no direct consequences, I would rather not want to know my genetic risk. However, if I would find out I had an increased risk for cancer, preventive surgery, if applicable, would be mandatory for me.



Example 4: A genetic test has to provide me with 100% certainty

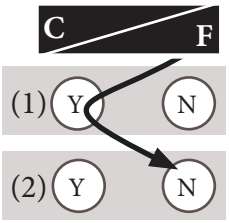
I generally think knowledge is useful and helpful for me to handle life, but a genetic test for inheritable cancer risk does not provide me with enough certainty. If the genetic test reveals that I am carrying a function-affecting genetic variant, there is still a good chance that I might not get sick at all. If the test result is negative, it does not mean that I will stay healthy, either. I thus refuse to be tested. If scientific progress led to better tests in the future, I would reconsider my decision and rethink it. Until then, I have enough preventive screenings to feel safe. I do not worry too much. If it gave me 100% certainty, I would even do a genetic test for a disease without preventive measures, because then I know, and feel in better control over my life.



Box 3-1 (continued).

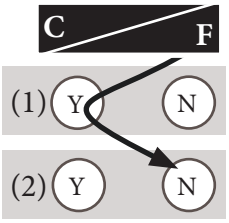
Example 5: I do genetic testing for my children, not for myself

Genetic testing is not for me. I know we have cancer running in our family, but I'd rather not know my exact genetic risk. However, I do it for my children, because they are young and still have the chance to prevent cancer. Thus, I intuitively decided to go for genetic testing. If they wish, I would even consider doing a test for unpreventable diseases, although I find this a difficult decision.



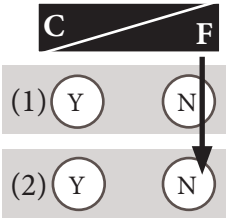
Example 6: I do genetic testing because others convinced me that it is useful for me

On my own initiative, I would not have done genetic testing for inheritable cancer risk. However, my health professionals and my relatives said that it would be useful and important for me to do it. That is mainly why I agreed to do testing. Having done testing does not bother me, I feel good with my decision. However, I would not undergo genetic testing for nonactionable diseases. I do not see the usefulness of this kind of testing; I think learning about a pathogenic genetic variant would bother me too much in that case.



Example 7: I do not want to know my genetic risk; I will leave my health-related future to fate

I know cancer is running in my family, but I do not want to know my genetic risk. I prefer leaving my future life to fate, to live in the present. It is like a gamble: the risk of losing is high, and I fear I could not handle it. Genetic testing cannot provide me with any certainty, and knowing about an elevated genetic risk would only make me miserable. At the same time, learning about a negative test result would not provide me with enough relief to take that risk.



Note: In order to illustrate the individuality of the positions, we describe the examples as subjective narratives, which are interpretative summaries of what our participants reported in the interviews. These are the words of the authors and do not correspond to quotes.

Table 3-2: Quotes supporting the results.
All names are pseudonyms. The quotes are referred to in the text by the codes in the left column.

Code	Quote
Q1	"If one hadn't done the test, one would only speculate, do I have it, do I not have it, and eh, well, in that case, that's when fear would actually be present. And now, it's just normal [...] and I, I think knowledge is always important." (Nick, 50+, healthy carrier)
Q2	"So now it's like a bit... now I can say, well it's fate now, maybe I will get cancer, maybe not. Ehm, I don't have it in my hands, it is as it is then. And if I tested myself, yes then I'd have a little, yes... I'd be a little more in charge again, although I perhaps couldn't change anything but... um, nevertheless, I knew it..." (Lea, 30+, untested)
Q3	"[...] I prefer to be informed, I prefer to know if there is something […]. Once I know that there might be something, once I'm told, do you want to know if there is a mutation on that gene, I could no longer say no I don't want to know." (Claire, affected non-carrier)
Q4	"Research in the genetic field will be massively improved in the coming years. Well, probably. [...] Maybe there will be much better answers in 5, 10 years if you do such a genetic examination because you can take in many more factors, many more influencing factors... And then there will probably be a better answer. And as soon as there is a better answer, I will probably think about it [genetic testing] again." (Doris, 40+, untested)
Q5	"I won't do the genetic test. [Because, after the genetic consultation], I thought again about the different aspects of the conversation. What are the results I could really get, specifically, from this genetic test. And namely, results with 100% certainty, so I thought about the certainty the test could provide me with. Or does it give me more uncertainty […] Would it really give me watertight answers. There might be other influencing factors, too. So regarding breast cancer, I couldn't get it off my mind after all, even if I had a negative genetic test result." (Doris, 40+, untested)

Table 3-2 (continued).

Code	Quote
Q6	"I then got counselling and listened to it all and thought about it, but then I decided against testing at that point. Because of my age. I was still younger than 40 at that time, and I just consciously wanted to take the time, wait until 40 and then I can bear the consequences of the test, and until then I just don't want to know. So I've been really, I've received good counselling about what are the consequences, what are the advantages, the disadvantages, of this test." (Daria, 40+, non-carrier)
Q7	"My main consideration […] is how the awareness [of knowing a genetic disease risk] would affect my decisions in life, actually. […] If having the mutation affects my decisions because of a certain sense of responsibility or something, which I would not do of my own free will." (Aaron, 20+, carrier)
Q8	"Had I been a carrier, I'd have had the surgeries. [...] That's what I'd have done. If there is no such consequence for people I think them doing the test isn't so important, but for me, that would have been an obvious consequence." (Silvia, 30+, non-carrier)
Q9	"So I think they [her two daughters] got tested simply because I said so, I think they didn't know what it meant at the beginning. Because everything was going so fast... I think I overwhelmed them a little bit, to be honest. Well, I don't regret that, I think it's important. I also think it's important that they just watch it afterwards. And are made aware of it." (Hanna, 50+, affected carrier)
Q10	"I was talking to my mother during the testing process, and otherwise I didn't really need anybody… I knew what I did, I knew what I was getting into, I knew… what the consequences would be, and so on. Yes." (Marissa, 50+, healthy)
Q11	"Well, the human psychological aspect, [...] I didn't think that it would have such an impact, I wouldn't have thought of that, suddenly it came. Over time, when I started to think a little bit." (Erich, 60+, non-carrier)
Q12	"I pay attention [to protecting my genetic data], yes, but it's not like it has greatly influenced my decision to do genetic testing." (Aaron, 20+, carrier)

Table 3-2 (continued).

Code	Quote
Q13	"At Cancer Aid, they told me that I would have difficulties insuring myself privately or semi-privately, that health insurance would refuse me because of the carrier test result. Yes, and that was in my head, and then I thought well, I try it anyway, and then, after the genetic consultation it was actually clear to me how to do it. I also asked for the legal text, that was sent to me by the geneticist… And then I called the health insurance company and said that I would like to change my insurance, that I had this problem. Eh, saying that there was the no-discrimination act, sending them all the information. It took about three weeks, and then they agreed, without reservation, without anything." (Nick, 50+, healthy carrier)
Q14	"But getting this information when you can't do anything about it anyway. I think I… I don't want this. Even though I agreed [to genetic testing]. That only occurred to me later. Interviewer: And why did you accept [genetic testing]? Participant: I don't know! I usually decide spontaneously and think about it afterwards [...] Interviewer: And did you consider stopping the process of testing? Participant: No. At home, my wife and daughter have really (laughs) taken sides and said, listen, it is not just for you. It's for your daughter as well." (Erich, 60+, non-carrier)
Q15	"Em, but... [if I hadn't done the genetic test] it would have been a bit like, I couldn't have taken part in certain things because... If I hadn't done it, I wouldn't have known, am I positive or not? [...] That's why it's actually, yes... I've actually wanted to know... uh... yes, how is it for me now, so that I simply know, am I in their group or am I, am I outside." (Eva, 40+, healthy carrier)
Q16	"I for myself, if my daughter hadn't come because she wanted to know that, I would not have done counselling in that sense. Well, that's, really, first and foremost for my daughters." (Emily, 60+, non-carrier)
Q17	"It wouldn't have left me alone to be told it [genetic testing] would be recommended [...] Then I'd preferred not to be told at all, see, now that is coming up again! If they wouldn't have told me anything, just said, well, we have taken out your cancer now, [...] now do these check-ups every five years - have a nice day. That would have been fine for me, yes, that would have been fine with me that alternative that they don't tell me about genetic testing..." (Erich, 60+, non-carrier)

Table 3-2 (continued).

Code	Quote
Q18	"For me, it's simply a derivation of fate - not fate, but human… needs, so if anything happens and... one lives shorter, or someone else could have protected himself and could not do it, that is careless towards their environment, in my opinion. If my sister died early, and her child lost her mother early, that would not be so cool." (Rolf, 50+, carrier)
Q19	"Well, for one [sister], it was simply because of the economic expense. If she goes to the gynaecologist twice a year and hasn't clarified the source of the problem beforehand then she does something useless. This is unnecessary and simply burdens the economy." (Rolf, 50+, carrier)
Q20	"Yeah, well, if the doctor tells me anything, I'll do it anyway, won't I?" (Gloria, 60+)
Q21	"The geneticist knew that I came from the medical field, and then it often happens, you notice that people say, ah you already know that anyway, but he still did it very professionally and as if I was a layperson, I liked that." (Silvia, 30+, non-carrier)
Q22	"Well, I'd be cautious... to discuss something like this with others. Because [...] I would get into a defensive position. [...] To defend that would not be so pleasant. Yes, I decided that way, that's the way it is, and I wouldn't want to defend it now." (Doris, 40+, untested)
Q23	"Well, in the first moment I was completely against it, I didn't even want to know anything about it, I didn't even want to think about it. And only after a certain amount of time I remembered it again, when the gynaecologist asked me about it, and then it went around in my head a bit. I also weighed up the advantages and disadvantages, what would happen if I would, or what would it mean, what would be the consequences, and then, yes, no, I don't want to... And what would be the reasons why I might do it after all… It has been a bit of a back and forth, but the tendency has always been more towards I don't want to know it." (Lea, 30+, untested)
Q24	"Yes, I think I already made the decision before the counselling session... And, I don't know what they could have told me that would have changed my opinion." (Lea, 30+, untested)

Table 3-2 (continued).

Code	Quote
Q25	"Well, in the end, it doesn't matter what the percentages are... Either I get it or I don't. Somehow it is always 50:50." (Daria, 40+, non-carrier)
Q26	"With a genetic cancer predisposition, I feel like, it's fate somehow, you just wait for the moment it strikes, and no matter what I do… it is predetermined. Yes." (Lea, 30+, untested)
Q27	"Internally, I anyway expected that it was just a strong family history. If I had feared a genetic cause, I would probably have reacted earlier." Interviewer: "So you didn't expect the detection of a genetic cause…" Participant: "Yeah, or… it was more towards the direction that they would not find anything. But that doesn't necessarily mean that that is the case, some perceptions are just repressed easily, right?" (Emily, 60+, non-carrier)
Q28	"Rationally, the probability of a finding was not very high […] I saw it black on white when going through the pedigree [with the counselling physician]. I'd say I have a big family […] and there are some cases of cancer that alerted me at first, but the majority is healthy. And that is why… Yes, the probability [to detect a function-affecting genetic variant] was reduced due to the many healthy people in my family." (Doris, 40+, untested)
Q29	"But, so for me, it was clear that there had to be something genetic because... Well, we had to find a cause. […] Well, once I got these results... I could hardly believe it, I was eh, yes, so eh, well, there were no mutations, I, I was really... But I immediately, immediately it came back to me, but, but... what is it then? So there's another one, there must be something else there!" (Claire, affected non-carrier)

Note: "Carrier" means being positively tested for a pathogenic variant; "non-carrier" refers to a test result of non-pathogenic variant or a variant of uncertain significance. "Untested" means the person decided against genetic testing. The participants referred to a pathogenic genetic variant when using the term "mutation".

3.4.2 Psychosocial factors influencing decision making

1. Looking for Certainty

At-risk individuals were looking for certainty when making their decision of whether to take genetic testing. The outcome of the decision depended on their life philosophy. On the one hand, for those preferring control, genetic testing reduced uncertainty because it provided facts and the possibility to make informed life decisions (example 1, Q3). They were sure knowing their genetic risk was beneficial and expressed confidence in scientific progress. On the other hand, genetic testing increased uncertainty for those leaving life to fate (examples 4 and 7). Because the genetic test result only provided disease probabilities, it did not provide certainty. Genetic testing was not considered meaningful as long as scientific knowledge is incomplete (Q4). Moreover, some individuals explained that life was full of uncertainties and risks; therefore, they preferred to live in the present rather than in the unforeseeable future (Q5).

2. Anticipating Consequences

Some at-risk individuals strongly focussed on the consequences of testing. They considered only genetic testing for cancer predisposition beneficial and would not take genetic testing for non-actionable diseases (examples 2 and 3). Therefore, their position on the control-fate-continuum is somewhat to the right of those who prefer control in any case (example 1).

Having children and being affected by cancer tended to strengthen at-risk individuals' focus on consequences. Some cancer patients perceived the testing procedure as intimidating and wanted to postpone it until after cancer treatment because they did not see immediate medical consequences. Moreover, young individuals perceived genetic testing as less urgent, and some postponed genetic testing until they reached the age when their healthcare professionals recommended preventive measures (Q6). Finally, some participant stated that they would refrain from testing if the consequences would determine their life too much (example 3, Q7).

Four levels of consequences that at-risk individuals anticipated were identified: medical, familial, psychological and discriminatory consequences. First, some individuals based their decision on the availability of medical consequences (examples 2 and 3). These individuals proactively thought about the integration of the test result and its consequences in their life and had already decided upfront on what preventive measures they would take if they were risk variant carriers (Q8). While some perceived frequent screening as a sufficient and satisfying measure, others needed the option of risk-reducing preventive surgery or medication to take the test. This might have helped them rationalise and structure their decision-making process, thus reducing uncertainty.

Second, some at-risk individuals strongly anticipated consequences for their family and considered this the main reason for genetic testing (example 5). While some of them accepted that some relatives might exercise their right not to now, others did not reflect on this aspect and motivated their relatives to get genetic testing without further consideration (Q9).

Third, most at-risk individuals anticipated adverse psychological reactions, as were discussed in genetic counselling. This anticipation helped them overcome feelings of anxiety and uncertainty (Q10). Still, one participant failed to anticipate psychological consequences and started to brood afterwards (Q11). Fourth, discriminatory consequences were anticipated by some few individuals. They were careful about whom to tell about their test result because they were aware of possible discrimination by employers or private insurance (in Switzerland basic health insurance is obligatory and the insurance companies can refuse no patient; however, this is not the case for private and semi-private insurance (Swiss Federal Council, 2004)). Still, participants did not refuse genetic testing for cancer predispositions out of privacy concerns (Q12). One interviewee even described a positive experience with his health insurance company (Q13).

3. Being Socially Influenced

A sense of duty and social influences from family, healthcare professionals and society left some at-risk individuals with no real alternative but to test, even if it was against their life philosophy (examples 5 and 6, Q14). First, family considerations had an influence (example 5): Some at-risk individuals felt excluded from their families without genetic testing (Q15). Others felt a duty towards their offspring and other family members despite their sceptical attitude towards genetic testing (Q16). Because of this feeling of obligation, one participant even wished he had never heard about the possibility of testing (Q17). Some at-risk individuals perceived not doing genetic testing as reckless, because to them, genetic testing implied taking responsibility for themselves and their social environment (Q18). Some also considered that their genetic test would reduce healthcare costs, which made them advocate the test to family members, too (Q19).

Second, despite the non-directiveness of genetic counselling, some affected individuals followed the advice of their counselling healthcare professional to be tested (example 6). That happened particularly in the context of a newly diagnosed cancer, where genetic testing might provide therapy-relevant information. The extent to which medical doctors influenced at-risk individuals in their decision-making process depended on their relationships as well as on the at-risk individuals' general attitudes towards doctors (Q20). One participant stressed that it was important to her to receive genetic counselling that was not influenced by her prior knowledge on the topic (Q21).

Third, we observed various societal influences. For instance, those who had decided against testing did not like to broadcast this decision because they did not want to justify their decision (Q22). By doing a genetic test, "one has done everything one can" (Doris, healthy, untested), which they felt was easier for others to understand. Moreover, online communities, such as Facebook groups or forums, provided another form of external influence. They had variable effects on at-risk individuals, some felt encouraged, others rather scared when reading how carriers dealt with their increased cancer risk. Some were emotionally neutral and used it as an additional information source in their decision-making process.

4. Deciding Intuitively vs Reflectively

Individuals at both extremes of the control-fate-continuum tended to decide quickly and intuitively: Testing was naturally good for those who gained control through it (example 1), and those who preferred to leave their future life to fate rejected genetic testing equally intuitively (example 7). Additionally, those who did testing mainly for their family or on their doctor's advice intuitively outsourced their decision to these third parties (examples 5-6; see also "Being Socially Influenced"). By contrast, those in the middle of the continuum tended to reflect upon genetic testing intensively, weighing pros and cons. Their decision depended on the circumstances of testing (examples 3-4) One participant first rejected genetic testing intuitively but made a more reflected decision later on (Q23).

Genetic counselling was vital for a reflected decision-making process (Q6). However, some individuals had already made their decision upfront and thus did not include the content of the genetic consultation in their decision (Q24). They went to the genetic consultation because it was required and to get detailed information about the testing procedure as well as the implications and consequences of the test.

5. Simplifying Risks

Because of the complexity of genetic risks, simplification was crucial for an effective decision-making process. While all at-risk individuals might simplified risks, they did so to different extents. Some individuals simplified risks and probabilities by breaking them down into a black-and-white picture, thus reinterpreting uncertainty (Q25). Consequently, not finding a function-affecting genetic variant sometimes led to the misconception that the risk of cancer was zero. In contrast, one individual felt that being a carrier of a function-affecting genetic variant would inevitably lead to cancer, even if in reality, the penetrance is not 100% (Q26). She thus rejected genetic testing due to fear.

Another simplification concerned the anticipation of the test result. Some individuals made an intuitive estimation (Q27); others received a risk calculation during genetic counselling (Q28). Those who thought their risk to be a carrier was high tended to decide clearly and intuitively – either in favour of or against genetic testing (Q29). Individuals thinking they were probably not carrying a function-affecting genetic variant tended to scrutinise the meaningfulness of genetic testing and tended to reflect more on their decision (see also *"Deciding Intuitively vs Reflectively"*).

3.5 Discussion

Our findings illustrate how people undergoing genetic counselling for cancer predisposition in Switzerland perceive the decision-making process for genetic testing. The study provides new findings of decision making by individuals at risk for hereditary cancer in continental Europe. Our results help healthcare professionals and at-risk individuals to structure the decision-making process. We propose that at-risk individuals position themselves on the control-fate-continuum, which derives of their general life philosophy. Medical doctors and genetic counsellors should understand and support individuals in this process. The position on the control-fate-continuum is an essential factor for genetic testing decision-making. Those who prefer a control-approach to life might benefit from genetic testing, even if the genetic variant that is tested for is not actionable (Wiggins et al. 1992). However, they might also overestimate genetic testing outcomes and feel determined to do genetic testing once it is offered. In contrast, those leaving their future life to fate might refuse genetic testing decisively and be better off without it, even if preventive measures could reduce their disease risk. Several psychosocial factors influence this underlying aspect of life philosophy and thus the decision-making process.

Our findings are in line with other studies of decision-making for genetic testing for cancer predisposition, in particular to the psychosocial factors identified. First, it is established that people are *looking for certainty* when they do genetic testing (Vos et al. 2013). Our study provides unique new evidence that the nature and level of certainty they attribute to genetic testing largely depends on their life philosophy. Second, people tend to anticipate consequences more when receiving genetic counselling. In settings without genetic counselling, many people do not anticipate the consequences of genetic testing (Roberts et al. 2017).

Moreover, *simplifying risks* due to misinterpretation, over- or underestimation is part of human psychology (Kahneman and Tversky 2009) and is thus commonly studied in the context of genetic testing (Kelly et al. 2005; Cicero et al. 2017; Goltz, Bergman, and Goodson 2016).

Additionally, while *intuitive decision-makers* need genetic counselling mainly for gathering more facts or because it is mandatory to get the genetic test, *reflective decision-makers* might need more psychosocial support and more time to reflect the pros and cons of genetic testing (Howard, A. F. et al. 2011; McAllister 2002). The *social influence* on decision-making leads back to the question of whether individuals have a real choice regarding genetic testing. Previous studies also conclude that some people agree to genetic testing out of a feeling of responsibility towards their relatives (Hallowell et al. 2003; Etchegary et al. 2009). While our theory represents this position as well, we suggest that individuals' life philosophy dominates this motive at the two extremes of the control-fate-continuum.

Our theory also accounts for the reasons of those who decline genetic testing. Previous studies showed that, even though some decliners do so unreflectively out of fear or lack of knowledge, many refuse genetic testing consciously because they do not see benefits for themselves (Keogh et al. 2017; White, V. et al. 2018; Peters et al. 2011). We suggest that genetic testing contradicts the life philosophy of some individuals. It is thus essential to counsel those deciding for genetic testing regarding how to inform these family members about the intention of being tested and test results. At-risk individuals have expressed difficulties with uninterested or resistant family members in previous studies (Forrest et al. 2003; Daly et al. 2016; Chopra and Kelly 2017), and our theory helps to explain why: different life philosophies can lead to difficulties in communication because those in need of control have difficulties in understanding why others might not want to learn about their genetic risk.

The scope of the control-fate-continuum theory covers genetic testing in both healthy and affected individuals provided that genetic testing has predictive components for some disease a person has not developed (yet). For instance, a breast cancer patient with a pathogenic BRCA variant still has an increased risk of ovarian cancer or bilateral breast cancer.

To our knowledge, this is the first study regarding the genetic testing decision-making process from the German-speaking region, which provides a different cultural setting from that of English-speaking countries, which are predominantly studied. Our interviewees were rarely worried about discrimination and did not take this variable into account in their decision-making process, as opposed to previous studies in other regions (Wauters and van Hoyweghen 2016). However, we acknowledge a possible bias in the sense that those worried regarding discrimination might have refrained from participating in this study. More research is needed to explore this potential cultural difference further.

3.5.1 Clinical implications

Our findings have implications for genetic counselling. It might be useful to discuss individuals' life philosophy and highlight that these might differ from those of their family members. Our framework can thus be used to support decision-making, anticipate potential conflicts within families and, potentially, helps to understand the reasoning of those declining genetic testing. The finding that some make their decisions intuitively while others need more support for thorough reflection indicates that tailoring genetic counselling resources to actual information needs might be useful. This becomes especially relevant in the context of limited counselling resources and increasing demand for genetic testing. However, more investigations, including quantitative studies, are necessary to assess the appropriateness and consequences of such prioritisation regarding autonomous decision-making.

3.5.2 Limitations

This is a qualitative study with eighteen at-risk individuals and two geneticists, and while this is a suitable number to get meaningful qualitative results (Hennink, Kaiser, and Marconi 2017), we propose using our theory to build hypotheses and to test them in more extensive studies. Despite actively searching for 18 months, we could only recruit three genetic testing decliners. Thus, we probably did not reach theoretical saturation for this subgroup. For ethical reasons, at-risk individuals had to contact us proactively in most cases, which gives a bias towards proactive and communicative people interested in genetic testing. Another possible bias results from the recruitment by the counselling doctors, as they might have preferred counsellees with characteristics other than the inclusion criteria. Moreover, qualitative analyses require interpretation, but we minimised personal interpretation bias by frequent team discussions, continuous personal reflection, constant comparison and other methods suggested for the grounded theory approach (Strauss and Corbin 1990).

3.5.3 Conclusion

Our study illustrates the enormous interpersonal variability of the genetic testing decision-making process. Professional counselling is a way to account for this variability and ensure proper support in the complex decision-making process. The control-fate-continuum can help medical geneticists and genetic counsellors to consider this interpersonal variability and provide counselling that respects and anticipates people's wishes and needs so that counsellees can make informed choices. It also helps their counselling in terms of information dissemination to family members by learning about potential differences in life philosophy, interest, and decision-making

regarding genetic testing. Moreover, our findings help future research studies for hypothesis testing and result quantification. Because genetic testing is becoming more common, stratifying people regarding their information needs and decision-making strategy helps to rationalise genetic counselling in the future, by saving resources and adapting healthcare services to the needs of those at risk for hereditary cancer.

3.6 References

Annells, Merilyn. 1996. "Grounded Theory Method: Philosophical Perspectives, Paradigm of Inquiry, and Postmodernism." Qual Health Res 6 (3): 379–93.

Battistuzzi, Linda, Marzena Franiuk, Nadine A. Kasparian, Nadia Rania, Laura Migliorini, and Liliana Varesco. 2019. "A Qualitative Study on Decision-Making About BRCA1/2 Testing in Italian Women." Eur J Cancer Care 28 (5): e13083. doi:10.1111/ecc.13083.

Binetti, Giuliano, Luisa Benussi, J. S. Roberts, Aldo Villa, Patrizio Pasqualetti, Ching-Fan Sheu, Lara Gigola et al. 2006. "Areas of Intervention for Genetic Counselling of Dementia: Cross-Cultural Comparison Between Italians and Americans." Patient Educ Couns 64 (1-3): 285–93. doi:10.1016/j.pec.2006.03.008.

Castilla, Lucio H., Fergus J. Couch, Michael R. Erdos, Kent F. Hoskins, Kathy Calzone, Judy E. Garber, Jeff Boyd, Matthew B. Lubin, Michelle L. Deshano, and Lawrence C. Brody. 1994. "Mutations in the BRCA1 Gene in Families with Early-Onset Breast and Ovarian Cancer." Nat Genet 8 (4): 387–91.

Charmaz, Kathy. 2006. Constructing Grounded Theory: A Practical Guide Through Qualitative Analysis. Introducing Qualitative Methods Series. London: SAGE Publications.

Chopra, Ishveen, and Kimberly M. Kelly. 2017. "Cancer Risk Information Sharing: The Experience of Individuals Receiving Genetic Counseling for BRCA1/2 Mutations." J Health Commun 22 (2): 143–52. doi:10.1080/10810730.2016.1258743.

Cicero, Giuseppe, Rosella de Luca, Patrizia Dorangricchia, Giuseppe Lo Coco, Cinzia Guarnaccia, Daniele Fanale, Valentina Calò, and Antonio Russo. 2017. "Risk Perception and Psychological Distress in Genetic Counselling for Hereditary Breast And/or Ovarian Cancer." J Genet Couns 26 (5): 999–1007. doi:10.1007/s10897-017-0072-0.

Claes, Erna, Lieve Denayer, Gerry Evers-Kiebooms, Andrea Boogaerts, and Eric Legius. 2004. "Predictive Testing for Hereditary Non-Polyposis Colorectal Cancer: Motivation, Illness Representations and Short-Term Psychological Impact." Patient Educ Couns 55 (2): 265–74. doi:10.1016/j.pec.2003.11.002.

Daly, Mary B., Susan Montgomery, Ruth Bingler, and Karen Ruth. 2016. "Communicating Genetic Test Results Within the Family: Is It Lost in Translation? A Survey of Relatives in the Randomized Six-Step Study." Fam Cancer 15 (4): 697–706. doi:10.1007/s10689-016-9889-1.

Etchegary, Holly, Fiona Miller, Sonya deLaat, Brenda J. Wilson, June C. Carroll, and Mario Cappelli. 2009. "Decision-Making About Inherited Cancer Risk: Exploring Dimensions of Genetic Responsibility." J Genet Couns 18 (3): 252–64. doi:10.1007/s10897-009-9218-z.

Forrest, Karen, Sheila A. Simpson, Brenda J. Wilson, Edwin R. van Teijlingen, Lorna McKee, Neva Haites, and Eric Matthews. 2003. "To Tell or Not to Tell: Barriers and Facilitators in Family Communication About Genetic Risk." Clin Genet 64 (4): 317–26. doi:10.1034/j.1399-0004.2003.00142.x.

Foster, Claire, Maggie Watson, Clare Moynihan, Audrey Ardern-Jones, and Rosalind Eeles. 2002. "Genetic Testing for Breast and Ovarian Cancer Predisposition: Cancer Burden and Responsibility." J Health Psychol 7 (4): 469–84. doi:10.1177/1359105302007004627.

Godino, Lea, Leigh Jackson, Daniela Turchetti, Catherine Hennessy, and Heather Skirton. 2018. "Decision Making and Experiences of Young Adults Undergoing Presymptomatic Genetic Testing for Familial Cancer: A Longitudinal Grounded Theory Study." Eur J Hum Genet 26:44–53. doi:10.1038/s41431-017-0030-1.

Goltz, Heather H., Margo Bergman, and Patricia Goodson. 2016. "Explanatory Models of Genetics and Genetic Risk Among a Selected Group of Students." Front Public Health 4:111. doi:10.3389/fpubh.2016.00111.

Hallowell, Nina, Audrey Ardern-Jones, Ros Eeles, Claire Foster, Anneke Lucassen, Clare Moynihan, and Maggie Watson. 2005. "Communication About Genetic Testing in Families of Male BRCA1/2 Carriers and Non-Carriers: Patterns, Priorities and Problems." Clin Genet 67 (6): 492–502. doi:10.1111/j.1399-0004.2005.00443.x.

Hallowell, Nina, Claire Foster, Ros Eeles, Audrey Ardern-Jones, V. Murday, and Maggie Watson. 2003. "Balancing Autonomy and Responsibility: The Ethics of Generating and Disclosing Genetic Information." J Med Ethics 29 (2): 74-9; discussion 80-3. doi:10.1136/jme.29.2.74.

Harper, Joyce, Joep Geraedts, Pascal Borry, Martina C. Cornel, Wybo J. Dondorp, Luca Gianaroli, Gary Harton et al. 2014. "Current Issues in Medically Assisted Reproduction and Genetics in Europe: Research, Clinical Practice, Ethics, Legal Issues and Policy." Hum Reprod 29 (8): 1603–9. doi:10.1093/humrep/deu130.

Hennink, Monique M., Bonnie N. Kaiser, and Vincent C. Marconi. 2017. "Code Saturation Versus Meaning Saturation: How Many Interviews Are Enough?" Qual Health Res 27 (4): 591–608. doi:10.1177/1049732316665344.

Hoffman, Richard M., Carmen L. Lewis, Michael P. Pignone, Mick P. Couper, Michael J. Barry, Joann G. Elmore, Carrie A. Levin, John van Hoewyk, and Brian J. Zikmund-Fisher. 2010. "Decision-Making Processes for Breast, Colorectal, and Prostate Cancer Screening: The DECISIONS Survey." Med Decis Making 30 (5 Suppl): 53S-64S. doi:10.1177/0272989X10378701.

Howard, A. Fuchsia, Lynda G. Balneaves, Joan L. Bottorff, and Patricia Rodney. 2011. "Preserving the Self: The Process of Decision Making About Hereditary Breast Cancer and Ovarian Cancer Risk Reduction." Qual Health Res 21 (4): 502–19. doi:10.1177/1049732310387798.

Kahneman, Daniel, and Amos Tversky. 2009. "Prospect Theory: An Analysis of Decision Under Risk." In Judgement and Decision Making: Volume 2, edited by Nick Chater. 4 vols, 152–79. London: SAGE Publications.

Kalokairinou, Louiza, Heidi C. Howard, Santa Slokenberga, Eva Fisher, Magdalena Flatscher-Thöni, Mette Hartlev, Rachel E. van Hellemondt et al. 2018. "Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape." J Community Genet 9 (2): 117–32. doi:10.1007/s12687-017-0344-2.

Kanga-Parabia, Anaita, Clara Gaff, Louisa Flander, Mark Jenkins, and Louise A. Keogh. 2018. "Discussions About Predictive Genetic Testing for Lynch Syndrome: The Role of Health Professionals and Families in Decisions to Decline." Fam Cancer 17 (4): 547–55. doi:10.1007/s10689-018-0078-2.

Kelly, Kimberly M., Howard Leventhal, Michael Andrykowski, Deborah Toppmeyer, Judy Much, James Dermody, Monica Marvin, Jill Baran, and Marvin Schwalb. 2005. "Using the Common Sense Model to Understand Perceived Cancer Risk in Individuals Testing for BRCA1/2 Mutations." Psycho-oncology 14 (1): 34–48. doi:10.1002/pon.805.

Keogh, Louise A., Heather Niven, Alison Rutstein, Louisa Flander, Clara Gaff, and Mark Jenkins. 2017. "Choosing Not to Undergo Predictive Genetic Testing for Hereditary Colorectal Cancer Syndromes: Expanding Our Understanding of Decliners and Declining." J Behav Med 40 (4): 583–94. doi:10.1007/s10865-016-9820-0.

McAllister, Marion. 2002. "Predictive Genetic Testing and Beyond: A Theory of Engagement." J Health Psychol 7 (5): 491–508. doi:10.1177/1359105302007005628.

Miki, Yoshio, Jeff Swensen, Donna Shattuck-Eidens, P. A. Futreal, Keith Harshman, Sean Tavtigian, Qingyun Liu, Charles Cochran, L. M. Bennett, and Wei Ding. 1994. "A Strong Candidate for the Breast and Ovarian Cancer Susceptibility Gene BRCA1." Science 266 (5182): 66–71. doi:10.1126/science.7545954.

Peters, June, Regina Kenen, Lindsey M. Hoskins, Laura M. Koehly, Barry Graubard, Jennifer T. Loud, and Mark H. Greene. 2011. "Unpacking the Blockers: Understanding Perceptions and Social Constraints of Health Communication in Hereditary Breast Ovarian Cancer (HBOC) Susceptibility Families." J Genet Couns 20 (5): 450–64. doi:10.1007/s10897-011-9370-0.

Press, Nancy, Jennifer R. Fishman, and Barbara A. Koenig. 2000. "Collective Fear, Individualized Risk: The Social and Cultural Context of Genetic Testing for Breast Cancer." Nurs Ethics 7 (3): 237–49. doi:10.1177/096973300000700306.

Roberts, J. S., Michele C. Gornick, Deanna A. Carere, Wendy R. Uhlmann, Mack T. Ruffin, and Robert C. Green. 2017. "Direct-to-Consumer Genetic Testing: User Motivations, Decision Making, and Perceived Utility of Results." Public Health Genom 20 (1): 36–45. doi:10.1159/000455006.

Schlich-Bakker, Kathryn J., Herman F. J. ten Kroode, Carla C. Wárlám-Rodenhuis, Jan van den Bout, and Margreet G. E. M. Ausems. 2007. "Barriers to Participating in Genetic Counseling and BRCA Testing During Primary Treatment for Breast Cancer." Genet Med 9 (11): 766–77. doi:10.1097/GIM.0b013e318159a318.

Skirton, Heather, Lesley Goldsmith, Leigh Jackson, and Aad Tibben. 2013. "Quality in Genetic Counselling for Presymptomatic Testing — Clinical Guidelines for Practice Across the Range of Genetic Conditions." Eur J Hum Genet 21 (3): 256–60. doi:10.1038/ejhg.2012.174.

Soini, Sirpa. 2012. "Genetic Testing Legislation in Western Europe-a Fluctuating Regulatory Target." J Community Genet 3 (2): 143–53. doi:10.1007/s12687-012-0078-0.

Strauss, Anselm, and Juliet M. Corbin. 1990. Basics of Qualitative Research: Grounded Theory Procedures and Techniques. Newbury Park, Calif: SAGE Publications.

Swiss Federal Council. 2004. "CC 810.12 Federal Act of 8 October 2004 on Human Genetic Testing (HGTA)." Accessed January 23, 2020. https://www.admin.ch/opc/en/classified-compilation/20011087/index.html.

Vos, Joël, Fred H. Menko, Jan C. Oosterwijk, Christi van Asperen, Anne M. Stiggelbout, and Aad Tibben. 2013. "Genetic Counseling Does Not Fulfill the Counselees' Need for Certainty in Hereditary Breast/ovarian Cancer Families: An Explorative Assessment." Psycho-oncology 22 (5): 1167–76. doi:10.1002/pon.3125.

Wauters, Annet, and Ine van Hoyweghen. 2016. "Global Trends on Fears and Concerns of Genetic Discrimination: A Systematic Literature Review." J Hum Genet 61 (4): 275–82. doi:10.1038/jhg.2015.151.

White, V. B., Kendall K. Walsh, Kimberly S. Foss, Lisa Amacker-North, Stacy Lenarcic, Lindsay McNeely, and Richard L. White. 2018. "Genetic Testing for Hereditary Breast Cancer: The Decision to Decline." Am Surg 84 (1): 154–60. doi:10.1200/jco.2015.33.15_suppl.1552.

Wiggins, Sandi, Patti Whyte, Marlene Huggins, Shelin Adam, Jane Theilmann, Maurice Bloch, Samuel B. Sheps, Martin T. Schechter, and Michael R. Hayden. 1992. "The Psychological Consequences of Predictive Testing for Huntington's Disease. Canadian Collaborative Study of Predictive Testing." N Engl J Med 327 (20): 1401–5. doi:10.1056/NEJM199211123272001.

Zimmermann, Bettina M., Noah Aebi, Steffen Kolb, David Shaw, and Bernice S. Elger. 2019. "Content, Evaluations and Influences in Newspaper Coverage of Predictive Genetic Testing: A Comparative Media Content Analysis from the United Kingdom and Switzerland." Public Underst Sci 28 (3): 256-274. doi:10.1177/0963662518816014.

Chapter 4: "Is it wise to know everything?" Motivations to and barriers of information-seeking behaviour regarding genetic testing

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4.1 Abstract

Objective: This study aims to illustrate what at-risk individuals consider motivators of and barriers to information-seeking before and after genetic testing for cancer predisposition and to assess what they think the public should know about genetics.

Methods: Semi-structured interviews with people seeking genetic counselling in Switzerland were analyzed qualitatively using thematic analysis. Wilson's model of information behaviour was the theoretical framework.

Results: We identified several factors portraying motivators and barriers for seeking information: *Life philosophy, Decision-making style, Previous knowledge, Trust in physicians, Family position, Emotions, Risk perception, Age, Disease, Feeling of responsibility,* and *Poor physician knowledge of genetics.*

Conclusion: While various existing interdisciplinary theories and models verify these factors, this empirical approach helps healthcare professionals to understand their patients' behaviour and wishes concerning information more concretely. Some participants also stated that, based on their own experience, giving too much information to the public is neither necessary nor beneficial for individuals.

Practice implications: The findings help healthcare professionals involved in pre- or post-test healthcare to understand motivators of and barriers to information seeking and support those affected according to their individual needs. Professionals involved in health communication should consider providing less, but more concise, information to the public.

4.2 Introduction

The genetic testing decision-making process for cancer predispositions is inherently complex. One level of complexity is the interpretation of a genetic test result and the notion of risk. Not being a carrier of a pathogenic variant does not mean there is no risk of cancer because environmental factors could cause cancer as well. Moreover, sometimes the test reveals a variant of unknown significance, which means that whether the detected genetic variant is pathogenic is unknown (Mahon 2015). Being a carrier, by contrast, does not always lead to cancer because the penetrance is not 100% (Evans, J., Skrzynia, and Burke 2001). Human beings have difficulties in rationally assessing risks appropriately (Kusev et al. 2019). Psychosocial challenges add to this difficulty (Winchester and Hodgson 2006): a test result also affects blood-related family members, knowing the genetic risk might cause depression and anxiety, and there is a potential risk

of discrimination. Information dissemination to family members can be a burden for carriers because of the complexity of information and established family communication patterns (Dancyger et al. 2011). Furthermore, choices and decisions regarding measures to prevent cancer in carriers can be complicated and emotionally laden (Evans, J., Skrzynia, and Burke 2001).

4.2.1 Informing at-risk individuals through genetic counselling

Because of this multi-layered complexity, it is challenging to obtain truly informed consent. In order to ensure the right level of information and understanding, genetic counselling usually accompanies genetic testing for cancer predisposition (Paluch-Shimon et al. 2016). The first consultation takes place before genetic testing: the genetic counsellor explains the testing procedure, possible outcomes, and consequences to at-risk individuals in order to enable informed decision-making. The second genetic consultation takes place after genetic testing and includes the discussion of the genetic test result, its meaning, and the next steps.

While genetic counselling has been the most essential information source for those seeking genetic testing, it faces challenges. Because of the increasing use of genetic testing worldwide, there are not enough genetic counsellors to cover the increasing demand (Ormond et al. 2018). This leads to long waiting lists and alternative models of counselling, such as group, online, or telephone counselling (Cloutier et al. 2017; Rayes et al. 2019). To meaningfully develop alternative information models, the information-seeking behaviour of at-risk individuals before and after genetic testing is highly relevant (Johnson et al. 2005). While studies so far have focused on information needs (e.g., Dean et al. 2017; Miller 1995), online information seeking (e.g., Abrahamson et al. 2008; Kaphingst et al. 2010) and the general extent of information-seeking behaviour (e.g., Cypowyj et al. 2003; Mills et al. 2015), there is a lack of empirical evidence around factors that motivate or hinder information-seeking among at-risk individuals.

4.2.2 Informing the public

Moreover, awareness, interest, and accessibility of genetic testing are increasing among the general population. Direct-to-consumer genetic testing (DTC-GT) companies sell genetic tests through the internet, without individual counselling and result interpretation. Even though some countries have banned DTC-GT (Kalokairinou et al. 2018), interested individuals can still purchase them elsewhere over the internet, making genetic testing accessible to any individual. Moreover, high-profile cases like Angelina Jolie's statement that she had preventive surgeries because of an increased genetic risk to develop cancer (Jolie Pitt, March 24, 2015; Jolie, May 13, 2013) increased public awareness of the existence of such predictive genetic tests and led

to a sustained increase in referrals worldwide (Freedman et al. 2017; Evans, D. G. et al. 2014). Because of these developments, thinking about how to inform the public is becoming a relevant issue. Most considerations are theoretical (Pickersgill 2011; Miah 2005), or survey studies (Abrahamson et al. 2008; Condit and Shen 2010; Jensen et al. 2017), and to our knowledge, at-risk individuals have never been asked about their opinion to what extent the public should be informed about genetics in any previous study.

4.2.3 Theoretical framework

Wilson's model of information behaviour serves as the theoretical framework for this study (Wilson 1997). He describes information-seeking behaviour as cyclic: Information needs lead to information-seeking behaviour, and the processing and use of this information might lead to new information needs. Activating factors and intervening variables are a central part of Wilson's model. They explain why some people seek more information than others do (despite similar information needs). The intervening variables include emotional, educational, demographic, social, environmental, and economic variables, as well as personal and source characteristics. Wilson also refers to interdisciplinary theories to explain the activating factors: Following stress/coping theory, whether people perceive a situation as stressful, and how they cope with stress influences information needs (Folkman 1984). Those intolerant of uncertainty but tolerant of arousal tend to have high information needs – they cope with stressful situations by using "consistent monitoring" ("monitors"). In contrast, those tolerant of uncertainty but stress-intolerant tend to avoid stressful situations; they thus do not engage in information-seeking behaviour and cope better with less information ("blunters") (Miller 1995; Miller and Mangan 1983). This coping behaviour influences information needs. Wilson connected two theories to the extent of information-seeking behaviour: high perceived risk might lead to more information-seeking behaviour according to risk/reward theory (Murray, K. 1991), and the theory of self-efficacy proposes that behaviour is guided by previous experiences of success or failure (Bandura 1977a). Wilson suggested this might also apply to information-seeking behaviour.

4.2.4 Aims

This article covers two aims. First, we aim to illustrate what at-risk individuals see as motivations and barriers for information-seeking behaviour before and after genetic testing. Second, we aim to assess what participants think the public should know about genetics.

4.3 Methods

4.3.1 Data collection

This study is part of a larger project, and the grounded theory approach (Strauss and Corbin 1990) was the methodological basis for data collection. The same interviews were analyzed concerning genetic testing decision-making[1], and for the purpose of this study, participants were particularly prompted to talk about their information-seeking strategies. Participants were eligible for the study if they were 18-70 years old, went to at least one genetic consultation concerning risk and testing for hereditary breast/ovarian cancer syndrome or Lynch syndrome and were psychologically stable. As the study progressed, we further specified the inclusion criteria, looking specifically for men and individuals deciding against genetic testing after genetic counselling, because these characteristics were interesting for our research questions and underrepresented. Table 4-1 portrays the characteristics of interview participants. We chose this heterogeneous population because it allows us to look at many perspectives regarding information-seeking behaviour before and after genetic consultation. All participants provided informed consent prior to the interview,[2] and the regional ethics committees of Northwest/Central Switzerland and Bern approved the research project (No 2017-00316).

Four healthcare professionals who provide genetic counselling in two Swiss university hospitals recruited participants by providing them with study information. Generally, participants proactively contacted the interviewer (BZ) if they were interested in participating, but four participants allowed the interviewer to contact them directly. We recruited participants between August 2017 and February 2019 and then stopped for reasons of time. The interviewer (BZ) held all interviews face-to-face at the hospital or the patients' home (according to participants' choice) and used a semi-structured interview guide.[3] We asked participants about the reasons, attitudes and information strategies regarding genetic testing.

4.3.2 Data analysis

All interviews were recorded and anonymized upon transcription. The language of the interviews varied: 15 interviews were in a Swiss-German dialect, two in High German, and one in French. The first author (BZ) translated the Swiss-German interviews to High German upon transcription and transcribed the remaining *ad verbatim*. The MaxQDA 2018 software (VERBI

1 See chapter 3.

2 See Appendix A-3 for the study information leaflet and the informed consent form.

3 See Appendix A-1 and A-2 for the interview guides.

Table 4-1: Characteristics of interview participants (n=18).

Gender	Female	14 (78%)
	Male	4 (22%)
Age	18-29	1 (6%)
	30-39	5 (28%)
	40-49	3 (17%)
	50-59	5 (28%)
	60+	4 (22%)
Genetic test result	Carrier of a pathogenic variant (self-reported)	7 (47%)
	Non-carrier of a pathogenic variant (including variants of unknown significance, self-reported)	8 (53%)
	No genetic test	3 (17%)
Syndrome tested for	Lynch syndrome	2 (11%)
	Hereditary breast/ovarian cancer	16 (89%)
Health status	Affected with cancer	6 (33%)
	Healthy at the time of genetic counselling	12 (66%)
Family member already diagnosed (mutation-positive)		5 (28%)
Blood-related children		12 (67%)
Mean interview duration (min)		59 (27-101)

GmbH) was used for assisting the analysis, which was conducted after data collection was completed (February to July 2019). We used qualitative thematic analysis on an interpretative level for examining underlying ideas of our participants, and we applied a realist approach, assuming a direct line between meaning, experience, and language (Braun and Clarke 2006). While thematic analysis is mainly known as an inductive methodology, Braun and Clarke (2006) also mention a deductive variant of thematic analysis, which they call "theoretical thematic analysis". Inspired by this methodological framework (see also Hayes 1997), we used Wilson's model of information-seeking behaviour (Wilson 1997) as a theoretical guide for data interpretation. Two researchers (BZ and JF) highlighted all parts of the interviews relevant for the research question and inductively assigned codes to these data, which were compared and discussed in detail. As a second step, they sorted these codes systematically, using Wilson's model as a framework, and extended the coding tree in several rounds of analysis and discussion. For example, we sorted codes that were relevant for "information needs", or that correspond to "activating mechanisms", and looked for additional data that fit these categories. We went through the data several times in order to catch all relevant codes. Then we applied Wilson's framework to each interview by writing interview-specific memos and subsequently wrote code-specific memos, where we summarized and interpreted all codes from each interview. Finally, we produced a descriptive report, which was translated into English by JF (proofread by BZ) and then reviewed

and commented on separately by three co-authors (IK, DS, MR). The final report served as a basis for the reported results. Reported quotes were translated into the English language by JF (native German speaker), double-checked by BZ (native German speaker, fluent in French), and proofread by DS (native English speaker).

4.4 Results

4.4.1 The cycles of information-seeking: motivation and barriers

We adopted Wilson's model on information-seeking behaviour in the context of genetic testing, identifying two cycles of information-seeking behaviour: before and after genetic testing. The outcomes of each cycle are informed decision-making as well as knowledge transfer to family members or others. In this study, we will focus on the mechanisms and variables that stand between information needs and information gathering.

The information-seeking process usually starts when learning about the possible relevance of genetic testing for the at-risk individual, which creates information needs. Participants mentioned cognitive information needs before genetic testing, including details on the testing procedure and general biological background knowledge, their risk of receiving a positive test result or developing the disease in the event of a positive result, and consequences in case of a positive test result.

> *"In the beginning, I didn't really know how the procedure works. I didn't inform myself about it; I just knew that I was just going to do the test. And then I arranged the first genetic consultation [...] I went to this first session completely unprepared, but the decision to do the test was very clear to me, I had thought about it for years. But rather on a relational basis, not through media or something... and not scientifically or anything. And then, the first consultation was very informative for me." (Mario, 20+, healthy)*

We also observed a need to elucidate and confirm already-existing beliefs and values. While some participants reported they needed information to evaluate whether genetic testing makes sense for them, others had already made their decision regarding genetic testing before starting to seek information – the primary function of the latter was to confirm their beliefs.

> *"It was important to me to hear their [the counselling healthcare professionals'] opinion. And I was glad that they also considered it [genetic testing] necessary. Because I had already lived for eight years with the idea that it was necessary, and if they had said, no, your risk is not high enough, then I would have found that kind of difficult." (Katy, 30+, healthy)*

Based on these information needs, at-risk individuals gather information, using a variety of sources (Table 4-2). Most participants used several sources: besides the genetic consultation, many searched for information online and added some additional, individual sources.

A few participants also mentioned that they had no further information needs after genetic counselling. The medical experts mentioned the importance of appropriate information sources and the lack of availability. They indicated that they produce their own information material and make their patients aware of appropriate information channels and materials (both online and offline).

Table 4-2: Information sources that participants used before or after genetic testing decision-making.

Information source	Evaluation	Experience	When used
Genetic counselling session	Mostly very good and trustworthy	For many the main source of information	Before and after
Online search	Some pointed out that it was difficult to find trustworthy sources	The main source to look for additional information, low barriers	Before and after
Online forums	More of emotional support, factual information is treated suspiciously	Not considered as a very important source of information	Before and after
Exchange with family members	Helpful or difficult, depending on the family situation	Emotional support and information dissemination from previously tested individuals	Before and after
Public information events / lectures	Mostly trustworthy information if held by professionals	Difficult to learn when these take place	Before and after
Self-help groups	Members appreciate these meetings a lot	Emotional support and some factual information, opportunity to ask questions	After
Scientific literature	Very trustworthy, but risk to get lost in details	Only available for highly educated individuals	Before and after
Information sheets from hospitals	Helpful but limited in the amount of information		Before and after
Healthcare professional (gynaecologist, GP, enterologist)	Many do not know much about genetics	Passive information gathering, highly dependent on the healthcare professional	Before and after
Newspapers, radio, TV, magazines	Coverage often perceived as superficial and populist	Coverage is not very salient; many do not recall having read anything related to genetic testing	Before and after
Guidelines (e.g. WHO)	Give objective facts and advice		After

After genetic testing, the information-seeking process starts again, especially when the genetic test came back positive. Information needs arise when concrete preventive measures are an option, and when the fear of getting sick is increasing. The main interest of those carrying a pathogenic variant concern the next steps, disease risk, information dissemination to relatives and children, psychological follow-up care, and discrimination.

> *"I was interested in the WHO [World Health Organisation] statistics because that must be meaningful; that's why I was looking for it. I thought, well, since the WHO deals with it, it is useful. I especially looked for BRCA." (Edwin, 50+)*

Most of those who tested negatively for a pathogenic variant no longer have any need for information; they consider their risk as settled. Even though a negative test result does not necessarily mean that there is no need for precautions, many of those with negative results have a strong feeling of security and relief.

> *"Well, the counsellor said the test results were somehow, eh, just so that you would call it negative... Negative, and not positive... But there's apparently something they still can't decipher today, and they could know more about that in a few years, but I realized then that this was getting too complicated for me. I don't really want to know all that, then I think in the end you almost live with that feeling, eh, what's still sleeping inside me. That is when I feel like, I'm good now, that's enough." (Susanna, 60+)*

Several factors motivate or hinder at-risk individuals' information gathering, even if there is an information need (Tables 4-3 and 4-4).

Table 4-3: Motivators for information-seeking behaviour.

Motivators		**Illustrating quotes**
Trust in physicians	Urge to double-check the doctors' assessments and recommendations	"I have read a lot of these scientific articles, and, well I like statistics and scientific methods, that's why I am quite capable of reading them, even if it is not my area of expertise. And... I like it that I can inform myself because I don't necessarily trust doctors easily if they tell me something." (Pamela, 30+)
Risk perception	High perceived risk	"My mother died from cancer many years ago, and the oncologist told us back then that it could be genetic [...] And more recently, my aunt, my mother's sister got the same kind of cancer. And that of course meant a red alert for me, I thought, well, probably it's genetic anyway. And I directly told my gynaecologist and asked him, could that be genetic and should I see a geneticist." (Carmen, 50+)

Table 4-3 (continued).

Motivators		**Illustrating quotes**
Previous knowledge	Catalyzes information search; the search is more targeted	"[…] I guess I had enough prior knowledge that […] the genetic consultation resulted in another discussion. So maybe if one has to, well, start with the basics, then I can imagine it is difficult, then it's an awful lot of information one has to understand and digest and uhm… And I think I didn't have to go through this process, somehow. Or didn't need it." (Brigitte, 60+)
Poor physician knowledge of genetics	Need and urge to acquire knowledge to manage prevention plan independently	"If you see a doctor and tell him, I want to have a regular check-up because I am at risk, then he doesn't know anything about it. He doesn't know [about genetic risks]. A family doctor usually knows about it; the specialists don't [...] In genetic counselling, some recommendations were made about how often I should go to preventive screenings. And with that information you go to your doctor, so you actually go to the doctor with more knowledge than he has, and that is certainly a good thing." (Edwin, 50+, mutation-positive)
Life philosophy	Knowledge is always important, mutation is not a disease, allows proactive prevention	"I just want to know, I think I'm just this kind of person, I can cope better, although I know it is... difficult, but I cope better." (Carmen, 50+)
Feeling of responsibility	Not getting informed is perceived as irresponsible	"My sisters are not really interested in genetic testing. I heard that one of my sisters said she doesn't want to know our mother's test result, and she isn't thinking about doing a test herself, she just doesn't seem to care. Both of my sisters are somehow not interested at all. But I sometimes think... they haven't even thought about the possibility that this could have an impact on them, or that their risk could be increased, they just don't care about this. [...] I just think that is a little... careless, maybe." (Donna, 30+)
Family position	Index patients need more information	"I was always the frontrunner, I was the first in my family who went to see a geneticist [for genetic counselling], and my cousins went there after me [...]." (Carmen, 50+)
Emotions	A feeling of hope for genetic progress enhances general interest in genetics	Rebecca had experiences related to genetics that she did not want to discuss during the interview. She seemed to be very emotional about it. These emotions drove her interest in getting more information: "Erm, there is a topic related to genetics in my life… Ha, I have difficulties talking about this right now, to be honest, to disclose this, too... Uhm… I would like to be informed, yes. What happens in the future. In genetics. But how one gets the information... Uhm, well… I think if I really had the need for information, I would really talk to an expert. Uhm… But it actually interests me what will happen in research." (Rebecca, 40+)

Table 4-3 (continued).

Motivators		**Illustrating quotes**
Disease	Wanting to know the cause of the disease	"Both, BRCA1 and BRCA2 were negative, there was nothing there. And at that point, it's true that… I was relieved, but I immediately asked myself, but where does it come from then? My cancer must have another cause then. And then my doctor proposed testing other genes." (Pamela, 30+)
Decision-making style	Weighted decision-making	"Before I did the genetic test, I went to counselling one more time and, before that, I informed myself in an online forum. [...] And gathered information about, what do you test for, what is important…" (Elsa, 40+)
Age	Being at the high-risk age	Interviewer: "What were you especially interested in?" Participant: "In the next step. Without having the result yet, what would it mean for me to be mutation-positive, what would be the next step." Interviewer: "And what did you find out?" Participant: "Well, had I been mutation-positive, it would have been about removing my breasts. That would have been the next step. Because I knew, the geneticist had also suggested it, that if I did the genetic examination, to be aware of possible consequences. And think about them upfront. And that's what I was searching the internet for, actually." (Marisa, 50+)

Table 4-4: Barriers to information-seeking behaviour.

Barriers		**Illustrating quotes**
Trust in physicians	Inherent trust towards doctors	"Yeah, well, if the doctor tells me anything, I'll do it anyway, won't I." (Josephine, 60+)
Risk perception	Low perceived risk	"What also likely had an impact [on deciding against testing] was, what is the probability that I actually have a mutation…? Ehm, rationally, it is not high. If I had a big, a really huge probability [of testing positive] […], then I'd say, well I basically must do it, it would be, eeeh, well, irresponsible - I also have small children - it would be irresponsible if I said no, and if there are options to do something [preventive] upfront. [...] But when I saw in black and white how small the probability is that I'm affected, it holds up for me that I made this decision." (Rebecca, 40+)
Previous knowledge	Feeling of already knowing enough	Interviewer: "During the whole process, how did you inform yourself about genetic testing?" Participant: "Uhm… did I even do this much…? I think I already knew a lot from my medical studies." (Ramona, 30+)

Table 4-4 (continued).

Barriers		**Illustrating quotes**
Poor physician knowledge of genetics	Feeling paralyzed and overstrained, need for guidance	"But I feel a bit like I'm in a vacuum because there is very little information, the gynecologist didn't know much, he simply referred to the geneticist for detailed information. Or regarding the breast removal, he said he could recommend a plastic surgeon, but he himself didn't know much. That was a bit frustrating because he was actually my first contact person. And somehow you feel like, phew, what am I doing now, or, where can I go to get information, and that's difficult." (Carmen, 50+)
Life philosophy	Feeling comfortable without knowing everything	"I was told once, when my mother died [...] that I was genetically at risk. I wish they hadn't told me that, I have to admit. [...] I think it would have been better if they had said, now that your mother died, the whole story is over." (Ted, 60+)
Feeling of responsibility	Protect from responsibility: once you know, you cannot go back	"I think […] many people don't want to know too many details, they don't want to have to deal with all of it. They don't want to think about this themselves at all, it's… a lot of responsibility, because once you know... you cannot go back." (Pamela, 30+)
Family position	Profit from family member's information	"My aunt and her husband, they have sent me all their documents, and they contain those WHO statistics, but you can find them online as well." (Edwin, 50+)
Emotions	Fear and negative emotions due to family history or own disease	Interviewer: "You said your wife was watching these health programs." Participant: "Yes." Interviewer: "But you don't watch them." Participant: "Me, not at all, no." Interviewer: "Why not?" Participant: "Ah I don't want to. Disease, well that's totally… You know, I think I might be a bit emotionally charged because of my mother. That wasn't nice to watch, how she died back then [...]" (Ted, 60+)
Disease	Lack of energy or lack of time	"I really consciously put the genetic test aside and simply waited until I was 40, and didn't let that come close to me. I didn't have time, anyway. I was really busy with my baby daughter and my breast cancer, so… yeah. I somehow lacked the energy, it wasn't important enough." (Elsa, 40+, in remission)
Decision-making style	Intuitive decision-making	"I knew quite quickly that I would do genetic testing, because […] I knew a little about it... It was offered to me, and... yes, it was clear to me that I wanted to know because I have two daughters." (Mariella, 50+, in remission)
Age	Young age – disease risks still low Old age - implications of test and its outcome not of personal relevance	"Well, I'm not at the age where I could get pregnant anymore, that's all over, I think age probably plays a role there as well. [...] I think today it is much more difficult for the young people than it used to be. With all that genetics. Not easier. Because our generation didn't have to think about that. And today young people must decide whether they want such a test or not, they have to learn to actively say no." (Brigitte, 60+)

4.4.2 Information to others

Some at-risk individuals stated that informing the public about genetics is not useful, because "nobody is interested in that" (Mariella, 50+, in remission). However, Elsa (40+, in remission) stated: "That is alright. People don't need to know that. Because if someone is personally affected, he or she will get the information. The system here at the hospital works well; people cannot slip through."

Others found that informing the public was important: "If someone has to decide and doesn't have previous knowledge, it is easier for them to be introduced to the topic if it becomes relevant, I'd say." (Ramona, 30+) Some participants consider it generally important to talk about disease risk and to break the existing taboo around it: "I feel the fear of people of becoming old and sick. Somehow, I have advanced knowledge, and if colleagues talk with me and there is an occasion, I tell them my story…" (Edwin, 50+, carrier). Additionally, participants considered it essential that people received appropriate counselling and thought about consequences before having a genetic test.

Another advantage of an informed public that participants mentioned was that it makes it easier for carriers to talk about their diagnosis. Some feel tired from explaining everything repeatedly to their unknowledgeable friends. It would make it easier to exchange their situation and fears. Generally, those in our sample who are carriers of a pathogenic genetic variant considered informing the public as more important than those who are not carriers.

Most participants used the internet in addition to genetic counselling. However, "it is difficult to find good quality information online. Nowadays, everybody writes something somehow […]" (Ted, 60+). The same is true for newspaper coverage: "There has been a lot of… populist, or unobjective, reporting I feel." (Donna, 30+) Some participants, however, have never even read, watched or heard something about genetics in mass media.

4.5 Discussion and Conclusion

4.5.1 Discussion

This study provides multiple motivators and barriers from qualitative interview data with people undergoing genetic counselling for testing regarding inheritable cancer risk. Established psychological theories help explain many of the motivators and barriers identified. First, the influence of life philosophy leads back to coping theory: those who cope better with more information ("monitors") show more information-seeking behaviour than those who prefer little

information ("blunters") (Miller 1995). According to social learning theory (Bandura 1977b), if collecting information enhances perceived self-efficacy, it provides peace of mind and security, reduces diffuse anxiety, and can calm people down. However, if individuals perceive their self-efficacy in gathering information as low, for example, due to overstraining because of the lack of guidance or negative emotions, it hinders their information-seeking behaviour. Second, the theory of decision-making styles (Scott and Bruce 1995), which says that individuals have different styles of decision-making that they apply quite consistently, confirms our observation that intuitive decision-making hinders information-seeking behaviour because these individuals have already made their decision before starting information gathering. In contrast, weighted decision-making (in the literature referred to as "rational decision-making" (Scott and Bruce 1995) is connected to extensive information gathering. Third, risk/reward theory (Murray, K. 1991) helps explain the influence of risk perception on information-seeking behaviour: If the perceived risk of being a carrier of a pathogenic genetic variant is low, there is a high chance that information is gathered for nothing, and thus that there is no reward for the effort. On the other hand, a high perceived risk leads to higher reward expectation if the anticipated event occurs. Moreover, Wilson suggested five intervening variables in his model of information behaviour (Wilson 1997), which help to explain the remaining motivating and hindering factors we identified. The demographics describe the factors age and previous knowledge (as it connects back to the educational background). The family position connects to the social role of the individual. The relationship with medical doctors describes the credibility that individuals assign to this information source. The role of disease is an environmental variable. Finally, the feeling of responsibility, as well as emotions, are part of individuals' psychological predisposition. Several studies indicate that anxiety leads to information avoidance (Case et al. 2005; Miles et al. 2008).

This study also provides insights into the opinion of at-risk individuals regarding the extent to which the public should be informed about genetics. As some of our participants pointed out, the complexity of genetic testing makes it beneficial to provide the public with some general background information, a demand in line with the findings of another study (Mesters, Ausems, and de Vries 2005). However, too much information might lead to information overload, stress, or overemphasis, meaning that people think they are at risk when they are not. For example, press coverage regarding predictive genetic testing for hereditary breast/ovarian cancer exploded after the public statement of Angelina Jolie that she had a preventive mastectomy due to her genetic risk (Zimmermann et al. 2019; Jolie, May 13, 2013), leading to cases of unjustified requests for genetic testing and preventive surgery (Freedman et al. 2017).

Based on our results and the conclusions drawn from theories above, it is not beneficial, nor necessary, to intensively "educate" the public about all the complexities of genetic testing. A better approach is to provide some general background information that makes it easier for them to seek information if genetic testing becomes personally relevant to them. This contradicts the assumption that more information is always better. While certain general background information should be available for everybody, the details of genetic testing are too complex and too individual to be given in a general manner to the public. However, general background information should function as a motivator for information-seeking behaviour: a certain degree of previous knowledge, which we have identified as one of the motivators, should not be limited to highly educated individuals or healthcare professionals. To have equal access to information is essential for individual information seeking; this is a matter of social justice and helps reduce inequalities connected to health (Hill, L. 2004). What exactly general background information entails, however, is highly context- and culture-dependent because countries differ significantly regarding legislation (Kalokairinou et al. 2018; Soini 2012), public attitudes (Gaskell et al. 2000) and pre-existing mass media portrayal (Zimmermann et al. 2019). In general, we think that ethical, legal, and social issues regarding genetic testing are of high relevance for the general public and should be prioritized (Zimmermann, Elger, and Shaw 2019; Miah 2005).

Swiss mass media seem to fail to fulfil people's expectations regarding balanced and informative reporting since some participants complained about the populistic and imprecise media coverage. This finding needs to be investigated in other contexts, since the media portrayal of genetic testing is more comprehensive in other countries, such as the UK (Zimmermann et al. 2019). At the same time, if someone actively seeks information regarding genetic testing online, it is difficult for laypeople to distinguish between accurate and misleading information. The issue of finding too little and, at the same time, too much information online is also described for other health-related issues (Synnot et al. 2016). Thus, strategies are needed to deal with these issues of inadequate information provision and information overload. Research on online information-seeking behaviour advises health professionals to recommend appropriate information material to patients, help patients to search and evaluate online information more effectively, and get involved in developing high-standard information material (Morahan-Martin 2004). While our medical experts confirm this pathway, resources are limited and sharing reliable information online and offline is crucial to enhancing access and quality of information outside the clinical setting.

The first limitation of this study concerns the sample size. We conducted and analyzed 18 interviews, which provided comprehensive insight into the motivators and barriers, but do not allow quantitative measures of, for example, how important each factor is. Even though we strived to reduce personal bias through constant reflection, team discussions, and a rigorous methodology, qualitative analysis is an inherently interpretative process, and another research group might have framed results differently. Moreover, choosing a different theoretical model as the basis for analysis might also have altered the results. We chose Wilson's model because it explicitly emphasizes motivations and barriers, which is an underrepresented aspect in most other models on information-seeking behaviour (Case et al. 2005). Selection bias is another limitation of this study. The recruitment procedure demanded most participants to contact the interviewer proactively if they wanted to participate. Anxious and avoiding at-risk individuals thus are probably underrepresented in our sample. However, we do have some individuals that we identified as "blunters" (Miller 1995), and we also have detailed descriptions of our participants about family members who were anxious and avoiding information. Moreover, since we asked participants not only about their information-seeking strategies but also aimed to conduct a grounded theory study on decision-making,[4] the other part of the project may have influenced our results. However, because of the exploratory nature of this study, our findings are still valid and, in fact, even enriched by the broader scope we obtained through the grounded theory study.

4.5.2 Conclusion

We empirically identified motivators of and barriers to information-seeking behaviour regarding genetic testing for cancer predisposition. While existing models and interdisciplinary theories verify and contextualize our findings, our results are more concrete and help healthcare professionals, especially genetic counsellors, oncologists, general practitioners and other medical doctors involved in post-testing cancer prevention to understand and support information seeking based on individual needs. Based on the participants' assessment, it seems sufficient to provide the public with general background information regarding genetic testing in order to facilitate information seeking if it becomes relevant for an individual. This avoids information overload at the moment of genetic counselling and accounts for limited financial resources. Professionals involved in public health communication should consider providing less but more concise information to the public, and invest energy in providing holistic and in-depth information for those at risk.

4 See chapter 3.

4.5.3 Practice implications

Our findings have several implications for clinical practice. First, they provide a concrete narrative for healthcare professionals to understand their patients' patterns and wishes concerning information-seeking behaviour. For instance, they should make monitors aware of websites with high-quality information on their topic of interest. Moreover, we conclude that there are different types of "blunters": on the one hand, those who act in line with their life philosophy, who are happy with little information and feel comfortable in outsourcing decisions to third parties (such as healthcare professionals). On the other hand are those who are in emotional distress, information overload, or under social pressure. While the first type can give informed consent, the second cannot, and might need further support. For instance, they could be sent to psychotherapy to discuss their avoiding behaviour. At the same time, healthcare professionals should be aware of these issues, respect that some are happy with little information and not push them into becoming "monitors" against their will. More research is needed to develop analytical models for such an assessment in clinical practice.

4.6 References

Abrahamson, Jennie A., Karen E. Fisher, Anne G. Turner, Joan C. Durrance, and Tammara C. Turner. 2008. "Lay Information Mediary Behavior Uncovered: Exploring How Nonprofessionals Seek Health Information for Themselves and Others Online." J Med Libr Assoc 96 (4): 310–23. doi:10.3163/1536-5050.96.4.006.

Bandura, Albert. 1977a. "Self-Efficacy: Toward a Unifying Theory of Behavioral Change." Psychol Rev 84 (2): 191–215.

———. 1977b. Social Learning Theory. Englewood Cliffs, NJ: Prentice-hall.

Braun, Virginia, and Victoria Clarke. 2006. "Using Thematic Analysis in Psychology." Qual Res Psychol 3 (2): 77–101. doi:10.1191/1478088706QP063OA.

Case, Donald O., James E. Andrews, J. David Johnson, and Suzanne L. Allard. 2005. "Avoiding Versus Seeking: The Relationship of Information Seeking to Avoidance, Blunting, Coping, Dissonance, and Related Concepts*." J Med Libr Assoc 93 (3): 353–62.

Cloutier, Mireille, Lauren Gallagher, Claire Goldsmith, Salwa Akiki, Nick Barrowman, and Shawna Morrison. 2017. "Group Genetic Counseling: An Alternate Service Delivery Model in a High Risk Prenatal Screening Population." Prenat Diagn 37 (11): 1112–19. doi:10.1002/pd.5149.

Condit, Celeste M., and Lijiang Shen. 2010. "Public Understanding of Risks from Gene-Environment Interaction in Common Diseases: Implications for Public Communications." Public Health Genom 14 (2): 115–24. doi:10.1159/000314915.

Cypowyj, C., Francois Eisinger, M. Morin, A. Mogoutov, H. Sobol, and C. Julian-Reynier. 2003. "Information-Seeking Behaviour and Psycho-Social Interactions During the Genetic Testing Process." Community Genet 6 (4): 224–34. doi:10.1159/000079384.

Dancyger, Caroline, Mel Wiseman, Chris Jacobs, Jonathan A. Smith, Melissa Wallace, and Susan Michie. 2011. "Communicating BRCA1/2 Genetic Test Results Within the Family: A Qualitative Analysis." Psychol Health 26 (8): 1018–35. doi:10.1080/08870446.2010.525640.

Dean, Marleah, Courtney L. Scherr, Meredith Clements, Rachel Koruo, Jennifer Martinez, and Amy Ross. 2017. ""When Information Is Not Enough": A Model for Understanding BRCA-Positive Previvors' Information Needs Regarding Hereditary Breast and Ovarian Cancer Risk." Patient Educ Couns 100 (9): 1738–43. doi:10.1016/j.pec.2017.03.013.

Evans, D. Gareth R., Julian Barwell, Diana M. Eccles, Amanda Collins, Louise Izatt, Chris Jacobs, Alan Donaldson et al. 2014. "The Angelina Jolie Effect: How High Celebrity Profile Can Have a Major Impact on Provision of Cancer Related Services." Breast Cancer Res. 16 (5): 442. doi:10.1186/s13058-014-0442-6.

Evans, James P., Cécile Skrzynia, and Wylie Burke. 2001. "The Complexities of Predictive Genetic Testing." BMJ 322 (7293): 1052–56. doi:10.1136/bmj.322.7293.1052.

Folkman, Susan. 1984. "Personal Control and Stress and Coping Processes: A Theoretical Analysis." J Pers Soc Psychol 46 (4): 839–52.

Freedman, Rebecca, Helen Mountain, Dian Karina, and Lyn Schofield. 2017. "A Retrospective Exploration of the Impact of the 'Angelina Jolie Effect' on the Single State-Wide Familial Cancer Program in Perth, Western Australia." J Genet Couns 26 (1): 52–62. doi:10.1007/s10897-016-9982-5.

Gaskell, George, Nick Allum, Martin W. Bauer, John Durant, Agnes Allansdottir, Heinz Bonfadelli, Daniel Boy et al. 2000. "Biotechnology and the European Public." Nat Biotechnol 18:935 EP -. doi:10.1038/79403.

Hayes, Nicky. 1997. "Theory-Led Thematic Analysis: Social Identification in Small Companies." In Doing Qualitative Analysis in Psychology, edited by Nicky Hayes, 93–114. Hove: Psychology Press.

Hill, Lilian H. 2004. "Health Literacy Is a Social Justice Issue That Affects Us All." Adult Learning 15 (1-2): 4–6. doi:10.1177/104515950401500101.

Jensen, Jakob D., Manusheela Pokharel, Courtney L. Scherr, Andy J. King, Natasha Brown, and Christina L. Jones. 2017. "Communicating Uncertain Science to the Public: How Amount and Source of Uncertainty Impact Fatalism, Backlash, and Overload." Risk Anal 37 (1): 40–51. doi:10.1111/risa.12600.

Johnson, J. David, Donald O. Case, James E. Andrews, and Suzanne L. Allard. 2005. "Genomics - the Perfect Information-Seeking Research Problem." J Health Commun 10 (4): 323–29. doi:10.1080/10810730590950048.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jolie Pitt, Angelina. 2015. "Diary of a Surgery." The New York Times, March 24. Accessed June 07, 2017. https://www.nytimes.com/2015/03/24/opinion/angelina-jolie-pitt-diary-of-a-surgery.html?_r=0.

Kalokairinou, Louiza, Heidi C. Howard, Santa Slokenberga, Eva Fisher, Magdalena Flatscher-Thöni, Mette Hartlev, Rachel E. van Hellemondt et al. 2018. "Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape." J Community Genet 9 (2): 117–32. doi:10.1007/s12687-017-0344-2.

Kaphingst, Kimberly A., Colleen M. McBride, Christopher Wade, Sharon H. Alford, Lawrence C. Brody, and Andreas D. Baxevanis. 2010. "Consumers' Use of Web-Based Information and Their Decisions About Multiplex Genetic Susceptibility Testing." J Med Internet Res 12 (3): e41. doi:10.2196/jmir.1587.

Kusev, Petko, Paul van Schaik, Rose Martin, Lars Hall, and Petter Johansson. 2019. "Preference Reversals During Risk Elicitation." J Exp Psychol Gen Advance online publication. doi:10.1037/xge0000655.

Mahon, Suzanne M. 2015. "Management of Patients with a Genetic Variant of Unknown Significance." Oncol Nurs Forum 42 (3): 316–18. doi:10.1188/15.ONF.316-318.

Mesters, Ilse, A. Ausems, and Hein de Vries. 2005. "General Public's Knowledge, Interest and Information Needs Related to Genetic Cancer: An Exploratory Study." Eur J Cancer Prev 14 (1): 69–75. doi:10.1097/00008469-200502000-00010.

Miah, Andy. 2005. "Genetics, Cyberspace and Bioethics: Why Not a Public Engagement with Ethics?" Public Underst Sci 14 (4): 409–21. doi:10.1177/0963662505056616.

Miles, Anne, Sanne Voorwinden, Sarah Chapman, and Jane Wardle. 2008. "Psychologic Predictors of Cancer Information Avoidance Among Older Adults: The Role of Cancer Fear and Fatalism." Cancer Epidemiol Biomarkers Prev 17 (8): 1872–79. doi:10.1158/1055-9965.EPI-08-0074.

Miller, Suzanne M. 1995. "Monitoring Versus Blunting Styles of Coping with Cancer Influence the Information Patients Want and Need About Their Disease. Implications for Cancer Screening and Management." Cancer 76 (2): 167–77.

Miller, Suzanne M., and Charles E. Mangan. 1983. "Interacting Effects of Information and Coping Style in Adapting to Gynecologic Stress: Should the Doctor Tell All?" J Pers Soc Psychol 45 (1): 223.

Mills, Rachel, Jill Powell, William T. Barry, and Susanne B. Haga. 2015. "Information-Seeking and Sharing Behavior Following Genomic Testing for Diabetes Risk." J Genet Couns 24 (1): 58–66. doi:10.1007/s10897-014-9736-1.

Morahan-Martin, Janet M. 2004. "How Internet Users Find, Evaluate, and Use Online Health Information: A Cross-Cultural Review." Cyberpsychol Behav 7 (5): 497–510. doi:10.1089/cpb.2004.7.497.

Murray, Keith B. 1991. "A Test of Services Marketing Theory: Consumer Information Acquisition Activities." J Marketing 55 (1): 10–25. doi:10.2307/1252200.

Ormond, Kelly, Mercy Y. Laurino, Kristine Barlow-Stewart, Tina-Marié Wessels, Shelley Macaulay, Jehannine Austin, and Anna Middleton. 2018. "Genetic Counseling Globally: Where Are We Now?" Am J Med Genet C 178 (1): 98–107. doi:10.1002/ajmg.c.31607.

Paluch-Shimon, Shani, Fátima Cardoso, Cristiana Sessa, Judith Balmaña, Maria J. Cardoso, Fiona Gilbert, and Elżbieta Senkus. 2016. "Prevention and Screening in BRCA Mutation Carriers and Other Breast/ovarian Hereditary Cancer Syndromes: ESMO Clinical Practice Guidelines for Cancer Prevention and Screening." Ann Oncol 27 (suppl 5): v103-v110. doi:10.1093/annonc/mdw327.

Pickersgill, Martyn D. 2011. "Research, Engagement and Public Bioethics: Promoting Socially Robust Science." J Med Ethics 37 (11): 698–701. doi:10.1136/jme.2010.041954.

Rayes, Nadine, Deborah J. Bowen, Tara Coffin, Denise Nebgen, Christine Peterson, Mark F. Munsell, Kathleen Gavin et al. 2019. "MAGENTA (Making Genetic Testing Accessible): A Prospective Randomized Controlled Trial Comparing Online Genetic Education and Telephone Genetic Counseling for Hereditary Cancer Genetic Testing." BMC Cancer 19:648. doi:10.1186/s12885-019-5868-x.

Scott, Susanne G., and Reginald A. Bruce. 1995. "Decision-Making Style: The Development and Assessment of a New Measure." Educational and Psychological Measurement 55 (5): 818–31. doi:10.1177/0013164495055005017.

Soini, Sirpa. 2012. "Genetic Testing Legislation in Western Europe-a Fluctuating Regulatory Target." J Community Genet 3 (2): 143–53. doi:10.1007/s12687-012-0078-0.

Strauss, Anselm, and Juliet M. Corbin. 1990. Basics of Qualitative Research: Grounded Theory Procedures and Techniques. Newbury Park, Calif: SAGE Publications.

Synnot, Anneliese J., Sophie J. Hill, Kerryn A. Garner, Michael P. Summers, Graziella Filippini, Richard H. Osborne, Sue D. P. Shapland, Cinzia Colombo, and Paola Mosconi. 2016. "Online Health Information Seeking: How People with Multiple Sclerosis Find, Assess and Integrate Treatment Information to Manage Their Health." Health Expect 19 (3): 727–37. doi:10.1111/hex.12253.

Wilson, Thomas D. 1997. "Information Behaviour: An Interdisciplinary Perspective." Comm Com Inf Sc 33 (4): 551–72. doi:10.1016/S0306-4573(97)00028-9.

Winchester, Ellen, and Shirley V. Hodgson. 2006. "Psychosocial and Ethical Issues Relating to Genetic Testing for BRCA1 and BRCA2 Breast Cancer Susceptibility Genes." Women's health 2 (3): 357–73. doi:10.2217/17455057.2.3.357.

Zimmermann, Bettina M., Noah Aebi, Steffen Kolb, David Shaw, and Bernice S. Elger. 2019. "Content, Evaluations and Influences in Newspaper Coverage of Predictive Genetic Testing: A Comparative Media Content Analysis from the United Kingdom and Switzerland." Public Underst Sci 28 (3): 256-274. doi:10.1177/0963662518816014.

Zimmermann, Bettina M., Bernice S. Elger, and David Shaw. 2019. "Media Coverage of Ethical Issues in Predictive Genetic Testing: A Qualitative Analysis." AJOB Empirical Bioethics 10 (4): 250–64. doi: 10.1080/23294515.2019.1670275.

Chapter 5: Autonomy and social influence in predictive genetic testing decision-making: a qualitative interview study

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5.1 Abstract

Beauchamp and Childress' definition of autonomous decision-making includes the conditions of intentionality, understanding, and noncontrol. In genetics, however, a relational conception of autonomy is implemented, which has been increasingly recognised. This article aims to empirically assess aspects of social influence in genetic testing decision-making and to connect these with principlist and relational theories of autonomy. We interviewed eighteen genetic counsellees and two healthcare professionals in Switzerland and performed a qualitative analysis based on a grounded theory study about predictive genetic testing decision-making. We found that some participants agreed to predictive genetic testing predominantly because relatives wanted them to do it, with some even acting contrary to their own convictions. Others, in contrast, based their decision on purely individualistic reasons but expressed difficulties in explaining their decision to their social environment. Healthcare professionals had a critical influence on decision-making is many cases without being manipulative, as perceived by counsellees. Still, cases of coercion and social pressure occurred within social relationships. In conclusion, predictive genetic testing decision-making includes relational and individualistic aspects, and both are compatible with autonomous decision-making. However, in both cases, individuals might face implicit or explicit social pressure. Therefore, while the principlist and relational notions of autonomy are competing on a theoretical level, they are two sides of the same medal when used as lenses of analysis for genetic testing decision-making.

5.2 Introduction

According to Beauchamp and Childress, an autonomous action requires intention, understanding and the absence of controlling influences, meaning it must be made "freely in accordance with a self-chosen plan" (Beauchamp and Childress 2013, 101). Even though Beauchamp and Childress acknowledge that social influences are unavoidable and permissible until a certain extent (Beauchamp and Childress 2013, 104–5), feminist philosophers have criticised the underlying conception of autonomy as too individualistic, disregarding the influence of social relations in human actions and decisions (Donchin 2001; Mackenzie and Stoljar 2000).

Their alternative conception of relational autonomy embeds social relationships and their influence on decisions and actions. Relational autonomy theories can be classified as substantive, meaning value-laden, or procedural, meaning content-neutral (Stoljar 2018). We here use the procedural conception of relational autonomy, thus examining the decision-making process in predictive genetic testing irrespective of the decision. In John Christman's procedural conception,

decisions are autonomous if individuals are cognitively and normatively competent during the decision-making process (i.e., rational and self-controlled), and their decision is self-reflected (Christman 2009). Marilyn Friedman has a similar conception of procedural autonomy with a lower threshold, in the sense that she considers a decision as autonomous as soon as a person is minimally self-reflective (Friedman, M. 2003). Both, Christman and Friedman, emphasise the influence of the social environment for this self-reflection but refrain from the claim that social relations constitute autonomy, meaning that social relations are necessary to act autonomously (Christman 2004). Accordingly, Bruce Jennings observed a "relational turn" in the conception of autonomy in biomedical ethics:

> *"The challenge of respecting the autonomy of persons is not to avoid relationality but to distinguish those forms of relationship and life worlds that are functional for the maintenance of communal meaning and integrity from those that are not." (Jennings 2016, 13)*

Especially in clinical genetics, the influence of social relations has been increasingly recognised (Ho 2008). Empirical inquiries have confirmed that family considerations often influence genetic testing decision-making (Etchegary et al. 2009; Gilbar and Barnoy 2018). Particularly in predictive genetic testing, the decision to test not only uncovers information about the tested individual but also reveals facts about blood relatives. Therefore, relatives also receive predictive information about disease risks and thus may face the decision of whether or not they want to perform such a test. Because each person has a right to know, but at the same time also a right not to know genetic risk information (Andorno 2004), this leads to potential dilemmas if individuals from the same family have divergent preferences (Hallowell et al. 2003).

Here, we examine autonomy in the context of decision-making in predictive genetic testing for Hereditary Breast and Ovarian Cancer syndrome (HBOC) or Lynch Syndrome, which increases the risk of colon cancer (Lynch, H., Smyrk, and Lynch, J. 1997). Predictive genetic testing is, per definition, performed in asymptomatic individuals (Skirton et al. 2013). Those carrying a risk-increasing genetic variant can opt for more regular preventive screening, or, in the case of HBOC, for preventive, risk-reducing surgery (Llort et al. 2015; Stoffel et al. 2015).

Empirical studies on genetic testing decision-making mainly come from North America (Etchegary et al. 2009; Hamilton, R. and Bowers 2007) and the United Kingdom (Foster et al. 2002; Hallowell et al. 2005). More recently, such studies have been conducted in other European countries such as Italy (Godino et al. 2018) or Spain (Rivera-Navarro, Cubo, and Mariscal 2015), but not in Switzerland. The legal and clinical context in Switzerland differs from other countries. Genetic counselling before and after predictive genetic testing from a specialised medical doctor

is legally required (Swiss Federal Council 2004, Art 14). Moreover, the Swiss health insurance covers the costs of genetic testing and genetic counselling under certain conditions, including genetic testing for cancer predisposition in the case of relevant family history or early-onset cancer (Swiss Federal Council 1994, Art 26). Only few genetic counsellors work in Swiss clinics (Abacan et al. 2019), and it is not an officially accredited profession.

This article aims to explore aspects of social influences in predictive genetic testing decision-making in Switzerland and discusses consequences on the conception of autonomy in that context. This is the first study providing empirical evidence about the social influence in genetic testing from Switzerland.

5.3 Methodology

This study is part of a grounded theory analysis based on an interview study with people undergoing genetic counselling for cancer predisposition (HBOC or Lynch syndrome) in a clinical setting in Switzerland. We asked participants in semi-structured face-to-face interviews about their reasons, attitudes and information strategies regarding predictive genetic testing and analysed the interviews in terms of the decision-making process and information-seeking behaviour. For data triangulation, we also interviewed two counselling physicians regarding the topics that came up during analysis. For this study, we reanalysed the codes obtained from the analysis of the decision-making process from the theoretical perspective of relational and individualistic autonomy (Frith 2012).

Detailed information regarding recruitment and characteristics of participants are presented in the primary publication.[1] Inclusion criteria were participation in at least one genetic counselling session, healthy mental state, non-pregnancy and 18-75 years of age. We included both healthy individuals and cancer patients, as genetic testing HBOC and Lynch syndrome also provides cancer patients with predictive information about risks of other cancer types. Both acceptors and decliners of genetic testing were eligible. Participants were recruited from several German-speaking university hospitals in Switzerland through their counselling physicians. We conducted the interviews between September 2017 and January 2019.

Following the grounded theory approach (Corbin and Strauss 1990), we first openly coded the interviews "line by line", and started building concepts by combining similar codes. Data analysis was an ongoing process, and we adapted the interview guide and refined the inclusion

1 See chapter 3.

criteria during the course of the study to reach theoretical saturation. We constantly compared codes within and in-between interviews and among each other. At an advanced stage of analysis, we started to interrogate the data to maintain our openness towards rebuilding and discarding existing concepts. Throughout the analysis process, we wrote memos and exchanged our interpretations with each other. For this part of the study, we systematically examined our concepts and categories for their connection to relational and individualistic aspects of autonomous decision-making.

The first author (BZ) conducted all interviews in either German, Swiss-German dialect, or French. The quotes were translated to English by BZ and proofread by IK and DS. The study was approved by the ethics committees of Northwest and Central Switzerland and Bern (ID number 2017-00316). All participating at-risk individuals signed an informed consent form before the interview.

5.4 Findings

We conducted eighteen interviews with counsellees who have a family history of cancer or were diagnosed with cancer at a young age themselves, and two with counselling doctors. Counsellees' age ranged from 27-70 years of age, fourteen of them were female, and twelve had blood-related children. Six of them had a cancer diagnosis when genetic testing was offered. In five cases, a pathogenic genetic variant was already diagnosed in a relative. Three participants refused genetic testing. We use five case stories from our sample to illustrate different aspects of social influence in the decision-making process (Box 5-1).

5.4.1 Responsibility towards relatives

The first aspect is the responsibility that at-risk individuals feel towards their relatives, particularly offspring. As illustrated by Mary's and Paul's (names are pseudonyms) case stories (Box 5-1), some participants even valued their children's interests higher than their own and agreed to a genetic test even though they were not convinced about its usefulness for themselves. Even some of the childless participants emphasised the importance of predictive genetic testing for offspring, criticising family members with children who were not interested in predictive genetic testing. Some participants tried to persuade family members to consider testing for their offspring:

> *"I think it would be useful that my sisters would… sacrifice themselves for their children and get tested, to just know for certain, to protect their children, because children are the greatest good for a family, in my opinion." (Jack, 50+, carrier in remission)*

Box 5-1: Case stories illustrating aspects of autonomous decision-making.

Case 1 – Mary: Doing predictive genetic testing for her daughter.

One of Mary's grown up daughters underwent genetic counseling because of the family's cancer history. Since Mary's affected sister refused genetic testing, Mary agreed to do it for her daughters. She had been aware that cancer was common in her family, but had always refrained from predictive genetic testing. "For myself, if my daughter hadn't come to me because she wanted to know, I wouldn't have had the test. I really primarily did it for my daughters." (Mary, 60+, healthy non-carrier)

Case 2 – Paul: Torn between his own interests and the interests of his daughter.

Paul is affected by cancer, and since his mother was, too, a genetic test was recommended. He initially agreed, thinking mainly about the interests of his daughter, but started to brood while waiting for the test result, doubting that knowing an increased genetic risk would do him any good. "And sometimes I started to brood, and started thinking, what consequences will this test result have for me… And then I switched sides and thought, no, it's good to know that, for my daughter it makes sense, she is young, she has children, and so on. And then you think about yourself again… […] My wife and daughter really took sides and said, listen, it's not just for you. It's for your daughter as well. And I think it is also reasonable for me to do it. […] So no, I wouldn't have stopped the testing process, that would have been too selfish. And: it wouldn't have left me alone. […] Not doing it would have made me very insecure." (Paul, 60+, non-carrier in remission)

Case 3 – Margaret: Perceiving genetic testing as her own private matter.

After genetic counseling, Margaret, who is healthy and childless, decided not to undergo predictive genetic testing despite many cases of cancer in her family. For her, a positive test result would not have resulted in any actionable consequences, just in anxiety and worries. Margaret described her difficulties in asking her family members for information she needed for her pedigree: "My goal would have been to talk to my relatives in a small circle, and that got a bit out of control. But they don't know anything about my decision now, that is my personal thing, my decision. My friend knows about it, but I haven't told anyone else, that is really my personal thing, yes." (Margaret, 30+, healthy, untested)

Case 4 – Ruth: Doing predictive genetic testing to belong.

Because Ruth's cousin and sister are carriers of a pathogenic genetic variant, they urged Ruth to undergo predictive genetic testing, too. Ruth at first perceived this as unnecessary, since she intended to take preventive measures in any case. However, after a while she felt like she needed to know anyway: "My sister, my cousin and I, we are like a fate community now, and that's good, because now we know we all have the same, we can talk about it, […] we are really on the same boat now. Eh, but it was like, if I didn't get tested, I couldn't have taken part in certain talks, I wouldn't have known if I have the mutation or not… […] Ehm, and that's why I actually wanted to know, too. And also to know, am I in their group or am I, am I outside." (Ruth, 40+, healthy carrier)

Case 5 – Anne: Listening to the doctor's advice.

Anne is affected by cancer, like her sister many years ago, and her treating physician recommended predictive genetic testing for a cancer predisposition. Even though Anne was convinced that the test would come back negative (based on her family history) she did not want to go against her treating physician and agreed to do the test. Her test result revealed a variant of unknown significance, but Anne was not interested in the meaning of this: "[My physician] simply said that the test result was somehow, eh, just so that one would classify it as negative, eh... as negative, not positive... But apparently they found something they still cannot decipher today, and they might be able to in a few years, but that was when I noticed that it was getting too complicated for me. I do not want to know all that at all, it seems to me one ends up like running around and thinking, eh, what is slumbering inside me, so... At that point I realized that that is ok for me." (Anne, 60+)

This focus on the interest of family members sometimes led to dilemma situations if family members had competing interests. For example, Mary explained how her second daughter refused to learn about the predictive genetic test result while her first daughter wanted to know her genetic risk:

> *"And one of my other daughters said she didn't want to know anything about it, that she didn't want to know my test result […] And because I suspected that she didn't want to know anything about that, there was some ambiguity, I had to inform her that I intended to do such a test, but in fact, she really doesn't want to know all that. That is just a tricky situation." (Mary, 60+, healthy non-carrier)*

5.4.2 Healthcare professionals' influence

Secondly, participants mentioned the role of healthcare professionals in their decision-making process. As in the case of Paula, physicians usually recommended predictive genetic testing if it influenced cancer treatment. However, healthy participants also described how medical professionals influenced their interest in predictive genetic testing on different levels:

> *"Because my gynaecologist had retired, I had to look for someone new at that time and I think that was an input for me somehow; it's a young practice, and it came up, also from her side; that she would like to send me there [to genetic counselling]. Ehm, not necessarily to do the [genetic] test but just so that she would know a little bit more about how to organise [cancer] prevention, even if I didn't do the test. [...] And then I thought, yes, I'll go to genetic counselling. And listen to what they say." (Jakobia, 30+, healthy, untested)*

> *"During the genetic consultation, I knew quite quickly that I wanted to be tested. [The genetic counsellor] rather tended towards me doing it, he also wrote down my family history and said he would recommend that I do it. That it was up to me, but, ehm, that I wouldn't block myself from anything." (Fabia, 50+, healthy carrier)*

One participant explained how she and her counselling doctor decided together how to proceed with genetic testing:

> *"She [the counselling physician] is really passionate, she's really interested... And... she speaks frankly with me, openly. So I don't feel like she's hiding things from me or making decisions for me [...] She was prepared, she had already thought about it herself, okay, we're looking for other [genes], which ones, she had already done all the research according to what the geneticist had given her, and what she knew. So she was ready to offer me... so that we could discuss [which genes to test further], the options we had." (Helen, 30+, non-carrier in remission)*

Even though none of the participants explicitly expressed that they were treated or counselled paternalistically, healthcare professionals considerably influenced their decision-making process. Still, healthcare professionals related to the principle of nondirective counselling:

> *"Ideally, genetic counselling should be nondirective […] Nondirectiveness concerns the decision regarding genetic testing itself rather than recommendations for cancer prevention. The latter for me has nothing to do with nondirectiveness. Nondirectiveness mainly concerns the genetic test. To say one should absolutely do genetic testing. Because there should be no such thing, shouldn't it, there are good individual reasons to reject a genetic test." (geneticist 1)*

5.4.3 Individualistic decisions and social relationships

Another aspect participants described was the tension between individualistic reasons (not) to perform genetic testing and their social relationships. Not all at-risk individuals took relational aspects into account in their decision-making process. Margaret, for example, based her decision against predictive genetic testing on individualistic reasons. However, she and another participant refusing genetic testing expressed difficulties talking about their decision to family and friends because they feared the judgment of others:

> *"I am reluctant to discuss my decision with others. Because they all have their history, their opinions… I'd be afraid someone would try and persuade me, influence me, and judge me…" (Michaela, 40+, healthy, untested)*

Moreover, as Paul's case illustrates, some at-risk individuals had contradictory feelings because, from an individualistic perspective, they would prefer not to know their genetic risk, but they felt an obligation towards their offspring to agree to predictive genetic testing. Paul said that once he was informed about the option of predictive genetic testing, he felt obliged to do it (Box 5-1).

Social influence was often implicit and did not influence all individuals to the same extent. As Eva's case illustrates (Box 5-1), knowing one's genetic risk can be a means to know in which group ("positive" or "negative") one belongs. While Eva's description could refer to implicit social pressure, she perceived her decision to have predictive genetic testing as autonomous. One of the geneticists confirmed that social influence was a common phenomenon that he frequently discussed in genetic counselling:

> *"What is not actually perceived [by counsellees], but what we also address in predictive testing is, um, it's actually a well-known phenomenon that if whole families get tested [...] then, of course, there are those who have it and those who don't. And [...] there is a bit of grouping in that sense. That those who have it are more likely to stick together at family gatherings." (geneticist 1)*

Several participants stated that it was important to them that their grown-up children had a genetic test. Some participants even admitted having nudged or persuaded their adult children to be tested. This was especially the case for participants who perceived predictive genetic testing as beneficial and who found it difficult to understand why someone would refuse it.

> *"I think my daughters just got tested because I said so (laughs). I don't think they knew what that meant at the beginning. Because it went so fast… I think I overruled them a little bit, to be honest. Well, I don't regret it, I think it is important." (Rose, 50+, carrier in remission)*

5.5 Discussion

We interviewed people in Switzerland undergoing genetic counselling for a predisposition to cancer (Hereditary Breast and Ovarian Cancer syndrome or Lynch Syndrome) about their decision-making process and analysed how social relationships influenced their decisions as part of a grounded theory study.[2] Our findings indicate that both relational and individualistic reasons influence decision-making, and in both cases, individuals might face implicit or explicit social pressure. The following paragraphs explore the theoretical and practical implications for autonomous decision-making.

5.5.1 External influences allowing for autonomous decision-making

In line with previous studies, we show that family considerations can dominantly influence predictive genetic testing decision-making (Hallowell et al. 2003; Hallowell et al. 2006; d'Agincourt-Canning 2006; Etchegary et al. 2009; Foster et al. 2002). Additionally, our results suggest that the wish of belonging to a social group can be a decisional factor. In both cases, decisions are still autonomous when applying a relational conception of autonomy. The impact of relational aspects in genetic testing decision-making has been increasingly recognised (Dove et al. 2017; Gilbar and Barnoy 2018). Still, clinical guidelines only superficially reflect on this aspect, as they only superficially recommend making psychosocial aspects and communication to the family a subject of discussion (Skirton et al. 2013; Deutsche Gesellschaft für Humangenetik e.V. and Berufsverband Deutscher Humangenetiker e.V. 2018). Given the importance of relational aspects in genetic testing decision-making, we think this should be explicitly mentioned in clinical guidelines.

2 See chapter 3.

Some participants felt torn between their feeling of responsibility towards relatives and their own interest. This feeling of inner conflict illustrates the potentially contradicting elements of relational and individualistic aspects in genetic testing decision-making. Such cases are particularly delicate and need special attention to ensure autonomous decision-making, as those individuals might be particularly susceptible to undue influences from their social environment. If family members accompany at-risk individuals to genetic counselling, it might be helpful having an additional session without family members present.

Previous studies also revealed that healthcare professionals influence genetic testing decision-making by the way they frame information (Scott, D. et al. 2019). This can support autonomous decision-making if counsellees are carefully guided in their decision-making process and adequate information is provided (Manson and O'Neill 2007). Relational autonomy, as well as theories on shared decision-making, support this approach (White 1998; Osuji 2018). However, this renders the principle of nondirectiveness impractical. While these considerations have been discussed at length in the past decade (Evans et al. 2004; Pennacchini and Pensieri 2011; Weil et al. 2006), Swiss law and German guidelines still hold on to this principle (Swiss Federal Council 2004, Art 14; Deutsche Gesellschaft für Humangenetik e.V. and Berufsverband Deutscher Humangenetiker e.V. 2018, section 1.5).

5.5.2 Undue social pressure preventing autonomous decision-making

We observed that decliners of genetic testing did not want to justify their decision towards others. They perceived their decision as concerning only their own life and thus made it based on individualistic reasons only. Besides, a previous study showed that decliners might also anticipate negative consequences for their relatives, which to them was another reason to decline (d'Agincourt-Canning 2006). While decliners perceived that their decision needed social justification, those agreeing to genetic testing for individualistic reasons did not express such social pressure. This indicates that declining genetic testing might provoke social pressure. The implicit nature of this pressure is problematic, as it might leave some individuals with the feeling of having no alternative but to test. To prevail true freedom of choice, which we think is crucial in predictive genetic testing, social acceptance to refuse such testing should be improved. As mass media coverage has a predominantly positive narrative when it comes to genetic testing for actionable diseases (Zimmermann, Elger, and Shaw 2019), reasons to decline should be made more transparent in the public discourse.

Additionally, some participants expressed their incomprehension about family members who refused genetic testing. These participants had a relational perception of genetic testing and perceived genetic testing for cancer predisposition as beneficial. This indicates a potential dispute between at-risk individuals with an individualistic and those with a relational conception regarding genetic testing decision-making. In some cases, this even led to coercive situations, which has also been reported and discussed in previous studies with test takers (Etchegary et al. 2009) and healthcare professionals (Gilbar and Barnoy 2018). Anita Ho argued that healthcare professionals should first carefully examine the family context and talk to the patient and family before making a hasty judgement about family coercion (Ho 2008). We propose that genetic counsellors should make counsellees proactively aware of the possibility that relatives might invoke their right not to know. This is especially important if people display unreflected approval of any genetic testing. Because our interviews revealed that most potentially problematic situations of social pressure arose out of unawareness, reflecting on the situation in genetic counselling might already improve this issue.

5.5.3 A model to illustrate social influences in PGT decision-making

To illustrate the role of individualistic and relational reasons in predictive genetic testing decision-making, we propose an integrating model of the aspects of social influence on autonomous predictive genetic testing decision-making (Figure 5-1). This model reflects the empirical data in this context and is an extension of both the principle of respect for autonomy of Beauchamp and Childress (2013) and the conceptions of relational autonomy proposed by Friedman (2003) and Christman (2009). We propose that there is a continuum between individualistic and relational aspects of decision-making and that individuals place themselves on this continuum for their decision-making regarding predictive genetic testing. Where they place themselves on

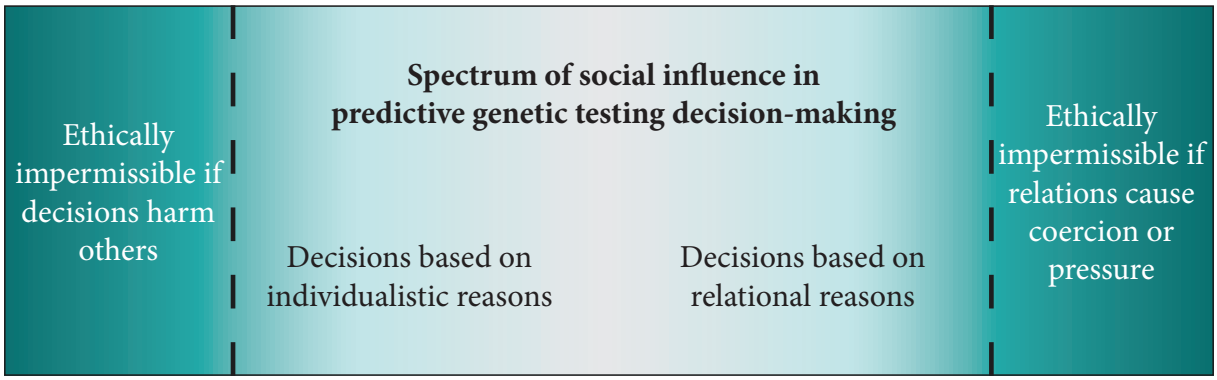

Figure 5-1: Model on the social influence in predictive genetic testing decision-making.

this continuum is context-dependent. Contextual factors can include the family situation, the character of the person, the cultural background, and other aspects. Individuals should not be unduly influenced when positioning themselves on the continuum. Furthermore, the model sees autonomy as a matter of degree, following Beauchamp and Childress (2013) and Friedman (2003). Consequently, there is a threshold when a decision is not autonomous anymore. This threshold is also context-dependent: it depends on the specific decision at stake, but also on cultural settings, as in some non-Western cultures where autonomy is to a much larger extent associated with the family's well-being.

5.5.4 Generalizability and limitations

The data informing the model we are proposing are based on considerations regarding predictive genetic testing for HBOC and Lynch syndrome. Previous studies on predictive genetic testing decision-making for Huntington's disease, where to date no meaningful medical actions are available, show that relational considerations (especially regarding offspring and other relatives) also influence decision-making to different degrees (Ibisler et al. 2017; Rivera-Navarro, Cubo, and Mariscal 2015). While the influencing role of healthcare professionals might differ from our findings, we propose that our model generally can be extended to such applications of predictive genetic testing, too. As the family has a distinct role in predictive genetic testing decision-making where the predisposition is heritable, applying the here-proposed model to other contexts would need further investigation.

This study was conducted in Switzerland, where genetic testing decision-making has not been investigated before. Swiss law requires that predictive genetic testing is covered by health insurance if medically indicated (Swiss Federal Council 1994, Art 26); "non-directive" genetic counselling is mandatory before and after predictive genetic testing (Swiss Federal Council 2004, Art 14), and medical doctors are responsible for providing such counselling. As such, the context differs from well-investigated countries, such as the United States or the United Kingdom.

We emphasise that our model focuses solely on how to enhance autonomous decision-making, focusing on social influence, but autonomy can legitimately be restricted if it comes into conflict with other ethical principles, for example, if the autonomous action of a person hurts other persons (Beauchamp and Childress 2013). Besides, our model operates under the premise that there are true choices available for individuals regarding predictive genetic testing decision-making, excluding other factors such as funding constraints.

Finally, this study had some methodological limitations.Reports about persuasion and coercion

mostly came from third-party descriptions and often did not concern the person interviewed, but his or her family members. This is because people refusing genetic testing were challenging to reach with our sampling strategy, which did not allow for snowballing due to ethical considerations. Those who participated and refused genetic testing still at least considered testing as an option, but our study lacks participants who refuse even to learn about the option of predictive genetic testing. Also, participants explained their decision-making process retrospectively, and their perception might have changed unconsciously after learning the outcome of the test. Still, our data set is rich in a variety of contexts and opinions and our results are in line with previous studies, which increases the credibility of our empirical findings. As with every secondary analysis, our primary purpose was not to collect information on relational aspects of autonomous decision-making; instead, they occurred spontaneously in our data.

5.5.5 Conclusion

This study presented a qualitative analysis of Swiss genetic counsellees, analysing social influences in decision-making for cancer predisposition genetic testing. We suggested that both relational and individualistic reasons play a role in predictive genetic testing decision-making, which affects the conception of autonomous decision-making. While the principlist and relational conception of autonomy are competing concepts in the theoretical debate, they are two sides of the same medal when using them as lenses of analysis for predictive genetic testing decision-making. Still, we showed that those declining genetic testing based on individualistic reasons might face implicit or explicit social pressure and that some tested individuals might persuade family members to test out of a sense of duty. However, individuals should decide themselves how much their social environment influence their decision for or against predictive genetic testing. Genetic counsellors should raise awareness of these issues. At the same time, public debates should focus more on the pros and cons of predictive genetic testing, avoiding positive bias.

5.6 References

Abacan, MaryAnn, Lamia Alsubaie, Kristine Barlow-Stewart, Beppy Caanen, Christophe Cordier, Eliza Courtney, Emeline Davoine et al. 2019. "The Global State of the Genetic Counseling Profession." Eur J Hum Genet 27 (2): 183–97. doi:10.1038/s41431-018-0252-x.

Andorno, Roberto. 2004. "The Right Not to Know: An Autonomy Based Approach." J Med Ethics 30 (5): 435-9; discussion 439-40. doi:10.1136/jme.2002.001578.

Beauchamp, Tom L., and James F. Childress. 2013. Principles of Biomedical Ethics. 7th ed. New York: Oxford University Press.

Bundesgesetz Über Die Krankenversicherung. KVG. Swiss Federal Council. 1994. Accessed December 23, 2019. https://www.admin.ch/opc/de/classified-compilation/19940073/index.html.

Christman, John. 2004. "Relational Autonomy, Liberal Individualism, and the Social Constitution of Selves." Philos Stud 117 (1): 143–64. doi:10.1023/B:PHIL.0000014532.56866.5c.

———. 2009. The Politics of Persons : Individual Autonomy and Socio-Historical Selves. Cambridge: Cambridge University Press.

Corbin, Juliet M., and Anselm Strauss. 1990. "Grounded Theory Research: Procedures, Canons, and Evaluative Criteria." Qual Soc 13 (1): 3–21.

d'Agincourt-Canning, Lori. 2006. "Genetic Testing for Hereditary Breast and Ovarian Cancer: Responsibility and Choice." Qual Health Res 16 (1): 97–118. doi:10.1177/1049732305284002.

Deutsche Gesellschaft für Humangenetik e.V., and Berufsverband Deutscher Humangenetiker e.V. 2018. "S2k-Leitlinie Humangenetische Diagnostik Und Genetische Beratung." medgen 30 (4): 469–522. doi:10.1007/s11825-018-0223-1.

Donchin, Anne. 2001. "Understanding Autonomy Relationally: Toward a Reconfiguration of Bioethical Principles." J Med Philos 26 (4): 365–86. doi:10.1076/jmep.26.4.365.3012.

Dove, Edward S., Susan E. Kelly, Federica Lucivero, Mavis Machirori, Sandi Dheensa, and Barbara Prainsack. 2017. "Beyond Individualism: Is There a Place for Relational Autonomy in Clinical Practice and Research?" Clinical Ethics 12 (3): 150–65. doi:10.1177/1477750917704156.

Etchegary, Holly, Fiona Miller, Sonya deLaat, Brenda J. Wilson, June C. Carroll, and Mario Cappelli. 2009. "Decision-Making About Inherited Cancer Risk: Exploring Dimensions of Genetic Responsibility." J Genet Couns 18 (3): 252–64. doi:10.1007/s10897-009-9218-z.

Evans, Marilyn, Vangie Bergum, Stephen Bamforth, and Sandra MacPhail. 2004. "Relational Ethics and Genetic Counseling." Nurs Ethics 11 (5): 459–71. doi:10.1191/0969733004ne724oa.

Foster, Claire, Maggie Watson, Clare Moynihan, Audrey Ardern-Jones, and Rosalind Eeles. 2002. "Genetic Testing for Breast and Ovarian Cancer Predisposition: Cancer Burden and Responsibility." J Health Psychol 7 (4): 469–84. doi:10.1177/1359105302007004627.

Friedman, Marilyn. 2003. Autonomy, Gender, Politics. Studies in feminist philosophy. Oxford: Oxford University Press.

Frith, Lucy. 2012. "Symbiotic Empirical Ethics: A Practical Methodology." Bioethics 26 (4): 198–206. doi:10.1111/j.1467-8519.2010.01843.x.

Gilbar, Roy, and Sivia Barnoy. 2018. "Companions or Patients? The Impact of Family Presence in Genetic Consultations for Inherited Breast Cancer: Relational Autonomy in Practice." Bioethics 32 (6): 378–87. doi:10.1111/bioe.12448.

Godino, Lea, Leigh Jackson, Daniela Turchetti, Catherine Hennessy, and Heather Skirton. 2018. "Decision Making and Experiences of Young Adults Undergoing Presymptomatic Genetic Testing for Familial Cancer: A Longitudinal Grounded Theory Study." Eur J Hum Genet 26:44–53. doi:10.1038/s41431-017-0030-1.

Hallowell, Nina, Audrey Ardern-Jones, Ros Eeles, Claire Foster, Anneke Lucassen, Clare Moynihan, and Maggie Watson. 2005. "Men's Decision-Making About Predictive BRCA1/2 Testing: The Role of Family." J Genet Couns 14 (3): 207–17. doi:10.1007/s10897-005-0384-3.

———. 2006. "Guilt, Blame and Responsibility: Men's Understanding of Their Role in the Transmission of BRCA1/2 Mutations Within Their Family." Sociol Health Illn 28 (7): 969–88. doi:10.1111/j.1467-9566.2006.00515.x.

Hallowell, Nina, Claire Foster, Ros Eeles, Audrey Ardern-Jones, V. Murday, and Maggie Watson. 2003. "Balancing Autonomy and Responsibility: The Ethics of Generating and Disclosing Genetic Information." J Med Ethics 29 (2): 74-9; discussion 80-3. doi:10.1136/jme.29.2.74.

Hamilton, Rebekah J., and Barbara J. Bowers. 2007. "The Theory of Genetic Vulnerability: A Roy Model Exemplar." Nurs Sci Q 20 (3): 254–64. doi:10.1177/0894318407303127.

Ho, Anita. 2008. "Relational Autonomy or Undue Pressure? Family's Role in Medical Decision-making." Scand J Caring Sci 22 (1): 128–35. doi:10.1111/j.1471-6712.2007.00561.x.

Ibisler, Aysegül, Sebastian Ocklenburg, Susanne Stemmler, Larissa Arning, Jörg T. Epplen, Carsten Saft, and Sabine Hoffjan. 2017. "Prospective Evaluation of Predictive DNA Testing for Huntington's Disease in a Large German Center." J Genet Couns 26 (5): 1029–40. doi:10.1007/s10897-017-0085-8.

Jennings, Bruce. 2016. "Reconceptualizing Autonomy: A Relational Turn in Bioethics." Hastings Cent Rep 46 (3): 11–16. doi:10.1002/hast.544.

Llort, G., I. Chirivella, R. Morales, R. Serrano, A. B. Sanchez, A. Teulé, E. Lastra, J. Brunet, J. Balmaña, and B. Graña. 2015. "SEOM Clinical Guidelines in Hereditary Breast and Ovarian Cancer." Clin Transl Oncol 17 (12): 956–61. doi:10.1007/s12094-015-1435-3.

Lynch, Henry T., Thomas Smyrk, and Jane Lynch. 1997. "An Update of HNPCC (Lynch Syndrome)." Cancer Genet Cytogen 93 (1): 84–99. doi:10.1016/S0165-4608(96)00290-7.

Mackenzie, Catriona, and Natalie Stoljar, eds. 2000. Relational Autonomy: Feminist Perspectives on Automony, Agency, and the Social Self. New York: Oxford University Press.

Manson, Neil, and Onora O'Neill. 2007. Rethinking Informed Consent in Bioethics. Cambridge: Cambridge University Press.

Osuji, Peter I. 2018. "Relational Autonomy in Informed Consent (RAIC) as an Ethics of Care Approach to the Concept of Informed Consent." Med Health Care Philos 21 (1): 101–11. doi:10.1007/s11019-017-9789-7.

Pennacchini, M., and C. Pensieri. 2011. "Is Non-Directive Communication in Genetic Counseling Possible?" Clin Ter 162 (5): e141-144.

Rivera-Navarro, Jesús, Esther Cubo, and Natividad Mariscal. 2015. "Analysis of the Reasons for Non-Uptake of Predictive Testing for Huntington's Disease in Spain: A Qualitative Study." J Genet Couns 24 (6): 1011–21. doi:10.1007/s10897-015-9840-x.

Scott, Danika, Sue Friedman, Melinda L. Telli, and Allison W. Kurian. 2019. "Decision Making About Genetic Testing Among Women with a Personal and Family History of Breast Cancer." J Oncol Pract, JOP1900221. doi:10.1200/JOP.19.00221.

Skirton, Heather, Lesley Goldsmith, Leigh Jackson, and Aad Tibben. 2013. "Quality in Genetic Counselling for Presymptomatic Testing — Clinical Guidelines for Practice Across the Range of Genetic Conditions." Eur J Hum Genet 21 (3): 256–60. doi:10.1038/ejhg.2012.174.

Stoffel, Elena M., Pamela B. Mangu, Stephen B. Gruber, Stanley R. Hamilton, Matthew F. Kalady, Michelle W. Y. Lau, Karen H. Lu, Nancy Roach, and Paul J. Limburg. 2015. "Hereditary Colorectal Cancer Syndromes: American Society of Clinical Oncology Clinical Practice Guideline Endorsement of the Familial Risk–colorectal Cancer: European Society for Medical Oncology Clinical Practice Guidelines." J Clin Oncol 33 (2): 209. doi:10.1200/JCO.2014.58.1322.

Stoljar, Natalie. 2018. "Feminist Perspectives on Autonomy." In the Stanford Encyclopedia of Philosophy, edited by Edward N. Zalta. Winter 2018: Metaphysics Research Lab, Stanford University. https://plato.stanford.edu/entries/feminism-autonomy/. Accessed October 14, 2019.

Swiss Federal Council. 2004. "CC 810.12 Federal Act of 8 October 2004 on Human Genetic Testing (HGTA)." Accessed January 23, 2020. https://www.admin.ch/opc/en/classified-compilation/20011087/index.html.

Weil, Jon, Kelly Ormond, June Peters, Kathryn Peters, Barbara B. Biesecker, and Bonnie LeRoy. 2006. "The Relationship of Nondirectiveness to Genetic Counseling: Report of a Workshop at the 2003 NSGC Annual Education Conference." J Genet Couns 15 (2): 85–93. doi:10.1007/s10897-005-9008-1.

White, Mary T. 1998. "Decision-Making Through Dialogue: Reconfiguring Autonomy in Genetic Counseling." Theor Med Bioeth 19 (1): 5–19. doi:10.1023/a:1009976119490.

Zimmermann, Bettina M., Bernice S. Elger, and David Shaw. 2019. "Media Coverage of Ethical Issues in Predictive Genetic Testing: A Qualitative Analysis." AJOB Empirical Bioethics 10 (4): 250–64. doi:10.1080/23294515.2019.1670275.

Chapter 6: Influence of content, events and culture on the public discourse about medical genetics in Switzerland – a quantitative media content analysis

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6.1 Abstract

Medical genetics is a broad and emerging field with many important implications for society, but knowledge about media coverage of this topic from recent years is lacking. This study aims to identify topics in medical genetics emerging in print media coverage in Switzerland by quantitatively analysing their occurrence in the public media discourse, and to assess culturally conditioned differences between Swiss language regions. We conducted a quantitative media content screening of print media and news agencies in the German- and French-speaking regions of Switzerland, and eight topics were identified. They demonstrate the large variety of topics in medical genetics present in the public discourse. Coverage was dominated by political votes and Angelina Jolie's preventive surgeries. We found only small differences between the language regions, and coverage was strikingly similar for most variables. This study gives an up-to-date quantitative overview of how print media cover different topics within the broad field of medical genetics. Because of the large variety of topics in the field, distinguishing between them is important for public discussions and further research.

6.2 Introduction

Media have a significant impact on the attitudes, perceptions and education of the public about science in general (Schäfer 2012) and genetics in particular (Condit 2001; Eyck and Williment 2003). They are the public's main source of information about genetics apart from school education (Garrett and Bird 2007). Assessing public debates and improving information provision and evidence-based discussion thus is crucial for democratic societies to be able to deal with upcoming decisions and policymaking (Dahlgren 2009).

The field of medical genetics is broad, ranging from basic research to practical clinical applications (Epstein 2006), and it is rapidly expanding due to technological advancements (Mei et al. 2016). These developments lead to both opportunities and risks, but conclusions relevant to policymaking are dependent on specific aspects within the field of medical genetics (Bunnik, Schermer, and Janssens 2011; Burke et al. 2015) and also require certain biological knowledge. Because of this, Kung and Gelbart have postulated a need for improved genetics education in high schools (Kung and Gelbart 2012). Citizens have a large impact on policymaking and public debates thus are of high intensity and importance, particularly in Switzerland with its political system of direct democracy.

With respect to the breadth and variety of the field of medical genetics, there is a lack of information about which topics within this field are picked up by mass media. Assessing mass media

coverage about medical genetics is important because it indicates what information the public is exposed to, and is considered an indicator of public debates, the level of public interest and its likely agenda (Kolb 2005). This paper, therefore, quantitatively investigates the content concerning medical genetics featured in print media and news agencies in the German- and French-speaking parts of Switzerland in the years 2013 to 2016. Three research questions are examined: (1) How many and which aspects of medical genetics were covered by print media and news agencies in Switzerland? (2) What factors triggered media coverage about different aspects of medical genetics? (3) What were the differences in coverage between German- and the French-speaking print media? Regarding the last research question, we hypothesized that there were statistically significant differences regarding amount and type of coverage, because of cultural differences in attitudes towards medical genetics, as mirrored in the results of recent political votes (Ballmer-Cao et al. 2016).

In the next section, we review the current literature on issue-attention cycles and science communication and provide some theoretical background. We will then briefly explain the methodology before answering our research questions in the results and discussion section.

6.3 Literature Review

Many studies have investigated coverage about genetic technologies in the 1990s and early 2000s (e.g., Görke, Kohring, and Ruhrmann 2000; Kohring and Matthes 2002; Leonarz 2002). In Germany, studies in agricultural genetics were predominant throughout the 1990s, whereas biomedical health applications of genetics only came into focus in the late 90s, with discussions around cloning and the Human Genome Project (Kohring and Matthes 2002). Cloning, stem cell and embryo research, in-vitro fertilisation and preimplantation genetic diagnosis (PGD) were the emerging topics in German TV programs in 2008-2009 (Ruhrmann and Milde 2011). The most recent study focused on media coverage after Angelina Jolie's announcement that she had a double mastectomy (Jolie, May 13, 2013). According to a media content analysis about this topic, conducted in 2013 for Canada, the USA and the UK, print media mostly reported positively about Angelina Jolie's decision to have a mastectomy but did not provide sufficient contextual information such as the rarity of her genetic condition (Kamenova, Reshef, and Caulfield 2014). Accordingly, a US survey revealed that while the majority of people were aware of Angelina Jolie's preventive surgery, only a small ratio gained more profound knowledge about her genetic risk (Borzekowski et al. 2014).

6.3.1 Issue-attention cycles and science communication

In this paper, we largely follow the research tradition of issue-attention cycles by investigating how media coverage develops over time. The issue-attention cycle theory claims that public interest in emerging issues has a cyclic development: first, an existing issue has not yet captured public attention, but one or several events suddenly increase public attention, so solutions need to be found by responsible stakeholders. Once the issue is addressed, and solutions are found, public interest in the issue will gradually decline (Downs 1972). Media coverage is considered a good indicator of the public's interest and attention and follows similar cycles (Kolb 2005). Many longitudinal media content analyses on genetics have been performed to confirm such a cyclic pattern (e.g., Görke, Kohring, and Ruhrmann 2000; Racine et al. 2006; Rödder 2005).

Gerhards and Schäfer (2007) compared coverage about human genome research in Germany and the US from 1999 to 2001, following the tradition of science communication. They found that in both countries newspaper coverage was similar in the sense that it was centred on events, commonly triggered by scientists, more positive in the assessment of genetic applications and mostly about scientific and medical implications (Gerhards and Schäfer 2006).

6.3.2 Attitudes to and coverage of genetics in Switzerland

The Swiss public's attitudes towards genetics have fluctuated repeatedly in the last 25 years. Media coverage increased mainly in the context of political votes, but attitudes did not necessarily become more positive towards genetics when media coverage increased (Bonfadelli, Dahinden, and Leonarz 2002). In the context of a controversial vote in 1998 on whether to prohibit genetically modified animals and plants in Switzerland (Swiss Federal Council 1998), the media coverage increased sharply and was greater and more controversial than in other European countries (Leonarz 2002). Both positive and negative statements were found more often and in more depth in the Swiss media coverage compared to other countries, and negative aspects were more differentiated than positive ones (Leonarz 2002). Concurrently, public attitudes in Switzerland changed from being sceptical to being rather positive towards genetics, an outcome Bonfadelli et al. (2002) interpreted as the result of a highly effective political campaign. Only a couple of years later, however, the public attitude had again changed to be rather sceptical, as demonstrated by a population-wide survey (Bonfadelli, Dahinden, and Leonarz 2002) and another political vote in November 2005, where a moratorium on importing and introducing genetically modified plants and animals in Switzerland was accepted (Swiss Federal Council 2005). In the latest relevant political votes, the Swiss people accepted the use of PGD for severe diseases (Swiss Federal Council 2015, 2016).

6.4 Materials and methods

The study period was 1 January 2013 to 31 December 2016 and covered the German and French language media in Switzerland. We chose the print media and the news agencies to represent mass media for two reasons. First, the print media often set the agenda for other mass media types (online platforms, TV and radio) and are frequently cited by them (Jarren and Vogel 2011). Second, print media allow for more reliable article selection than other mass media, including online media, where published content can be altered or removed easily. Thus, to assess the predominance of medical genetic topics present in public discourse, the print media and the news agencies as their most important sources are considered eligible.

Since we aimed to compare coverage in the German- and French-speaking parts of Switzerland, we chose the newspapers according to the functional equivalence approach (Wirth and Kolb 2004), meaning that we included newspapers fulfilling similar functions from each language region (Table 6-1). We chose the print media with the highest numbers of readers according to a media-independent Swiss survey (WEMF 2016) within each language-specific functional group because we considered these most important in the public debate. Sources to sample relevant articles are summarized in Table 6-1.

Table 6-1: Sampling of newspapers and article sampling tool.

Functional group	Newspapers German	Newspapers French	Sampling tool
Tabloid	Blick	Le Matin	Factiva
Freesheet	20 Minuten	20 minutes	www.20min.ch, pdf search
Broadsheets	*Neue Zürcher Zeitung* (NZZ)	Le Temps	Factiva
	TagesAnzeiger (TA)	La Tribune de Genève (LTG)	Factiva
News agency	sda	ats	LexisNexis

Note: According to the functional equivalence approach, we chose media that fulfilled similar functions in the German- and French-speaking parts of Switzerland, respectively. The sampling tools were chosen according to availability.

6.4.1 Data collection and reliability

The variables collected from each article were article length (word count), article title, date of publication and details about the medium in which the article was published (name, language, genre, column, author of the article). The article length was split into three groups for analysis: short notices (<200 words), news (200-800 words) and reports (>800 words), as an indicator of the content completeness of the coverage. Holtzman et al. (2005) have suggested that in the

English-speaking media, at least 800 words are necessary to have enough space to explain genetics as a complex topic complete and informative. Even though our analysis is based on French- and German-speaking articles, we adopted this threshold because journalistic articles are often counted in columns rather than words, and the same amount of words result in a similar amount of columns in all three languages. Thus, if newspapers include longer articles, they give these topics more value and include more information. This is confirmed by good journalistic practice, where it is taught that articles should be shortened from the end (Mast 2012).

We furthermore wanted to assess the content of coverage by determining the main topic of the articles. To our knowledge no current exhaustive and distinctive list of topics covering the whole field of medical genetics has been published, so we derived our own category system using an approach commonly used in quantitative media content analysis (MacQueen et al. 2009; Rössler 2010): We started with a list of topic categories that we derived from scientific literature and a preliminary content analysis through the Google search engine, and in parallel constructed German and French search algorithms with keywords covering these topic categories.

In the subsequent first round of sampling, we had 2444 search hits, of which 1829 articles met the inclusion criteria. We excluded double hits (same title, medium and publication date), press releases, content tables, glossaries and very short articles with one or two sentences only (no full story, no title) because we were interested in newspaper articles with journalistic content.

To ensure that the category system was valid, complete and selective, we used these 1829 articles to refine and improve our topic category list: all articles in the topic category "other" (459 articles) were reanalyzed, and the category system was expanded inductively with more subcategories as well as four more main categories. At the same time, we extended our search algorithms in order to capture full coverage of all emerging topics. They were used for the second round of article sampling, where we retrieved additional 479 units, 377 of which meeting the inclusion criteria.

Two researchers (BZ and FZ) then tested the list for further validation and to assess the interpersonal reliability of this topic category system. Both coded the main topic of the article in 194 randomly selected articles independently. Because our topic variable was nominal with many degrees of freedom, we used Scott's Pi to calculate the reliability score (Scott 2009). A reliability score higher than 0.7 is considered good, and we reached a score of 0.86 for the article topics. For publication, the first author (BZ) translated relevant article titles from German or French into the English language. DS double-checked the translation as a native English speaker.

6.4.2 Statistical analysis

Data collection and statistical analysis were performed with IBM SPSS statistics 24.0. For better comparability between the different topics, we normalized the data for language comparison using topic-specific Article Count Ratio (ACR), which is the number of articles counted per language region divided by the total number of articles within the topic-specific subsample.

We used Independent Samples t-test to compare article length in the French-speaking versus German-speaking media coverage. If the Levene-test for variance differences was significant ($p \leq 0.05$), we could not assume equal variance between the two language groups and thus used an Independent Sample t-test with Welch's correction. Because we performed multiple testing, Bonferroni-Holm correction was applied to control for the increased likelihood of a type I error.

6.5 Results and discussion

Data was collected for 2206 articles. Articles that did not treat any topic in the field of medical genetics in the title, subtitle or the first paragraph were excluded since they were not mainly about medical genetics (764 articles). Another 60 articles were double-registered, and 219 articles were not about medical genetics, but rather about forensic applications in crime, testing for familial relationships (e.g. paternity testing) and evolutionary or population-based genetics, genetic testing on animals or genetically modified plants or articles about non-reproductive medicine, indicating that many more aspects of genetics and genetic testing are covered in public discourse using the same keywords. This may make it even more complicated for nonspecialists to have an informed overview of and distinguish between the different aspects and applications of genetics and their specific implications and challenges.

A total of 1163 articles remained for subsequent statistical analysis, from which we identified eight topics that exhaustively and distinctively describe different content categories in the media coverage about medical genetics. Four articles (0.3%) could not be assigned to any of the presented topics and were assigned to the category "Other". We will now describe and discuss each of the topics and explain the most important characteristics we could derive from our data.

6.5.1 Preimplantation and prenatal genetic testing (*PPGT*): hot topic due to political votes

The most prevalent topic in our sample was *PPGT* (Figure 6-1). The majority (91%, N=465) of these articles were related to PGD (data not shown), where an in-vitro fertilized embryo is tested for genetic alterations prior to implantation in the mother's womb (Dahdouh et al. 2015). For

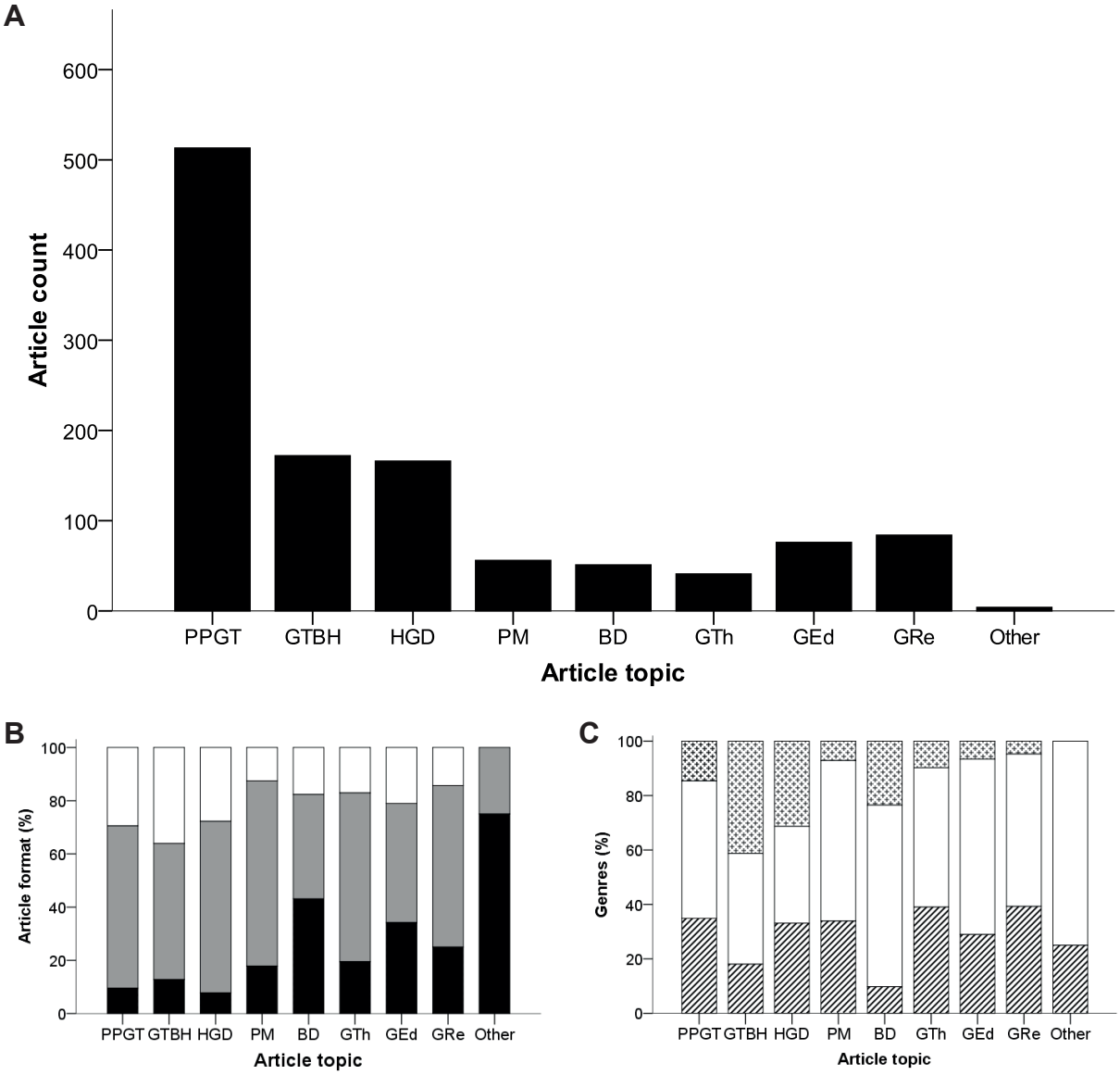

Figure 6-1: A) Frequency of the topics identified. B) Article format based on article length. Black = reports (>800 words); grey = news (200-800 words); white = notice (<200 words). C) Media genre ratio. Dashed = news agencies; white = broadsheets; crossed = tabloids (including cost-free press).

example, a headline in the French-speaking broadsheet newspaper Le Temps stated: "Pre-implantation genetic diagnosis: the open door to the 'improved' man" (Vögeli, May 28, 2015). The other articles assigned to this topic were about other or more general applications of genetics in reproductive medicine:

> *"Prenatal diagnosis: perfect babies soon?" (Beauté, September 07, 2013)*
>
> *" 'Women are not Petri dishes.' " (Brotschi, May 24, 2016)*

Regarding the development of coverage for *PPGT* over time, two peaks in May 2015 and May 2016 are conspicuous (Figure 6-2B). These are related to the two dates of public votes in Switzerland

about whether to permit PGD in June 2015 and June 2016. Between February and July 2015, 42% (N=217) of all *PPGT* articles were published, and between February and July 2016 another 115 articles (22% of *PPGT* coverage) were published, making the coverage regarding the second vote much lower compared to the first one. It seems that the topic was not assigned the same relevance and controversy, because it had already been covered and discussed the year before.

PPGT coverage was strongly connected to the two political votes, and given the predominance of *PPGT* when compared to the other topics, it shows that political topicality is the most potent driver for media coverage in medical genetics during the investigation period. A previous study already showed a similar development for a political vote related to genetics in 1998 (Bonfadelli, Dahinden, and Leonarz 2002). After July 2016, only four more articles about *PPGT* appeared, so the public debate diminished quickly once the political decision was final. We thus conclude that *PPGT* coverage in Switzerland was based more on political communication than on science communication.

Appositely, 90% of *PPGT* articles were news or short notices and only 10% were long reports providing background information (Figure 6-1B).

6.5.2 Genetic Testing on Born Humans (*GTBH*): celebrity boosts coverage

GTBH summarizes a high variety of articles about genetic testing on born humans. The articles were about the present and future applications of predictive genetic testing on born humans, for example, "Controversial Fertility Test leaves questions unanswered" (Straumann, March 22, 2013) and issues around direct-to-consumer genetic testing: "Genetic testing: Experts warn against excesses and charlatans" (sda, June 23, 2014).

However, the majority (52.9%) of *GTBH* articles were about Angelina Jolie's announcements that she had a double mastectomy (Jolie, May 13, 2013) and a hysterectomy (Jolie Pitt, March 25, 2015). This topic has been investigated in other countries previously and that has attracted the media's and the public's attention worldwide (Borzekowski et al. 2014; Kamenova, Reshef, and Caulfield 2014). We specifically searched for articles about Angelina Jolie's statement and its consequences in our search algorithms, and the actress' two announcements correspond well with the two peaks in coverage found for *GTBH* coverage (Figure 6-2C). The case of Angelina Jolie is a typical example of a key event that is interesting to cover for the media. For instance, Henderson and Kitzinger (1999) investigated British media coverage about breast cancer genetics, finding that "soft" topics such as case stories about hereditary breast cancer families and

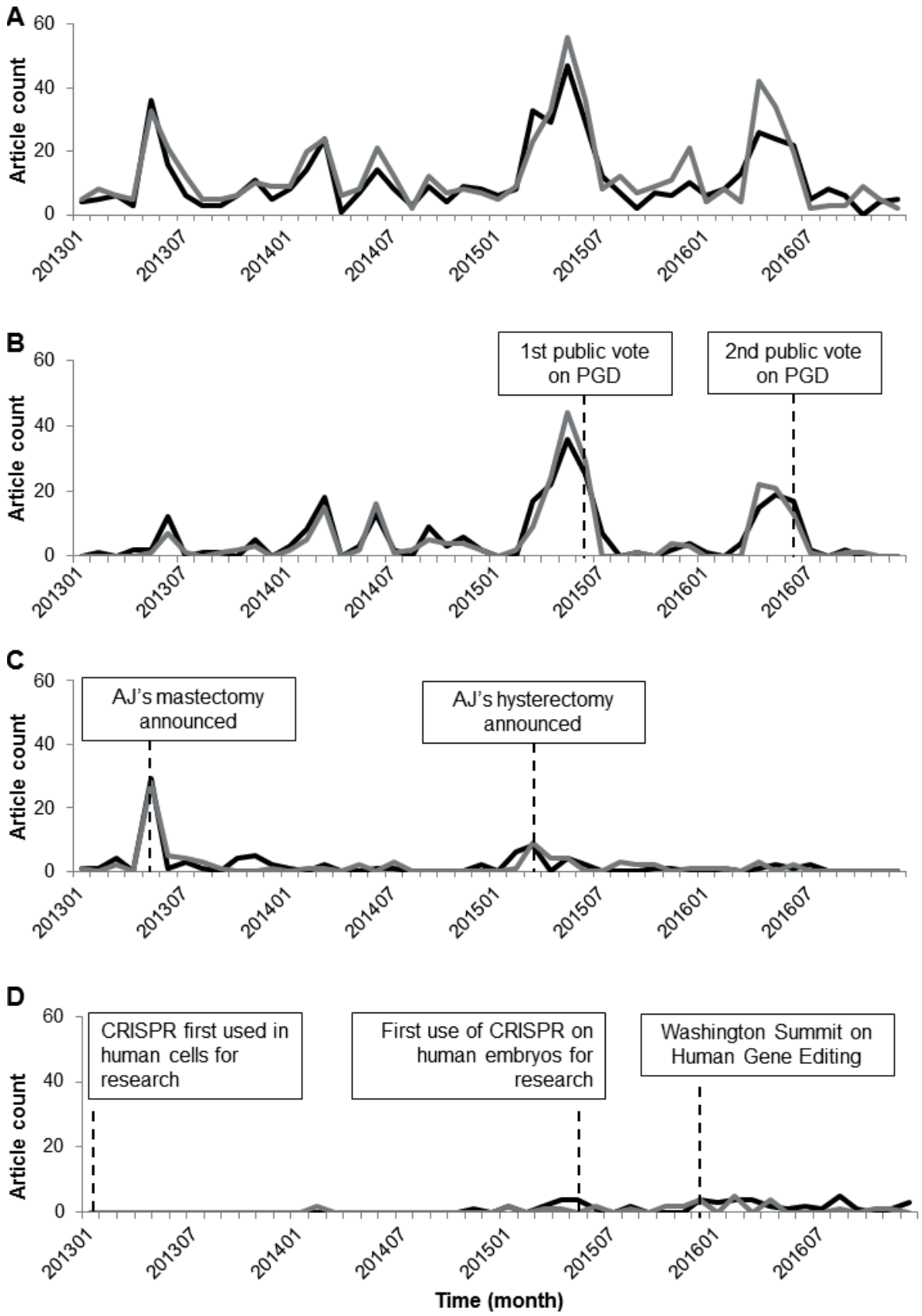

Figure 6-2: Development of topics over time illustrate that event-specific coverage is dominating. Black = German language, grey = French language. A) All articles (N=1163); B) Preimplantation and prenatal Genetic Testing (*PPGT*, N=513); C) Genetic Testing on Born Humans (*GTBH*, N=172); D) Genome Editing (*GEd*, N=76). See Figure B-2 in Appendix B-3 for time development of the other topics.

decisions about mastectomies rather than "hard" topics such as new research findings were most widely reported. Interview readers also remembered such "soft" stories best (Henderson and Kitzinger 1999). The celebrity, the controversy and issues of sexuality due to the mastectomy, made this topic especially interesting for tabloids, which had the highest coverage ratio for this topic (Figure 6-1C).

The overall distribution of topics over time (Figure 6-2A) illustrates that coverage was strongly influenced by the PGD votes and by Angelina Jolie's statements. No other events related to medical genetics were able to attract as much media coverage. This fits well with previous findings suggesting that the coverage about the Human Genome Project was largely event-centred (Gerhards and Schäfer 2006) and that political events, as well as the involvement of celebrities, seem to be most successful in attracting media coverage.

6.5.3 Human genetic diseases (*HGD*): focus on rare diseases

HGD comprised articles about cancer, Down syndrome and other genetic diseases. An important amount, 58.6% of the articles in *HGD* (N=102), were specifically about rare diseases (data not shown): "Genome clinic unravels the mystery of rare diseases" (Davaris, July 22, 2013).

Rare diseases were particularly emphasized in the French-speaking coverage (Figure 6-3A). The main reasons for coverage were charity events, but also new findings from local research groups regarding rare diseases, which is also why most articles (92%) were in a notice or news format (Figure 6-1B). This ratio was even higher in the French part alone (Figure 6-3B).

6.5.4 Personalized medicine (*PM*) and Gene therapy (*GTh*): little and superficial coverage

The articles assigned to *PM* focused on clinical, economic, scientific, social or ethical implications:

> *"Personalized medicine, a revolution in progress" (Boder, September 22, 2015)*
>
> *"Childhood cancer: towards personalized treatment" (Beauté, October 05, 2013)*
>
> *"'Personalized medicine will contribute to lower prices'" (Cepeda, July 12, 2015)*

GTh comprised articles about gene therapy in all stages including basic animal experiments, clinical trials and possible clinical applications, such as:

> *"A genetic pathway to treat eye diseases" (Gordon, February 02, 2014)*
>
> *"Parkinson's disease: first promising results for gene therapy" (Benkimoun, January 10, 2014)*
>
> *"Gene therapy against viscous lung mucus: treatment with foreign DNA can help Cystic Fibrosis patients" (Niederer, July 08, 2015)*

GTh and *PM* were both largely neglected by tabloids (Figure 6-1C). Only two longer reports about *PM* were published in the German-speaking part and eight in the French-speaking region (Figure 6-3B), indicating that highly informative coverage was generally low for *PM*, despite the fact that the media considered it as an important issue for the future of healthcare, as illustrated in the headlines above.

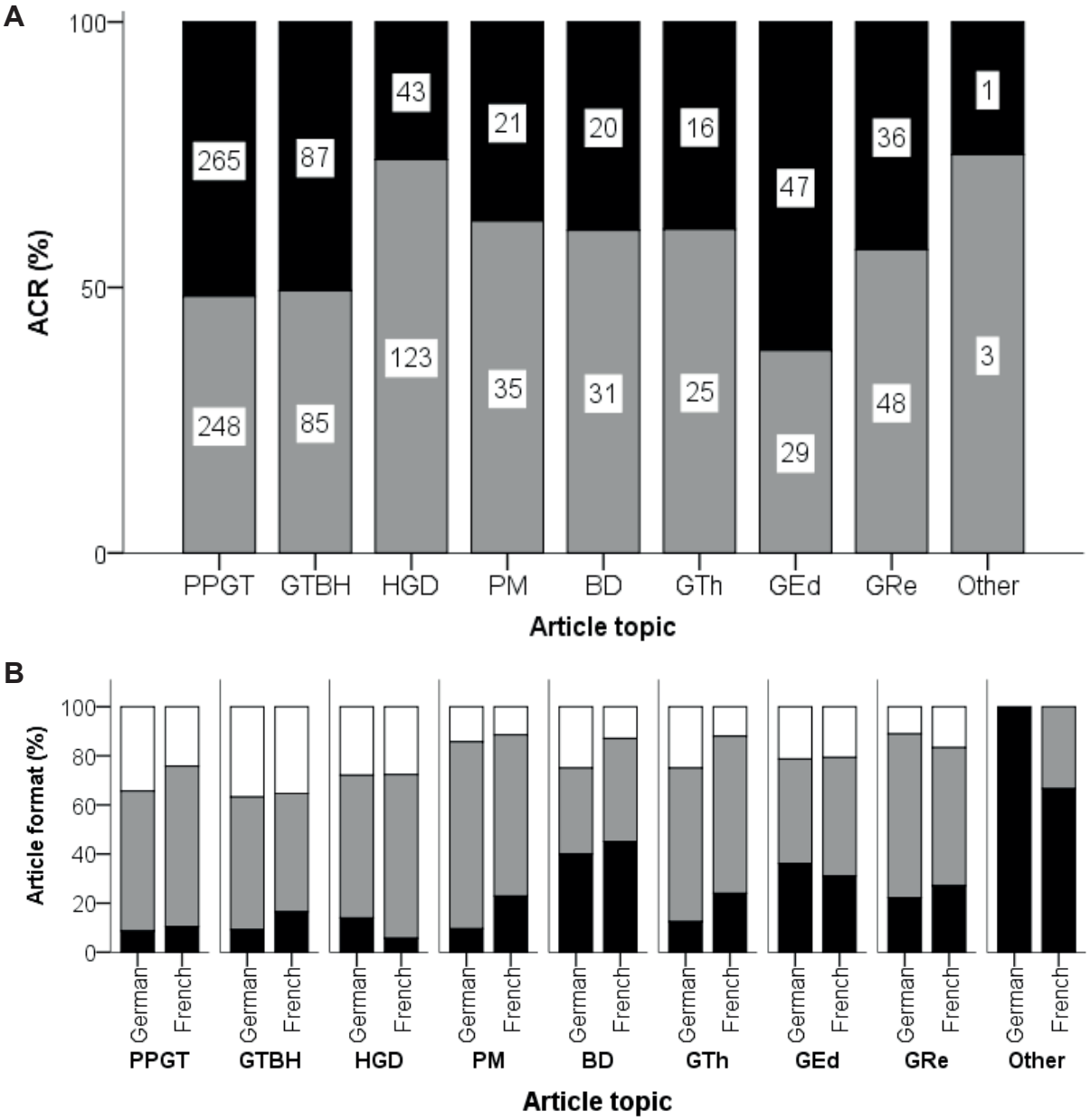

Figure 6-3: Comparison of language regions.
A) Article Count Ratio (ACR) between coverage of the two language regions. 50% means the same number of articles published in both regions. Total article counts stated in the bars. Black = German language, grey = French language. B) Language-specific comparison of article format, based on article length. Black = reports (>800 words); grey = news (200-800 words); white = notice (<200 words).

Although *GTh* is a highly scientific topic, only eight articles (20%) exceeded 800 words, two from the German-speaking coverage and six from the French-speaking coverage (Figures 6 1B and 6 3B). The ratio of highly informative reports thus is rather low, especially when compared to other scientific topics, such as *GRe* or *BD*.

6.5.5 Genome editing (*GEd*): little coverage despite important scientific progress

GEd contained technical, economic, ethical and medical implications of that technique, summarized by the headline: "A new genetic engineering tool is conquering laboratories; researchers speak of a revolution in biotechnology and a new future for gene therapy" (Stallmach, November 26, 2014). More specifically, most articles focused on a newly discovered engineering technique called CRISPR/Cas9: "Brave new world of gene design? 'Crispr' - how humans try to seize a technique from nature" (Kegel, March 09, 2016).

The first article about the genome editing application of CRISPR/Cas9 was published in 2012 (Jinek et al. 2012), and its application on human cells in January 2013 (Jinek et al. 2012; Jinek et al. 2013). In May 2015, the first report of the use of CRISPR/Cas9 in human embryos was released by a Chinese research team (Liang et al. 2015), which resulted in discussions about ethics in international media (Niemiec, Zimmermann, and Howard 2016) and led to the international Washington Summit on Human Gene Editing in December 2015 (Olson et al. 2016).

Generally, the coverage about *GEd* in our sample is much lower than for *PPGT* and *GTBH*, and event-specific peaks are not remarkable (Figure 6-2D). Based on the Issue-attention cycle theory (Downs 1972), we would have expected event-specific coverage for *GEd* as well, but the coverage began to be present in the investigated media only by January 2015, though on a low level compared to the other events affecting *PPGT* and *GTBH*. CRISPR/Cas9 thus was not a key event for Swiss media coverage: Even though the Swiss media picked up that topic some months before it started to become a subject of profound ethical discussion after its use on human embryos (Liang et al. 2015; Reardon 2015), the first emergence and potential use of CRISPR/Cas9-mediated genome editing in January 2013 was completely neglected. International events related to *GEd* such as the Washington Summit on Human Gene Editing (Olson et al. 2016) in December 2015 did not attract much attention, either. In particular, the French-speaking region with 29 articles had very low coverage about *GEd* considering the internationally discussed impact of CRISPR/Cas9 during the investigation period (Reardon 2015). However, 34% of *GEd* articles were longer than 800 words, which is a higher ratio than for most other topics, and only five *GEd*

articles were published in tabloid newspapers (Figure 6-1B and C). These indicators suggest that the coverage took into account the topic's complexity compared to other topics.

Researchers from Germany investigated the media coverage around genome editing for therapeutic purposes in German newspapers. They found that genome editing in somatic cells, which are not inheritable, were legitimized in media coverage because of the prospect of helpful new therapies. In contrast, media coverage about genome editing in inheritable germline cells was more controversial, because the focus was more on non-medical issues, such as the danger of designer babies (Voß 2010). Even though this study was conducted before the discovery of CRISPR/Cas9, the findings are comparable to ours: Some 18% of our articles were about medical and technical implications, which mainly concerned somatic applications and mostly highlighted advantages, e.g. "Crispr brings new hope to cancer patients" (Stallmach, July 01, 2016), but the majority of *GEd* articles (36%) focussed on germlines and were critical: "Ethically questionable gene therapy - Chinese researchers try to modify human embryos" (Stallmach, April 24, 2015).

6.5.6 Big Data in Medical Genetics and Health (*BD*) and Genetic research (*GRe*): better adapted to science communication than other topics

BD encompassed articles about the emergence and use of biobanks as databases for human genetic material: "'Biobanks will acquire more and more value'" (Dessibourg, April 28, 2016). Furthermore, issues of Big Data including data sharing, data protection, social and economic implications were addressed: "Big Pharma marries Big Data" (Schätti and Thier, July 16, 2014). *BD* had a very low ratio of news agency articles compared to the other topics (Figure 6-1C) and a high number of reports longer than 800 words (Figure 6-1B). This indicates that the Swiss print media reported more self-investigated and content-rich articles regarding *BD* compared to other topics, and thus seem to assign more relevance to *BD* than to *GTh* or *PM* for example, despite the fact that the number of articles was comparable across these three topics (Figure 6-1A).

GRe included articles from different research disciplines such as biotechnology, biology, medicine, sociology, bioethics and philosophy: "Only the best for the children; liberal ethicists advocate genetic improvement of children - which provokes not only religious people" (Hofmann, February 19, 2015). A total of 21 articles (25%) were long enough to be able to include rich background information, and only a small amount of coverage was present in tabloids (4 articles,

Figure 6-1B and C). This indicates that coverage about *GRe* – similar to *GEd* – took scientific complexity into account compared to other topics. In comparison to previous studies, the number of articles about *GRe* was low. During the success phase of the Human Genome Project in 2000, 395 articles were reported about it in Germany in just one year (coming from three newspapers), 49 articles in Ireland (four newspapers) and 260 in French-speaking Canada (five newspapers) (O'Mahony and Schäfer 2005; Racine et al. 2006). For Swiss studies, absolute numbers are not available, but *GRe* was the second most prevalent topic (after the political discussion about legal changes) in Swiss German coverage 1997-1999 (Leonarz 2002). Thus, even though direct comparability of numbers is limited, it shows that *GRe* is a rather underrepresented topic in Switzerland in recent years when compared to the late 1990s and early 2000s. This might be due to the lack of a particular event triggering *GRe* coverage during the investigation period.

6.5.7 Only small differences in the amount of coverage between the two language regions

Overall, we found that the total number of articles about medical genetics was slightly higher in the French-speaking part of Switzerland (54%). *HGD* was the topic with the most prominent difference between the language regions: 74.1% of all articles came from the French language region. The only topic that had considerably more articles in the German-speaking part was *GEd* (Figure 6-3A). All other topics related to recent translational genetic research, such as *GRe*, *PM*, *BD* and *GTh*, were predominant in the French-speaking region, but these differences were not statistically significant (data not shown).

Additionally, the distribution of published media articles over time followed very similar patterns between both language regions (Figure 6-2). This is an expected finding because the most prominent peaks were caused by clearly distinguishable events with national and international relevance. We also compared the mean length of articles between the two language regions for each topic as an indicator for the topic-specific depth of the coverage, but could not find any significant differences in article-length between the two language regions (data not shown). There were no significant differences regarding article formats (Figure 6-3B).

We had expected to find differences in media coverage between the two language regions because of cultural differences. Generally, the Swiss-French part is more open towards new genetic applications – PGD votes were accepted by a much higher proportion of voters in the French part (74% in favour) than in the German part (56% in favour) (Ballmer-Cao et al. 2016). The same was already true for the Gene Protection vote in 1998. The ban of genetically modified organisms

was accepted by 36% in the German but by only 19% in the French part (Swiss Federal Council 1998)). A vote-independent survey on attitudes towards genetic technology reported the same trend, namely that the French-speaking part had the most positive attitude (Longchamp et al. 2003). In another context, the same is true for organ donation attitudes (Schulz et al. 2006). However, these cultural differences were not significantly reflected in our data. It is possible that differences are more of a qualitative rather than quantitative nature, which would have to be investigated by comparing, for example, the content and the tone of the articles.

6.6 Conclusion

This study gives a quantitative overview over publicly debated issues in the field of medical genetics. Our results show that the Swiss print media and the news agencies cover a wide variety of genetic topics within medical genetics. The amount and nature of coverage differed significantly between the different topics. Political votes and a celebrity's statement attracted media attention and dominated the coverage. Conversely, important scientific events related to *GEd* were not mirrored in the Swiss coverage. Compared to topics more related to clinical aspects or politics (*PPGT*, *HGD*, *PM*) the media coverage of more scientific topics (i.e., *GEd*, *GRe*, *BD*, *GTh*) is not prevalent. It is no surprise that the articles concerning scientific topics generally contain more reports than the ones with less of a scientific angle, even though we would have expected coverage of *PM* and *GTh* to be more in-depth based on their content. As we did not find significant differences between the two language regions of Switzerland, the national debate about medical genetics seems to form in a rather similar way in the different parts of the country, without important culture-specific variations in coverage. National players in the pharmaceutical industry may add to this effect.

With our approach, we aimed to cover all topics in medical genetics extensively and conclusively, but we acknowledge that topics could be distinguished and defined in other ways. For reasons of resources, the data collected remained restricted to certain quantitative indicator variables, and the conclusions are based on selected theories and hypotheses. However, our results raise questions and provide insights for targeted in-depth content analysis, media effect studies and comparison with surveys on knowledge and attitudes of the public. Our extensive and conclusive topic list will allow the research community to investigate and address media coverage and public debates about genetics more precisely, with a well-defined scope and more precise keyword searches, and all topics have international relevance, allowing their translation to studies in other settings.

Researchers from other disciplines can use the results for planning public presentations of their results. Public and mass media are not really interested in scientific findings but they do intensively cover celebrities, political disputes and economic development of national players. Therefore, one way to promote scientific media coverage should be the establishment of cooperation with celebrities and political lobbying.

6.7 References

Ballmer-Cao, Thanh-Huyen, Urs Bieri, Marina Delgrande Jordan, Lukas Golder, Sibylle Hardmeier, Hans Hirter, Ulrich Klöti et al. 2016. "Vox 121 - Votation Du 05.06.2016.".

Beauté, Bertrand. 2013. "Diagnostic Anténatal: Bientôt Des Bébés Parfaits?" La Tribune de Genève, September 7.

———. 2013. "Cancers Chez L'enfant: Vers Un Traitement Personnalisé." La Tribune de Genève, October 5.

Benkimoun, Paul. 2014. "Maladie De Parkinson: Premiers Résultats Prometteurs Pour Une Thérapie Génique." Le Temps, January 10.

Boder, Willy. 2015. "La Médecine Personnalisée, Une Révolution En Marche." Le Temps, September 22.

Bonfadelli, Heinz, Urs Dahinden, and Martina Leonarz. 2002. "Biotechnology in Switzerland: High on the Public Agenda, but Only Moderate Support." Public Underst Sci (11): 113–30.

Borzekowski, Dina L. G., Yue Guan, Katherine Smith, Lori H. Erby, and Debra L. Roter. 2014. "The Angelina Effect: Immediate Reach, Grasp, and Impact of Going Public." Genet Med 16 (7): 516–21. doi:10.1038/gim.2013.181.

Brotschi, Markus. 2016. "«Die Frau Ist Keine Petrischale»." *Tages-Anzeiger*, May 24.

Bunnik, Eline M., Maartje H. N. Schermer, and A. C. J. W. Janssens. 2011. "Personal Genome Testing: Test Characteristics to Clarify the Discourse on Ethical, Legal and Societal Issues." BMC Med Ethics 12:11. doi:10.1186/1472-6939-12-11.

Burke, Wylie, Paul Appelbaum, Lauren Dame, Patricia Marshall, Nancy Press, Reed Pyeritz, Richard Sharp, and Eric T. Juengst. 2015. "The Translational Potential of Research on the Ethical, Legal and Social Implications of Genomics." Genet Med 17 (1): 12–20. doi:10.1038/gim.2014.74.

Cepeda, Ariel. 2015. "«La Médecine Personnalisée Contribuera À La Baisse Des Prix»." Le Matin, July 12.

Condit, Celeste M. 2001. "What Is 'Public Opinion' About Genetics?" Nat Rev Genet 2 (10): 811–15. doi:10.1038/35093580.

Dahdouh, Elias M., Jacques Balayla, François Audibert, R. Douglas Wilson, Jo-Ann Brock, Carla Campagnolo, June C. Carroll et al. 2015. "Technical Update: Preimplantation Genetic Diagnosis and Screening." J Obstet Gynaecol Can 37 (5): 451–63. doi:10.1016/S1701-2163(15)30261-9.

Dahlgren, Peter. 2009. Media and Political Engagement: Citizens, Communication, and Democracy. 1. publ. Communication, society and politics. Cambridge u.a. Cambridge University Press.

Davaris, Sophie. 2013. "La Clinique Du Génome Perce Le Mystère Des Maladies Rares." La Tribune de Genève, July 22.

Dessibourg, Olivier. 2016. "«Les Biobanques Acquerront De Plus En Plus De Valeur»." Le Temps, April 28.

Downs, Anthony. 1972. "Up and down with Ecology-the Issue-Attention Cycle." Public Interest 28 (Summer): 38–50.

Epstein, Charles J. 2006. "Medical Genetics in the Genomic Medicine of the 21st Century." Am J Hum Genet 79 (3): 434–38. doi:10.1086/507610.

Eyck, Toby A. T., and Melissa Williment. 2003. "The National Media and Things Genetic Coverage in the New York Times (1971–2001) and the Washington Post (1977-2001)." Sci Commun 25 (2): 129–52. doi:10.1177/1075547003259212.

Garrett, Jinnie M., and Stephanie J. Bird. 2007. "Ethical Issues in Communicating Science." Sci Eng Ethics 6 (4): 435–42. doi:10.1007/s11948-000-0001-7.

Gerhards, Jürgen, and Mike S. Schäfer. 2006. Die Herstellung einer öffentlichen Hegemonie: Humangenomforschung in der deutschen und der US-amerikanischen Presse. Wiesbaden: Verlag für Sozialwissenschaften.

Gordon, Elisabeth. 2014. "Une Piste Génétique Pour Traiter Des Maladies De L'œil." Le Matin, February 2.

Görke, Alexander, Matthias Kohring, and Georg Ruhrmann. 2000. "Gentechnologie in Der Presse." Pub 45 (1): 20–37. doi:10.1007/s11616-000-0048-0.

Henderson, Lesley, and Jenny Kitzinger. 1999. "The Human Drama of Genetics: 'Hard' and 'Soft' Media Representations of Inherited Breast Cancer." Sociol Health Illn 21 (5): 560–78. doi:10.1111/1467-9566.00173.

Hofmann, Markus. 2015. "Der Bundesrat öffnet den Sesam der Gentests: Der Schutz des ungeborenen Lebens soll gewährleistet bleiben." *Neue Zürcher Zeitung*, February 19.

Holtzman, Neil A., Barbara A. Bernhardt, E. Mountcastle-Shah, Joann Rodgers, Ellen S. Tambor, and Gail Geller. 2005. "The Quality of Media Reports on Discoveries Related to Human Genetic Diseases." Community Genet 8 (3): 133–44. doi:10.1159/000086756.

Jarren, Otfried, and Martina Vogel. 2011. "„Leitmedien" Als Qualitätsmedien: Theoretisches Konzept Und Indikatoren." In Krise Der Leuchttürme Öffentlicher Kommunikation: Vergangenheit Und Zukunft Der Qualitätsmedien, edited by Roger Blum, Heinz Bonfadelli, Kurt Imhof, and Otfried Jarren, 17–30. Wiesbaden: Verlag für Sozialwissenschaften.

Jinek, Martin, Krzysztof Chylinski, Ines Fonfara, Michael Hauer, Jennifer A. Doudna, and Emmanuelle Charpentier. 2012. "A Programmable Dual-RNA–Guided DNA Endonuclease in Adaptive Bacterial Immunity." Science 337 (6096): 816–21. doi:10.1126/science.1225829.

Jinek, Martin, Alexandra East, Aaron Cheng, Steven Lin, Enbo Ma, and Jennifer A. Doudna. 2013. "RNA-Programmed Genome Editing in Human Cells." eLife 2:e00471. doi:10.7554/eLife.00471.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jolie Pitt, Angelina. 2015. "My Children Won't Have to Say 'Mom Died of Ovarian Cancer'." The *Daily Telegraph*, March 25.

Kamenova, Kalina, Amir Reshef, and Timothy A. Caulfield. 2014. "Angelina Jolie's Faulty Gene: Newspaper Coverage of a Celebrity's Preventive Bilateral Mastectomy in Canada, the United States, and the United Kingdom." Genet Med 16 (7): 522–28. doi:10.1038/gim.2013.199.

Kegel, Bernhard. 2016. "Schöne Neue Welt Des Gen-Designs? «Crispr» - Wie Sich Der Mensch Einer Technik Der Natur Zu Bemächtigen Versucht." *Neue Zürcher Zeitung*, March 9.

Kohring, Matthias, and Jörg Matthes. 2002. "The Face(T)S of Biotech in the Nineties: How the German Press Framed Modern Biotechnology." Public Underst Sci 11 (2): 143–54. doi:10.1088/0963-6625/11/2/304.

Kolb, Steffen. 2005. Mediale Thematisierung in Zyklen: Theoretischer Entwurf und empirische Anwendung. Köln: Herbert von Halem.

Krippendorff, Klaus, and Mary A. Block, eds. 2009. The Content Analysis Reader. Thousand Oaks, Calif: SAGE Publications.

Kung, Johnny T., and Marnie E. Gelbart. 2012. "Getting a Head Start: The Importance of Personal Genetics Education in High Schools." Yale J Biol Med 85 (1): 87–92.

Leonarz, Martina. 2002. "Die Gentechnologie als kontroverses Medienthema: Eine Zeitungsinhaltsanalyse von 1997 bis 1999." In Gentechnologie in der öffentlichen Kontroverse: Eine sozialwissenschaftliche Analyse, edited by Heinz Bonfadelli and Urs Dahinden, 25–43. Zürich: Seismo.

Liang, Puping, Yanwen Xu, Xiya Zhang, Chenhui Ding, Rui Huang, Zhen Zhang, Jie Lv et al. 2015. "CRISPR/Cas9-Mediated Gene Editing in Human Tripronuclear Zygotes." Protein Cell 6 (5): 363–72. doi:10.1007/s13238-015-0153-5.

Longchamp, Claude, Urs Bieri, Silvia Ratelband-Pally, and Stefan Tschöpe. 2003. "Klare Präferenzen Bei Der Anwendung: Schlussbericht Zum Gentechnik-Monitor 2003 Für Die Interpharma." http://www.gfsbern.ch/de-ch/Detail/klare-praeferenzen-bei-der-anwendung.

MacQueen, Kathleen M., Eleanor McLellan, Kelly Kay, and Bobby Milstein. 2009. "Codebook Development for Team-Based Qualitative Analysis." In Krippendorff and Block 2009, 211–19.

Mast, Claudia. 2012. ABC Des Journalismus: UVK.

Mei, Yue, Yan Wang, Huiqian Chen, Zhong S. Sun, and Xing-Da Ju. 2016. "Recent Progress in CRISPR/Cas9 Technology." J Genet Genomics 43 (2): 63–75. doi:10.1016/j.jgg.2016.01.001.

Niederer, Alain. 2015. "Gentherapie Gegen Zähen Lungenschleim; Bei Patienten Mit Zystischer Fibrose Kann Eine Behandlung Mit Fremd-DNA Helfen." *Neue Zürcher Zeitung*, July 8.

Niemiec, Emilia, Bettina M. Zimmermann, and Heidi C. Howard. 2016. "Ethical Issues of Gene Editing: What Does Popular Media Report?" European Conference for Human Genetics 2016, Barcelona, May 24.

O'Mahony, Patrick, and Mike S. Schäfer. 2005. "The 'Book of Life' in the Press Comparing German and Irish Media Discourse on Human Genome Research." Soc Stud Sci 35 (1): 99–130. doi:10.1177/0306312705046542.

Olson, Steven, Committee on Science, Technology, and Law, Policy a. G. Affairs, and National Academies of Sciences, Engineering, and Medicine. 2016. International Summit on Human Gene Editing: A Global Discussion: National Academies Press.

Racine, Eric, I. Gareau, H. Doucet, D. Laudy, G. Jobin, and P. Schraedley-Desmond. 2006. "Hyped Biomedical Science or Uncritical Reporting? Press Coverage of Genomics (1992-2001) in Quebec." Soc Sci Med 62 (5): 1278–90. doi:10.1016/j.socscimed.2005.07.024.

Reardon, Sara. 2015. "Global Summit Reveals Divergent Views on Human Gene Editing." Nature 528 (7581): 173. doi:10.1038/528173a.

Rödder, Simone. 2005. Das Buch Des Lebens in Der Presse: Die Darstellung Der Entschlüsselung Des Menschlichen Erbguts in Deutschen Und Britischen Medien. Angewandte Medienforschung : Schriftenreihe für die Kommunikationsforschung 33. München: Verlag Reinhard Fischer.

Rössler, Patrick. 2010. Inhaltsanalyse. 2nd rev. ed. UTB für Wissenschaft: Uni-Taschenbücher. Konstanz: UVK.

Ruhrmann, Georg, and Jutta Milde. 2011. "Zum Nachrichtenwert Von Molekularer Medizin: Eine Inhaltsanalyse Von TV-Meldungen 1995 Bis 2004." In Molekulare Medizin Und Medien: Zur Darstellung Und Wirkung Eines Kontroversen Wissenschaftsthemas, edited by Georg Ruhrmann, Jutta Milde, and Arne F. Zillich, 99-119. Wiesbaden: Verlag für Sozialwissenschaften.

Schäfer, Mike S. 2012. "Taking Stock: A Meta-Analysis of Studies on the Media's Coverage of Science." Public Underst Sci 21 (6): 650–63. doi:10.1177/0963662510387559.

Schätti, Guido, and Jenni Thier. 2014. "Big Pharma Heiratet Big Data." Blick, July 16.

Schulz, Peter J., Kent Nakamoto, David Brinberg, and Joachim Haes. 2006. "More Than Nation and Knowledge: Cultural Micro-Diversity and Organ Donation in Switzerland." Patient Educ Couns 64 (1-3): 294–302. doi:10.1016/j.pec.2006.03.009.

Scott, William A. 2009. "Scott's Pi." In Krippendorff and Block 2009, 347–49.

sda. 2014. "Gentests: Experten Warnen Vor «Exzessen» Und «Scharlatanen»." June 23.

Stallmach, Lena. 2014. “Ein Neues Gentech-Werkzeug Erobert Die Labors; Forscher Sprechen Von Einer Revolution in Der Biotechnologie Und Von Einer Neuen Zukunft Für Die Gentherapie.” *Neue Zürcher Zeitung*, November 26.

———. 2015. “Ethisch Fragwürdige Gentherapie – Chinesische Forscher Versuchen, Menschliche Embryonen Zu Verändern.” *Neue Zürcher Zeitung*, April 24.

———. 2016. “Crispr Bringt Krebskranken Neue Hoffnung.” *Neue Zürcher Zeitung*, July 1.

Straumann, Felix. 2013. “Ein Fruchtbarkeitstest, der Fragen offen lässt.” *Tages-Anzeiger*, March 22.

Swiss Federal Council. 1998. “Eidgenössische Volksinitiative ‘zum Schutz von Leben und Umwelt vor Genmanipulationen (Gen-Schutz-Initiative)’.” Accessed January 23, 2020. https://www.bk.admin.ch/ch/d/pore/vi/vis240t.html.

———. 2005. “Bundesbeschluss über die Volksinitiative «für Lebensmittel aus gentechnikfreier Landwirtschaft»: Volksabstimmung vom 27.11.2005.” Accessed January 23, 2020. https://www.bk.admin.ch/ch/d/pore/va/20051127/det520.html.

———. 2015. “Volksabstimmung Vom 14.06.2015, Vorlage Nr. 592 Übersicht: Bundesbeschluss Vom 12.12.2014 Über Die Änderung Der Verfassungsbestimmung Zur Fortpflanzungsmedizin Und Gentechnologie Im Humanbereich.” Accessed January 23, 2020. https://www.admin.ch/ch/d/pore/va/20150614/det592.html#.

———. 2016. “Volksabstimmung Vom 05.06.2016, Vorlage Nr. 603 Übersicht: Änderung Vom 12.12.2014 Des Bundesgesetzes Über Die Medizinisch Unterstützte Fortpflanzung Und Gentechnologie Im Humanbereich.” Accessed January 23, 2020. https://www.admin.ch/ch/d/pore/va/20160605/det603.html.

Vögeli, Erika. 2015. “Diagnostic Préimplantatoire : La Porte Ouverte À L’homme «Amélioré».” Le Temps, May 28.

Voß, Miriam. 2010. Gesunde Gene: Die mediale Diskussion um die Gentherapie: transcript Verlag.

WEMF. 2016. MACH Basic 2016-2: Die Schweizer Leserschaftsstudie: WEMF AG für Werbemedienforschung.

Wirth, Werner, and Steffen Kolb. 2004. “Designs and Methods of Comparative Political Communication Research.” In Comparing Political Communication: Theories, Cases, and Challenges, edited by Frank Esser, 87–111. Communication, society, and politics. Cambridge: Cambridge University Press.

Chapter 7: Content, evaluations and influences in newspaper coverage of predictive genetic testing: A comparative media content analysis from the United Kingdom and Switzerland

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7.1 Abstract

Predictive genetic testing often entails challenging decisions about preventive measures and uncertain health-related risk predictions. Because of its increasing availability, it is important to assess how to debate it publicly. Newspaper content analysis represents a common and reliable way to investigate public discourse retrospectively. We thus quantitatively compare broadsheet newspaper coverage about predictive genetic testing in the United Kingdom and Switzerland during the period of 2011–2016 regarding content, evaluations, stakeholder influence, and trigger events. British coverage was more extensive and positive and included more personal stories. Swiss coverage had more focus on political issues. Angelina Jolie's announcement about her double mastectomy was the most important coverage trigger. Researchers were the most frequently cited stakeholder group, but stakeholders from government and civil society were also represented. Our results thus reflect a movement toward a more active public engagement with predictive genetic testing. The findings help to improve and enrich public engagement regarding predictive genetic testing.

7.2 Introduction

7.2.1 Predictive genetic testing

On 14 May 2013, the American actress Angelina Jolie announced that she had had a preventive double mastectomy due to an increased genetic risk of developing breast cancer (Jolie, May 13, 2013). Her decision, which attracted international media attention (Kamenova, Reshef, and Caulfield 2014) and enhanced the public's interest in genetic testing and preventive mastectomies (Borzekowski et al. 2014; Lebo et al. 2015), was based on predictive genetic testing. It involves the analysis of the genetic code prior to the development of a certain trait. Here, we focus on predictive genetic testing of born humans, excluding any genetic testing prior to birth, that is, prenatal screenings or preimplantation genetic testing. The traits investigated are usually mutations linked to hereditary diseases like certain cancer types (e.g. hereditary breast and ovarian cancer), metabolic disorders (e.g. familial hypercholesterinaemia), or neurological disorders (e.g. familial Alzheimer's disease). Predictive genetic testing is becoming more common in clinical practice all over the world, but its meaning, impact, and consequences are complex. The decision regarding whether or not to do such testing is a personal one and depends, inter alia, on the nature of the investigated disease, disease history in the family, environmental risk factors, the state of scientific knowledge, and personal factors (i.e. age, gender, family relationships,

psychological coping mechanisms, etc.). Predictive genetic testing usually gives a percentage of risk that a disease might be expressed throughout a person's life because most mutations are associated with less than 100% disease prevalence. This means that even upon diagnosis of the presence of a mutation, it is generally impossible to foresee with certainty if and when a patient will develop the corresponding disease. That is why high-quality and professional genetic counselling is crucial and forms a common part of the clinical process for predictive genetic testing. In contrast, direct-to-consumer genetic testing kits are freely available on the market, often without any offer of counselling prior to or after testing. Still, many of the results of direct-to-consumer genetic testing are of a predictive nature. Because of this commercialization as well as the increase in scientific knowledge and growing public interest, predictive genetic testing is becoming an important issue for societies worldwide.

For a growing number of well-investigated mutations, there are substantial medical benefits related to predictive genetic testing. The possibility of early identification of at-risk people often improves early detection or disease prevention, leading to a better clinical outcome and even the possibility of reducing long-term healthcare costs. Risks regarding predictive genetic testing often relate to a lack of or bad genetic counselling, for example, in direct-to-consumer genetic testing. Professional, non-directive genetic counselling is irreplaceable for predictive genetic testing with a health impact (Evers-Kiebooms et al. 2001), but it is important that non-specialists have access to reliable information and become aware of the most important (personal) risks and benefits around predictive genetic testing. This will give non-specialists the chance to engage actively with these issues and to become less prone to commercially or politically driven, one-sided information.

Predictive genetic testing is both a scientific and a medical topic. The scientific field of genetics is fast-growing and has many practical applications, both medical and non-medical. Predictive genetic testing is a medical application of that scientific field. Such health-related issues are of particular public interest. Science communication research about health-related scientific topics like predictive genetic testing is therefore particularly important. Investigating mass media coverage is a common, reliable, and stable way to analyze public debates about medico-scientific topics retrospectively.

7.2.2 Science communication, mass media, and the public

According to Haran and Kitzinger (2013), research on media coverage regarding human genetics followed four strands. (1) The *Public Understanding of Science* approach investigates whether

media accurately represent science in order to give readers a good understanding of genetics. Evidently, the mass media play an important role in communicating science to the public, particularly regarding genetics and diseases (Garrett and Bird 2007; Schwitzer et al. 2005). Broadsheet newspapers, in particular, aim to uncover complex issues and make them available to the public in an understandable way. They are one of the main public sources of scientific knowledge (Metag and Schäfer 2016), and still have an agenda-setting role for online news platforms (Maier 2010).

Taking the readers in focus, (2) *Health Education* assesses whether they have accurate and full information about genetics. (3) *Public Relations of Science* focuses more on political and industrial campaigns and their influence on readers. The effects of communication on recipients are complex and multi-layered (Condit 2001). According to the dynamic transactional model (Früh and Schönbach 1982, 2005), media effects on readers are individual and highly dynamic, depending, for example, on the recipient's personal situation, cognitive state, and emotions at the moment of consuming media content. Readers, in turn, also influence media content through their interests, opinions, and feedback. Supporting this model, empirical studies show that media coverage does have an important impact on health-related attitudes and behaviour (Bonfadelli 2002; Fishman and Casarett 2006), but the concrete effects remain unclear (Catalan-Matamoros and Peñafiel-Saiz 2017). However, it is important to distinguish between media effects on an individual level, and the effects of media coverage for a society, as it is described in science communication models. Even though they do not cover the whole public discourse, and do not directly reflect public opinion, an exploration of mass media coverage is an important part of grasping public opinion on a topic of public relevance (Condit 2001).

(4) *Public Engagement* with Science investigates active, controversial discussions between different stakeholder groups (Bubela 2006; Bucchi 2008; Gerhards and Schäfer 2009; Jones 2011; Locke 2011; Stilgoe, Locke, and Wilsdon 2014; Schäfer 2009). The engagement of public stakeholders takes place on many levels and has different definitions when translated into practice (Davies 2013). Mass media reporting, which is the basis for this study, is one type of engagement mechanism of public stakeholders, in distinction to publicity and public hearings or meetings (Rowe and Frewer 2005). Gerhards and Schäfer (2009) attempted to make the extent of public engagement within mass media reporting empirically measurable. According to their framework, the *"science-dominated scientific public sphere"* applies a passive role to media recipients. Media, therefore, should transport scientific content as communicated by scientists, but also in a way that is understandable for non-specialists. The *"contextualized scientific public sphere"* stands

for an active and controversial public discourse, where science is one stakeholder among many and needs public legitimation. A contextualized scientific public sphere is thus characterized by a high variety of stakeholders in media coverage (as opposed to a dominance of scientific stakeholders), the presence of positive, negative, and ambivalent assessments (instead of predominantly positive reporting), and a high variety of topics (instead of scientific topics dominating). Applying these public sphere models in a media content analysis about human genetic research in 1999–2001, Gerhards and Schäfer (2009) found what they called a "hegemony of supporters" in both the United States and Germany. Following the *"science-dominated scientific public sphere"* model, scientific stakeholders dominated media coverage of human genetic research with their positive assessments in both investigated countries.

Another approach to empirically measure public engagement in mass media reporting is *medialisation* (Weingart 2005), which was measured by the extensiveness of coverage about a certain topic, the diversity of stakeholders and content, and the level of controversial evaluations of the scientific topic (Schäfer 2009). The more extensive, pluralized, and controversial a topic is treated, the more it is medialized. Schäfer (2009) showed that the level of medialisation is topic-dependent. In media coverage reflecting active public engagement with science, we expect highly pluralized and controversial media coverage. We also expect that a higher extent of coverage is an indicator of higher engagement with the topic. Thus, high medialisation reflects higher public engagement, but given the dynamic nature of media effects on an individual level, this effect can only be true on a societal level.

Generally, empirical analyses focused on media coverage of genetic research (Henderson and Kitzinger 2007; O'Mahony and Schäfer 2005), or on more specific applications like pharmacogenetics (Almomani et al. 2015), genetic screening programs (Holton, Canary, and Wong 2016), prenatal genetic testing (Lewis, Choudhury, and Chitty 2015; Kamenova et al. 2016), and direct-to-consumer genetic testing (Lynch et al. 2011). To our knowledge, this is the first media content analysis about the particularities of predictive genetic testing.

7.2.3 Comparing Switzerland and the United Kingdom

Comparative research faces at least two major problems: the comparability of data and the choice of countries under scrutiny (Wirth and Kolb 2013). To assure comparability, the functional equivalence approach is commonly used in the social sciences as well as in psychology (Wirth and Kolb, 2004). The first part of Wirth's and Kolb's guideline to comparability proposes the examination of the functions of the topic and objects of research within the specific society

(e.g., Esser 2000; Patzelt 1997; Wirth and Kolb 2004): While the United Kingdom and Switzerland can be considered rather similar in terms of welfare and quality of healthcare, there are some important differences. First, the United Kingdom has a parliamentary democracy under a constitutional monarchy (gov.uk 2020) as a political system, whereas Switzerland has a direct democracy, meaning that the people have a direct say in all policy issues (Swiss Federal Council 2017). For example, in case of changes to legislation, it is the parliament that has the final decision in the United Kingdom, while in Switzerland, there is generally the possibility of a public vote. Second, the British media system is more liberal and adopted a commercial model earlier than in Switzerland, where the political party–related press dominated the media system until the mid-20th century (Hallin and Mancini 2011). Taking these differences into account, we consider the role of the mass media in any public discourse as functionally equivalent in both countries, because none of the differences affect the information function of mass media and especially of leading quality newspapers. Kolb (2005) presents an in-depth analysis of the comparability of Western countries' press and has come to the same conclusion for Germany, France, and the United Kingdom in a study of health risks potentially caused by car traffic.

An optimal rational choice of countries for comparison can be taken either based on the quasi-experimental *most similar systems design* (with a different outcome) or the *most different systems design* (with the same outcome) (Berg-Schlosser 2003; Teune and Przeworski 1970). We follow the first approach: the two countries should be similar with regard to the research object of media and society. Searching for specific differences is economical due to the limited number of differences that may occur in rather similar countries or cultures, and it is fruitful because of the possible integration in a quasi-experimental design that after finding some dissimilarity looks back at possible causes (Kolb 2004). Even after gathering the data and when interpreting it, a self-critical search for sources of bias and causal testing remains recommendable as will be seen in the discussion.

There are differences between Switzerland and the United Kingdom that could possibly cause differences in the results. First, their legislation is different regarding predictive genetic testing: in Switzerland, the Human Genetic Testing Act (Swiss Federal Council 2004) regulates very specifically who can prescribe predictive genetic testing and how it should be performed, but there is no specific legislation in the United Kingdom regarding predictive genetic testing. Second, there are important differences in the health insurance system: Switzerland has mandatory public health insurance with tax-independent financial contributions from individuals (Federal Office of Public Health 2019), complemented by a quite well-established private health insurance

sector. In contrast, the National Health Service (NHS) organizes the health care system in the United Kingdom; it is directly funded by national insurance, and the private health insurance sector is rather small (National Health Service 2020).

We compare public debates about predictive genetic testing from 2011 to 2016 in broadsheet newspaper coverage in the United Kingdom and the German-speaking part of Switzerland, which is the largest language region of the country, covering 65% of the Swiss population. We aim to assess the content and evaluation of media coverage of predictive genetic testing and determine which stakeholders and which events influenced the media reporting to what extent. We furthermore investigate the extent to which newspaper coverage of predictive genetic testing reflects public engagement with the topic by comparing it to the aforementioned science communication models (Gerhards and Schäfer 2009; Weingart 2005).

7.3 Methodology

7.3.1 Study design

We compared broadsheet newspaper coverage of predictive genetic testing from the German-speaking part of Switzerland and the United Kingdom. Broadsheet newspapers aim to provide in-depth information of high journalistic quality, and they are considered to serve as both an information source and agenda-setter for other media such as television and online media (Jarren and Vogel 2011). A population-wide Swiss survey revealed that the Swiss population mainly receives scientific news through newspapers (Metag and Schäfer 2016). Thus, to assess media coverage of a complex medico-scientific topic such as predictive genetic testing, broadsheet newspapers are a good indicator. For quantitative comparison, we chose a total of four broadsheet newspapers based on their functional equivalence in the country (Wirth and Kolb 2004). Each newspaper had high circulation and traditions on different political spectra to minimize newspaper-specific bias. For Switzerland, we chose *Tages-Anzeiger* from the left-liberal spectrum and the right-liberal *Neue Zürcher Zeitung*. Equivalently, we chose the left-liberal *The Guardian* and the right-conservative *Daily Telegraph* from the United Kingdom.[1] All chosen broadsheet newspapers target a national and, in the case of *The Guardian* and *Neue Zürcher Zeitung*, an international audience.

We used the online database Factiva (Dow Jones, New York, United States) for the sampling of relevant articles, generating separate search algorithms for English- and German-speaking

1 We chose The *Daily Telegraph* over *The Times* because of higher readership.

media based on a previous (unpublished) analysis from our group.[2] The precise search algorithms are available as supplementary material.[3] For the analysis, we only included articles where predictive genetic testing on born humans, its consequences or its social, economic, and political implications were the focus in at least 1/5th of the article body in order to avoid the inclusion of unspecific articles. We excluded articles about epigenetics, prenatal or preimplantation predictive genetic testing as well as post-symptomatic genetic testing, where a genetic test is done because of already existing symptoms or disease for diagnostic or therapeutic purposes (i.e. pharmacogenetics, or cancer diagnosis-related testing). Furthermore, we excluded articles about genetic testing on animals or plants and non-medical applications of genetic testing, such as forensics, evolutionary genetics, or ancestry-search as well as articles shorter than 100 words, because that content was not rich enough to be useful for our variables of interest. All articles collected from Factiva were screened for inclusion or exclusion by two researchers (B.Z. and N.A.) separately. In cases of disagreement, researchers discussed articles and reached consensus about whether to include the article based using the aforementioned criteria. The unit of analysis was each article, but interviews, infoboxes, and first-page short announcements were merged into one unit of analysis with the main article (defined as the longest piece of text) if they were published on the same day, in the same medium, and on the same topic. Editorials and opinion pieces always remained as a separate unit of analysis, even if the above criteria applied.

7.3.2 Codebook development

We used a mixed-method approach, starting with a summarizing qualitative content analysis. Following the approach of Mayring (2015), topic-specific text passages from all German-speaking articles were first paraphrased, then rewritten in a more abstract form. We then reduced the number of statements by summarizing similar passages. This was done by two researchers separately and then compared. We repeated this reduction step two times, aiming for agreement after each step. Having a list of 70 abstract content categories, we translated them into English and compared them with 20 randomly chosen articles from British coverage. If we found text passages in the UK coverage that were not reflected in our content category list yet, we extended the list until no more categories were found. We then further reduced these categories based on topicality, which resulted in 13 distinct topics. Again, this stepwise reduction was performed by two researchers separately, and an agreement was reached upon discussion.

2 See chapter 6.

3 See Appendix C-1.

The codebook for quantitative analysis and the stakeholder list were inspired by quantitative content analyses by Gerhards and Schäfer (2007) and Schäfer (2009) to allow for comparability of results. The complete codebook is available as supplementary material.[4] We tested and adapted the codebook in several steps, aiming to capture a high amount of content but allowed high inter-coder reliability. Scott's Pi was used to calculate the reliability score (Scott 2009). The mean reliability score was 0.72, ranging from 0.48 to 0.84. A reliability score higher than 0.7 is considered a good, higher than 0.4 a medium reliability. Variables with medium reliability were improved with additional inter-coder agreements in case of uncertainties. Furthermore, since the complexity of articles differed importantly (depending mainly on the numbers of stakeholders and topics that were covered), and reliability decreased with article complexity, we decided to increase the appropriateness of our data by double-coding articles that had more than seven stakeholders and more than one topic, or that were subjectively considered unclear by the coding researcher. A total of 43 articles (in addition to the 20 articles from reliability testing) were analyzed by two researchers separately and compared. In case of non-agreement, we reached agreement upon discussion.

7.4 Results

7.4.1 Content comparison

The Factiva keyword search resulted in 1154 hits. A total of 137 articles met the inclusion criteria. We found 2.5 times more articles about predictive genetic testing in the United Kingdom than in Switzerland throughout the investigation period. We assigned each article one or several topics, which were identified by qualitative content analysis and reflect the content of media coverage. In total, we assigned 195 topics to 137 articles (Table 7-1). The most common topic in both countries was *Public Debates*, which included celebrities going public with a genetic disease risk or a preventive treatment, such as Angelina Jolie, as well as political discussions. *Research*, accounting for research studies as well as clinical trials, was more common in the United Kingdom than in Switzerland. The same is true for *Preventive Treatment* in relation to predictive genetic testing and for *Genetics in Society*. The latter topic included articles about the implementation of predictive genetic testing into healthcare, general guidelines, the notion of the genetic revolution or personalized medicine. A common topic in Switzerland was *Direct-to-Consumer Genetic Testing*, which contained articles about genetic testing in a do-it-yourself manner by

4 See Appendix C-2

Table 7-1: Frequency and percentage of topics and stakeholders represented in media coverage.

		CH (%)	UK (%)	All (%)	All (N)
No of articles (total)		**N=38 (27.7%)**	**N=99 (72.3%)**	**100.0%**	**137**
Topic	Genetic Data	9.1%	2.1%	4.1%	8
	Research	5.5%	19.3%	15.4%	30
	Genetics in Society	9.1%	15.7%	13.8%	27
	Public Debates	27.3%	19.3%	21.5%	42
	Legal / Economic Aspects	7.3%	1.4%	3.1%	6
	Ethical Issues	5.5%	2.1%	3.1%	6
	Direct-to-Consumer Genetic Testing	21.8%	5.0%	9.7%	19
	Medical Use of Predictive Genetic Testing	5.5%	5.0%	5.1%	10
	Disclosure of Disease Risk	1.8%	0.7%	1.0%	2
	Personal Stories	0.0%	12.9%	9.2%	18
	Preventive Treatment	5.5%	16.4%	13.3%	26
	Other	1.8%	0.0%	0.5%	1
	Total topics	**100.0% (N=55)**	**100.0% (N=140)**	**100.0%**	**195**
Stakeholder	Media	11.6%	11.5%	11.6%	72
	Research (other)	8.5%	15.9%	13.6%	85
	Natural/medical science	17.5%	16.6%	16.9%	105
	Social science / art & humanities / law	4.8%	0.2%	1.6%	10
	Medical doctor / geneticist / nurse	12.7%	12.4%	12.5%	78
	Companies	5.8%	4.4%	4.8%	30
	Society/individual (other)	0.0%	0.2%	0.2%	1
	Patients & families	3.2%	12.2%	9.5%	59
	Celebrities	6.9%	7.4%	7.2%	45
	Politics	15.3%	1.8%	5.9%	37
	Governmental organizations	6.3%	6.5%	6.4%	40
	Non-governmental organizations	6.9%	10.6%	9.5%	59
	Other	0.5%	0.2%	0.3%	2
	Total stakeholder	**100.0% (N=189)**	**100.0% (N=434)**	**100.0%**	**623**

CH = German-speaking part of Switzerland, UK = United Kingdom.

sending DNA samples and receiving test results directly from a genetic testing company without passing through any health care provider. *Legal and Economic Aspects* of predictive genetic testing and *Ethical Issues* such as discrimination, stigmatization, privacy, data protection, data sharing, and confidentiality issues, were not frequently addressed in either country. Some articles focused on clinical aspects, such as the *Medical Use of Predictive Genetic Testing*, including new tests, healthcare procedures, clinical eligibility criteria for predictive genetic testing, and the *Disclosure of Disease Risk*, including doctor-patient relationships related to counselling and

information giving, the right not to know, the disclosure of incidental findings, and expression of uncertainty in the clinical context.

Genetic Data focused on predictive genetic testing related to Big Data, data sharing platforms, whole-genome sequencing (WGS), and other uses of genetic data in various contexts. This topic was more important in the German-speaking part of Switzerland than in the United Kingdom. Finally, *Personal Stories* did not occur at all in Switzerland but was frequently present in British articles. This topic included examples, cases, and statements of people living with a genetic risk and their families, but excluded celebrity stories, which were included in the topic *Public Debates*.

7.4.2 Evaluations

We also assessed how the different topics were evaluated. British coverage had significantly more positive evaluations (p = .002), whereas in Swiss-German articles, the topics were significantly more often negatively evaluated (p = .016, Figure 7-1A). Switzerland also had a higher ratio of ambivalent articles that covered both positive and negative aspects, but this difference was not statistically significant when applying the Benjamini–Hochberg (BH) correction for multiple testing (p = .046). Evaluations differed considerably between topics, but the trend of the United Kingdom having a higher ratio of positive and a lower ratio of negative articles ranged through nearly all topics (Figure 7-1B). The topic *Research* was significantly more neutral, and the topic *Direct-to-Consumer Genetic Testing* was significantly more negative than the other topics. Regarding stakeholder-specific evaluations (Figure 7-1C), stakeholders from research and medicine were more ambivalent and negative in Swiss-German reporting and more neutral in the United Kingdom. Economic stakeholders generally were more positive than the other stakeholders.

7.4.3 Stakeholders and events influencing media coverage

We collected all stakeholders that gave direct or indirect statements on a topic in each article as an indicator of the influence on media coverage. We coded a total of 623 stakeholders for 195 topics in 137 articles (Table 7-1). Stakeholders cited in coverage were distributed similarly in both countries, except research studies and patients and their families, which were significantly more often cited in the United Kingdom than in the German-speaking part of Switzerland. Instead, Swiss-German coverage had a higher ratio of political stakeholders. In both countries, stakeholders from the economic sectors, namely companies from the pharmaceutical industry, biotech industry, direct-to-consumer genetic testing companies, and health insurers occurred

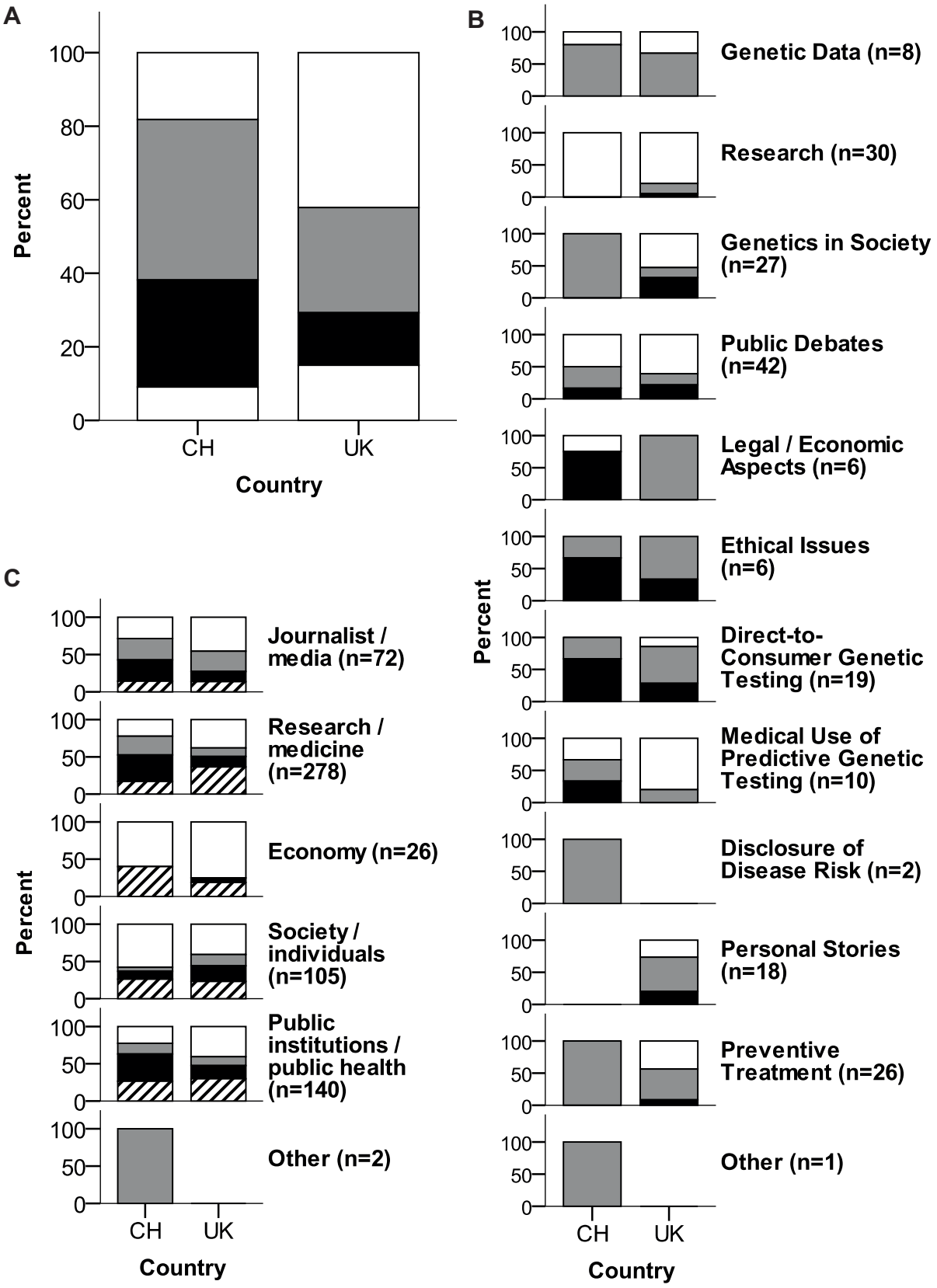

Figure 7-1: Evaluation of topics.
(A) Overall evaluation of topics per country, (B) topic-specific evaluation, and (C) stakeholder-specific evaluation. White = positive evaluation, gray = ambivalent (both positive and negative) evaluation, black = negative evaluation, dashed = neutral (no evaluation).

seldom, and so did experts from research fields of social sciences, humanities, or law. As another measure, we assessed which stakeholder initiated each article (Figure 7-2C). For example, stakeholders organizing press releases or conferences or writing a commentary were considered initiators of the corresponding article. In the United Kingdom, a higher proportion of research studies, as well as affected people and their families, initiated articles in the broadsheet newspapers. In Switzerland, politicians were more often initiators of articles, but not affected people. We also assessed which events triggered media coverage by analyzing the development of media coverage over time (Figure 7-2A). Angelina Jolie's two announcements about her double mastectomy and ovariectomy in May 2013 and March 2015 led to the most important peaks in media coverage of predictive genetic testing. In the United Kingdom, more articles appeared after Angelina Jolie's first announcement than before, and coverage increased even more after the second announcement, indicating that the effect was long-lasting (Figure 7-2B). Angelina Jolie's first announcement also caused coverage in the German-speaking part of Switzerland to peak, but not as much as in the United Kingdom, and there was no long-term increase in coverage after the announcements. Another peak in UK coverage was in the first quarter of 2013. This was mainly coverage of new guidelines and preventive treatment options. In Switzerland, the affair of Professor Ernst Hafen attracted the broadsheet newspaper's attention in the first quarter of 2012. Hafen wanted to launch a public campaign about genetic testing but was criticized in the media.

7.4.4 Connection with theory and empirical evidence

We compared our data with previous empirical studies to address the level of public engagement provided by mass media reporting. Compared to the data from Gerhards and Schäfer (2009), our data revealed a higher ratio of non-scientific stakeholders, and they took more ambiguous positions, particularly in Swiss-German coverage (Table 7-2). Even though UK stakeholders gave more positive assessments than Switzerland, their positioning was not as hegemonial as in Gerhards and Schäfer's study. *Research* was not the only dominant topic in our data. Even though it was an important topic in British coverage, our topic analysis showed that other topics had similar importance, also in the United Kingdom.

We extended the comparison with the medialisation model first applied by Schäfer (2009) (Table 7-3). By comparing our data with the three differently medialized topics in Schäfer's investigation, we are able to rank our data and make estimations about the level of medialisation. The extensiveness of coverage, measured by the number of articles appearing each year, was low

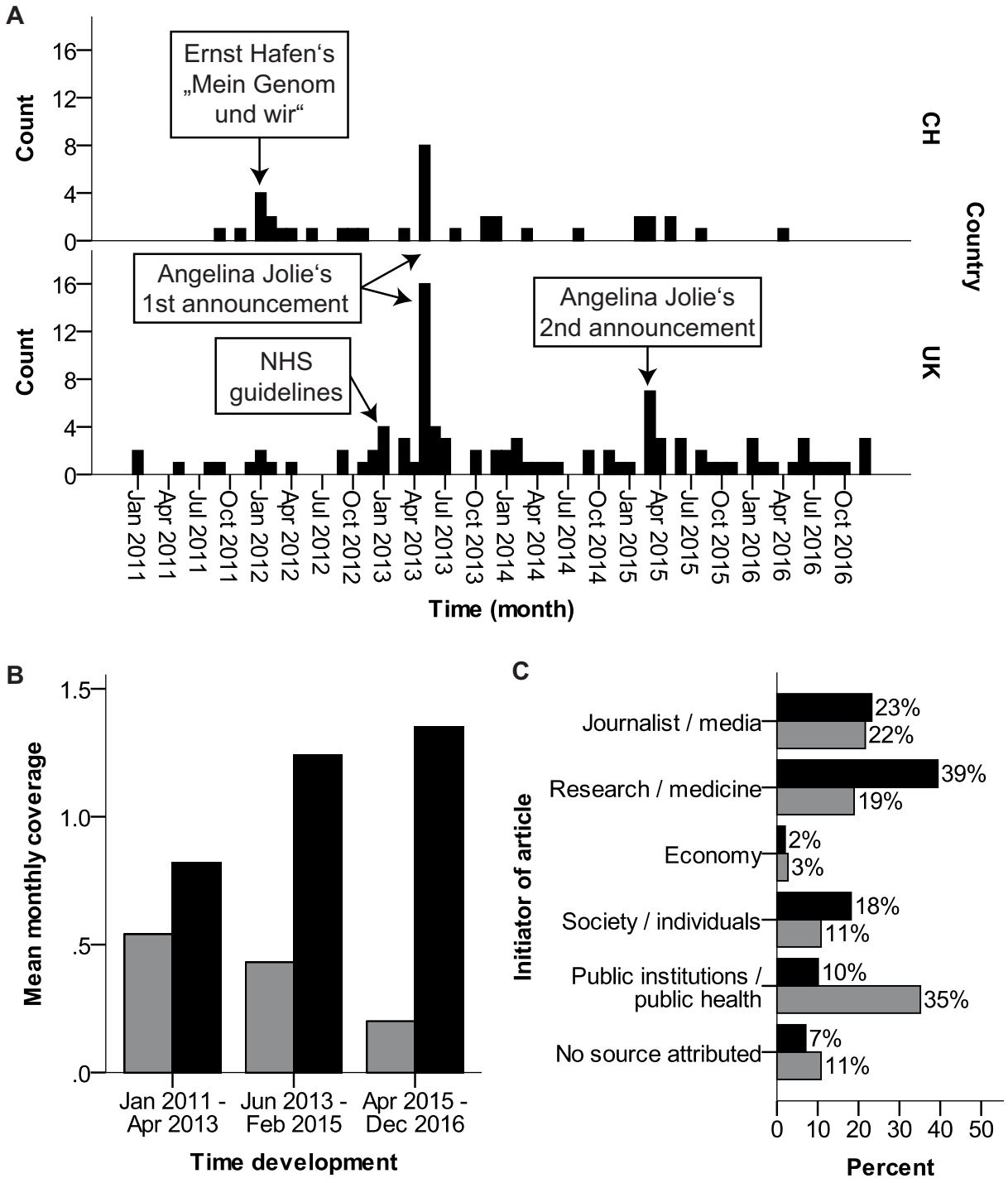

Figure 7-2: Triggers of media coverage.
A) Development of coverage over time (always three months added together for each quarter of the year). B) Mean number of articles per month before, between and after Angelina Jolie's statements in May 2013 and March 2015. C) Stakeholders initiating articles, e.g. by a press conference, press release or interview (only coded if made explicit in the respective article). The percentage was calculated for each country separately.
Black lines/bars = UK, grey lines/bars = German-speaking part of Switzerland.

Table 7-2: Comparison of our data to Gerhards and Schäfer, 2009.

		Zimmermann et al.		Gerhards & Schäfer, 2009	
		CH	**UK**	**Germany**	**USA**
Standing of stakeholders	Media & readers (%)	11.6	11.5	9.2	10.2
	Research in total (%)	43.4	45.1	56.1	54.5
	Natural/biomedical science (%)	17.5	16.6	39.1	48.9
	Social science / art & humanities / law (%)	4.8	0.2	10.4	3.1
	Medical doctor / geneticist / nurse (%)	12.7	12.4	n.c.	n.c.
	Other research stakeholder (%)	8.4	15.9	6.6	2.5
	Economic actors (%)	5.8	4.4	9.8	21.9
	Politics & governmental organizations (%)	21.6	8.3	17.8	10.1
	Civil society (%)	17.0	30.4	6.7	3.4
	Patients & families (%)	3.2	12.2	0.6	0.3
	Celebrities (%)	6.9	7.4	n.c.	n.c.
	Church (%)	0.0	0.0	1.2	0.1
	Non-governmental organizations (%)	6.9	10.6	1.1	0.5
	Other civil society (%)	0.0	0.2	3.8	2.5
	Various (%)	0.5	0.2	0.5	0.0
	N	**189**	**434**	**849**	**745**
Positioning of stakeholders	Positive evaluations (%)	37.3	58.7	49.6	72.9
	Ambivalent evaluations (%)	23.8	18.2	30.9	20.6
	Negative evaluations (%)	38.9	23.0	19.5	6.4
	N (neutral evaluations excluded)	**50**	**119**	**508**	**436**
Framing of topics	Scientific-medical frame (%)	49.2	48.5	57.1	68.5
	Genetic data / WGS (%)	9.1	2.1		
	Research on predictive genetic testing (%)	5.5	19.3		
	Direct-to-consumer genetic testing (%)	21.8	5.0		
	Medical use of predictive genetic testing (%)	5.5	5.0		
	Disclosure of disease risk (%)	1.8	0.7		
	Preventive treatment (%)	5.5	16.4		
	Economic frame (%)	7.3	1.4	6.9	10.5
	Legal / economic aspects (%)	7.3	1.4		
	Political frame (%)	36.4	35.0	9.8	4.6
	Genetics in society (%)	9.1	15.7		
	Public debates about predictive genetic testing (%)	27.3	19.3		
	Socio-ethical frame (%)	5.5	15.0	26.2	16.4
	Ethical issues (%)	5.5	2.1		
	Personal stories (%)	0.0	12.9		
	Other (%)	1.8	0.0		
	N	**55**	**140**		

Note: See Appendix C-3 for more detailed information. n.c. = not collected.

in both countries, especially in the German part of Switzerland, where on average one article appeared every 2 months. The level of pluralization, measured by the ratio of scientific stakeholders and the share of articles appearing in the science section of the newspaper, corresponded to medium or high medialisation. Our data showed a high level of controversy in both countries because a high ratio of statements were evaluative, and there were many ambivalent or negative statements. Swiss reporting was even more ambiguous than in the United Kingdom.

Table 7-3: Comparison of the level of medialisation with a study by Schäfer, 2009.

		Schäfer 2009			Zimmermann et al.	
		Stem Cell Research (medialized)	**Human Genome Research (partly medialized)**	**Neutrino research (not medialized)**	**CH**	**UK**
Extensiveness	Average number of articles per year	319	143	14	6	17
Pluralization	Pluralization: share of research actors (%)	18.3	40.1	86.0	43.4	45.1
	Pluralization: share of science newspaper section (%)	11.7	20.5	72.3	44.7	6.1
Controversy	Share of evaluations of all statements (%)	41.3	47.5	16.4	77.8	70.1
	Positive (%)	46.7	47.4	92.9	37.3	58.7
	Ambivalent (%)	28.4	36.1	7.1	23.8	18.2
	Negative (%)	24.9	16.5	-	38.9	23.0

7.5 Discussion

A large body of literature postulates a transition from passive public understanding to active public engagement with science (e.g., Bucchi 2008; Gerhards and Schäfer 2009; Jones 2011; Schäfer 2009), and we share the opinion that this is a desirable development. Media coverage of predictive genetic testing in both Switzerland and the United Kingdom reflected a more active public engagement than in previous studies (Gerhards and Schäfer 2009; Schäfer 2009). Despite a lack of extensiveness, we conclude that the content of broadsheet newspaper reporting about predictive genetic testing supports active public engagement with the topic. It needs further research doing similar analyses on other scientific topics to examine whether this is a general

trend or topic-related. Predictive genetic testing is of high interest to the general public because of its health-relevance, and it is well possible that this feature facilitates public engagement. Especially for topics with high social and ethical relevance like predictive genetic testing, where the solutions to specific problems (e.g. the availability of testing, disclosure of information, or cost coverage) are not black or white, journalists need to include all stakeholder groups in order to give a solid picture that provides readers with enough information to make opinions that go beyond a simplistic "right-or-wrong" scenario (Hivon et al. 2010). Besides scientific and medical stakeholders, governmental and non-governmental organizations were well represented in both countries' newspaper reporting, and Swiss-German coverage also often represented political stakeholders. The healthcare system in Switzerland is highly political, so it is more prone to discussing medico-scientific topics such as predictive genetic testing in a political manner. The UK NHS is publicly funded and also accountable to the political system, but most NHS-related issues were framed in a societal context rather than as political issues, with political stakeholders only rarely cited. It might be due to the lack of direct democracy in the United Kingdom that politicians are less salient in newspaper coverage.

Research on ethical, legal, and social issues has accompanied genetic research and its translation into practice since the 1980s (Caulfield et al. 2013). A media content analysis reported an intensive discussion on ethical issues in the United Kingdom on coverage about the Human Genome Project in the early 2000s (Smart 2003), and not only journalists but also quoted scientist initiated discussion of these issues (Henderson and Kitzinger 2007). Our results contradict these assessments, showing a lack of ethical topics and researchers cited from social science, humanities, and law in both countries. The public is exposed to the medical and scientific point of views regarding predictive genetic testing, but much less about ethical issues. Since broadsheet newspapers are known to address complex issues and have a higher capacity for well-investigated journalism than other media, we suggest that ethical issues are generally not addressed enough in the media, even though ethical considerations are of crucial relevance for public engagement with science (Miah 2005).

Based on earlier studies (Gerhards and Schäfer 2009; Hjörleifsson, Árnason, and Schei 2008) and the fact that large, international companies from pharmaceutical and biotech industry are headquartered in both countries, we would have expected a higher and more active influence of economic stakeholders in media coverage. Direct-to-consumer genetic testing companies are widespread in the United Kingdom, but not in Switzerland due to legal restrictions, but in both countries they are underrepresented. However, economic stakeholders might choose other

media types such as online media, television, or other public relations measures to place their messages into the public discourse. In addition, industry influence media coverage more indirectly, through lobbying, research collaborations, or targeted advertising in print media (Rinallo and Basuroy 2009; Weishaar et al. 2016). It is thus conceivable that the higher and less controversial coverage of predictive genetic testing in the United Kingdom might be partly due to the presence of direct-to-consumer genetic testing companies. Additional research is needed to reveal the extent of industrial influence on media coverage of genetic testing.

Previous studies suggested that patients are an underrepresented stakeholder group in the public debate (Weigold 2001). Others found that patients and patient organizations were represented in health-related media debates (Hivon et al. 2010). In our data, affected people had a major say in media coverage in the United Kingdom; however, in Switzerland, they were marginalized. This country-specific difference could be due to a possible cultural dependency, it might well be that British newspapers more often use personal stories in order to interest readers. Since personalization is a typical feature of tabloid media, and tabloids are much more established in the media landscape of the United Kingdom than in Switzerland, we suggest that this finding could be due to converging tendencies of broadsheets toward tabloids (Kolb 2015). Personalization in broadsheet newspapers is also higher in the United Kingdom than in Denmark and Spain (Jebril, Albæk, and de Vreese 2013).

Because previous studies suggested that the Angelina Jolie effect, meaning higher testing rates, more preventive mastectomies, and more public interest in predictive genetic testing, lasted at least several years (Evans et al. 2015; Freedman et al. 2017), we tested whether the same applied to media coverage. We found a long-term increase in media coverage in the United Kingdom but not in the German-speaking part of Switzerland. Because of the heterogeneity of topics published in the United Kingdom even after Angelina Jolie's announcements, we suspect that the observed increase in coverage is not exclusively due to the Angelina Jolie effect, but is affected by other factors, such as the increase in the number of scientific studies and the increasing implementation of predictive genetic testing applications within the NHS. Despite that, Angelina Jolie's first statement in May 2013 was the most important trigger for media coverage of predictive genetic testing in both countries. The lack of any long-lasting effect in Swiss-German broadsheet coverage, however, explains only part of the very low coverage about predictive genetic testing.

Swiss-German reporting was strongly characterized by controversial political discussions about direct-to-consumer genetic testing. In contrast, British coverage had a stronger focus on positively evaluated research aspects of predictive genetic testing, a finding in line with previous

research claiming that media hype genetic research (Bubela and Caulfield 2004; Caulfield 2004). Our findings suggest that Swiss-German coverage has a more critical view and is less prone to overhyping the future potential of research for predictive genetic testing. Overall, Swiss coverage reflects a more sceptical attitude toward predictive genetic testing compared to the United Kingdom, a deduction that is supported by the more restrictive policy strategy in Switzerland regarding the implementation of predictive genetic testing both in clinical practice and for direct-to-consumer testing (Swiss Federal Council 2004). To our knowledge, the only comparable survey data about public attitudes toward genetics is from 1997, when Swiss citizens had a more critical view toward gene technologies than other European countries, including the United Kingdom (Gaskell et al. 2000). However, until 2000, public attitudes in Switzerland became considerably more positive, indicating that fast changes in attitudes can occur (Bonfadelli, Dahinden, and Leonarz 2002). A more recent representative survey in Switzerland shows that attitudes in favour of or against genetic testing are in balance (comparis.ch 2012).

7.6 Conclusion

Our results show that newspaper coverage about predictive genetic testing reflects a trend toward more public engagement compared to previous studies on similar topics. However, the extent and nature of coverage varied considerably between the German-speaking part of Switzerland and the United Kingdom. UK coverage was more extensive and more positive. *Personal Stories* and *Research* were predominant foci in UK coverage; in Switzerland, political topics like *Direct-to-Consumer Genetic Testing* had a stronger focus. Research stakeholders were the most prominent stakeholder group, influencing media coverage with statements and coverage-initiating initiatives. Governmental and non-governmental organizations were frequently cited in both countries. In addition, political stakeholders in Switzerland and affected people in the United Kingdom had input into coverage. The Angelina Jolie case was the most important trigger for coverage but was followed by a long-term coverage increase only in the United Kingdom, not in the German-speaking part of Switzerland.

Our study gives important insights into how the public has been informed about predictive genetic testing in recent years. Media coverage reflects an important part of public discourse and is the only aspect that can be measured retrospectively in a consistent, comparable, and reliable manner. Cited stakeholders are a good and indeed the only analyzable indicator to address who is influencing this public discourse through media coverage. Extensive, controversial, and diverse media coverage in terms of aspects covered and stakeholders cited might even reinforce

the public's engagement with predictive genetic testing. Because media coverage is the most important source of information for non-specialists, we would also assume that it also increases the public's general knowledge about this topic. However, our analysis cannot directly reflect the public's knowledge of or attitudes toward predictive genetic testing. These factors need to be assessed through representative population surveys, and our data can inform the content of such research. An analysis of Swiss public discourse and attitudes toward biotechnology some 20 years ago revealed that media coverage reflected the public attitudes well at that time (Bonfadelli et al., 2002).

To avoid in-country language bias, our analysis was limited to the leading German-speaking newspapers in Switzerland, not addressing their French- and Italian-speaking counterparts. Because of different cultural and political attitudes in the different Swiss language regions, it is important to state that our results are valid for the German-speaking Swiss newspapers, which cover the majority (65%) of the Swiss population, but cannot necessarily be expanded to the whole country. Because we compared German with English-speaking newspaper coverage, there is a possible bias due to differences in language. However, comparing countries with such differences can also reveal interesting differences for broader applications allowing our results to be to some extent transferable to and comparable with other Western countries. Certain findings might also be biased due to the rather small sample size of our study. We only covered broadsheets in our analysis, despite the fact that online media and television are other important information sources for non-specialists. The high-quality broadsheet newspapers used in this study are known to have a focus on science communication more than other news media and serve as an information source for journalists as well. Thus, they are influencing the media content across the media system. We also aimed to include broadsheet newspapers that are representative of the newspaper landscape of the respective country, but it is possible that results would look different if we had selected other newspapers for our analysis.

Our study adds up to the empirical evidence of public engagement with science. We show that it is worthwhile making data collection comparable to previous studies because it provided context and knowledge by comparison with other scientific topics as well as between different countries. Public engagement with science seems to have increased in the media in the last decade, according to our data. Furthermore, our results are useful for science communication scholars, who can learn which aspects are lacking in terms of successful public engagement with the issues raised by predictive genetic testing. Researchers, clinical geneticists, and other stakeholders can also obtain insights into where and how to enhance their communication activities with

the mass media. In particular, we recommend that researchers from the fields of ethics, law, and social sciences intensify their communication activities in the mass media. Because their work has a high societal impact, their representation in media would help to produce richer, more holistic coverage about predictive genetic testing in both countries (Miah 2005). Acknowledging that this call is not easily accomplished and requires a lot of effort, time, manpower, special training, and collaborations with journalists, we also call on the responsible authorities to provide these researchers with the necessary financial, educational, and technological tools.

7.7 References

Almomani, Basima, Ahmed F. Hawwa, Nicola A. Goodfellow, Jeffrey S. Millership, and James C. McElnay. 2015. "Pharmacogenetics and the Print Media: What Is the Public Told?" BMC Med Genet 16:32. doi:10.1186/s12881-015-0172-3.

Bennet, David J., and Richard C. Jennings, eds. 2011. Successful Science Communication: Telling It Like It Is. Cambridge: Cambridge University Press.

Berg-Schlosser, Dirk. 2003. "Makro-Qualitative Vergleichende Methoden." In Vergleichende Politikwissenschaft, edited by Dirk Berg-Schlosser and Ferdinand Müller-Rommel. 4th ed., 103–25. Opladen: Leske + Budrich.

Bonfadelli, Heinz. 2002. "Gentechnologie im Urteil der Bevölkerung: Agenda-Setting - Wissensklüfte - Konsonanzeffekte." In Gentechnologie in der öffentlichen Kontroverse: Eine sozialwissenschaftliche Analyse, edited by Heinz Bonfadelli and Urs Dahinden, 47–96. Zürich: Seismo.

Bonfadelli, Heinz, Urs Dahinden, and Martina Leonarz. 2002. "Biotechnology in Switzerland: High on the Public Agenda, but Only Moderate Support." Public Underst Sci (11): 113–30.

Borzekowski, Dina L. G., Yue Guan, Katherine Smith, Lori H. Erby, and Debra L. Roter. 2014. "The Angelina Effect: Immediate Reach, Grasp, and Impact of Going Public." Genet Med 16 (7): 516–21. doi:10.1038/gim.2013.181.

Bubela, Tania M. 2006. "Science Communication in Transition: Genomics Hype, Public Engagement, Education and Commercialization Pressures." Clin Genet 70 (5): 445–50. doi:10.1111/j.1399-0004.2006.00693.x.

Bubela, Tania M., and Timothy A. Caulfield. 2004. "Do the Print Media "Hype" Genetic Research? A Comparison of Newspaper Stories and Peer-Reviewed Research Papers." Can Med Assoc J 170 (9): 1399–1407. doi:10.1503/cmaj.1030762.

Bucchi, Massimiano. 2008. "Of Deficits, Deviations and Dialogues: Theories of Public Communication of Science." In Handbook of Public Communication of Science and Technology, edited by Massimiano Bucchi and Brian Trench. Routledge international handbooks. . Edited by Massimiano Bucchi. London: Routledge.

Catalan-Matamoros, Daniel, and Carmen Peñafiel-Saiz. 2017. "The Use of Traditional Media for Public Communication About Medicines: A Systematic Review of Characteristics and Outcomes." Health Commun, 1–9. doi:10.1080/10410236.2017.1405485.

Caulfield, Timothy A. 2004. "Biotechnology and the Popular Press: Hype and the Selling of Science." Trends Biotechnol 22 (7): 337–39. doi:10.1016/j.tibtech.2004.03.014.

Caulfield, Timothy A., Subhashini Chandrasekharan, Yann Joly, and Robert Cook-Deegan. 2013. "Harm, Hype and Evidence: ELSI Research and Policy Guidance." Genome Med 5 (3): 21. doi:10.1186/gm425.

comparis.ch. 2012. "Zwei Drittel Wollen Keinen Gentest Machen: Comparis.Ch Zur Akzeptanz Von Analysen Des Eigenen Erbguts." Medienmitteilung. Accessed January 27, 2020. https://www.comparis.ch/-/media/files/mediencorner/medienmitteilungen/2012/krankenkasse/akzeptanz-gentests.pdf.

Condit, Celeste M. 2001. "What Is 'Public Opinion' About Genetics?" Nat Rev Genet 2 (10): 811–15. doi:10.1038/35093580.

Davies, Sarah R. 2013. "Constituting Public Engagement: Meanings and Genealogies of PEST in Two UK Studies." Sci Commun 35 (6): 687–707. doi:10.1177/1075547013478203.

Esser, Frank. 2000. "Journalismus vergleichen: Journalismustheorie und komparative Forschung." In Theorien des Journalismus: Ein diskursives Handbuch, edited by Martin Löffelholz. 1st ed., 123–46. Wiesbaden, s.l. Verlag für Sozialwissenschaften.

Evans, D. Gareth R., Julie Wisely, Tara Clancy, Fiona Lalloo, Mary Wilson, Richard Johnson, Jonathon Duncan, Lester Barr, Ashu Gandhi, and Anthony Howell. 2015. "Longer Term Effects of the Angelina Jolie Effect: Increased Risk-Reducing Mastectomy Rates in BRCA Carriers and Other High-Risk Women." Breast Cancer Res. 17 (1): 143. doi:10.1186/s13058-015-0650-8.

Evers-Kiebooms, Gerry, Erna Claes, Lieve Denayer, and Marleen Decruyenaere. 2001. "Predictive Genetic Testing: Psychological Impact." In Encyclopedia of Life Sciences. Chichester, UK: Wiley Publishing.

Federal Office of Public Health. 2019. "The Compulsory Health Insurance in a Nutshell: Your Questions, Our Answers." Accessed January 27, 2020. https://www.bag.admin.ch/dam/bag/en/dokumente/kuv-aufsicht/krankenversicherung/sie-fragen-wir-antworten-oblig-kv.pdf.download.pdf/broschuere-sie-fragen-wir-antworten-e.pdf.

Fishman, Jessica M., and David Casarett. 2006. "Mass Media and Medicine: When the Most Trusted Media Mislead." Mayo Clin Proc 81 (3): 291–93. doi:10.4065/81.3.291.

Freedman, Rebecca, Helen Mountain, Dian Karina, and Lyn Schofield. 2017. "A Retrospective Exploration of the Impact of the 'Angelina Jolie Effect' on the Single State-Wide Familial Cancer Program in Perth, Western Australia." J Genet Couns 26 (1): 52–62. doi:10.1007/s10897-016-9982-5.

Früh, Werner, and Klaus Schönbach. 1982. "Der Dynamisch Transaktionale Ansatz: Ein Neues Paradigma Der Medienwirkungen." Pub 27: 75–88.

———. 2005. "Der Dynamisch-Transaktionale Ansatz III: Eine Zwischenbilanz." Publizistik 50 (1): 4–20. doi:10.1007/s11616-005-0115-7.

Garrett, Jinnie M., and Stephanie J. Bird. 2007. "Ethical Issues in Communicating Science." Sci Eng Ethics 6 (4): 435–42. doi:10.1007/s11948-000-0001-7.

Gaskell, George, Nick Allum, Martin W. Bauer, John Durant, Agnes Allansdottir, Heinz Bonfadelli, Daniel Boy et al. 2000. "Biotechnology and the European Public." Nat Biotechnol 18:935 EP -. doi:10.1038/79403.

Gerhards, Jürgen, and Mike S. Schäfer. 2007. "Hegemonie der Befürworter. Der mediale Diskurs über Humangenomforschung." Soz Welt 58 (4): 367-395.

———. 2009. "Two Normative Models of Science in the Public Sphere: Human Genome Sequencing in German and US Mass Media." Public Underst Sci 18 (4): 437–51. doi:10.1177/0963662507082891.

gov.uk. 2020. "How Government Works." Accessed January 27, 2020. https://www.gov.uk/government/how-government-works.

Hallin, Daniel C., and Paolo Mancini. 2011. Comparing Media Systems Beyond the Western World: Cambridge University Press.

Haran, Joan, and Jenny Kitzinger. 2013. "Human Genetics and Cloning in the Media: Mapping the Research Field." In Handbook of Genetics and Society: Mapping the New Genomic Era, edited by Paul Atkinson, Peter Glasner, and Margaret Lock. Abington: Routledge.

Henderson, Lesley, and Jenny Kitzinger. 2007. "Orchestrating a Science 'Event': The Case of the Human Genome Project." New Genet Soc 26 (1): 65–83. doi:10.1080/14636770701218175.

Hivon, Myriam, Pascale Lehoux, Jean-Louis Denis, and Melanie Rock. 2010. "Marginal Voices in the Media Coverage of Controversial Health Interventions: How Do They Contribute to the Public Understanding of Science?" Public Underst Sci 19 (1): 34–51. doi:10.1177/0963662508088668.

Hjörleifsson, Stefán, Vilhjálmur Árnason, and Edvin Schei. 2008. "Decoding the Genetics Debate: Hype and Hope in Icelandic News Media in 2000 and 2004." New Genet Soc 27 (4): 377–94. doi:10.1080/14636770802485467.

Holton, Avery E., Heather E. Canary, and Bob Wong. 2016. "Business and Breakthrough: Framing (Expanded) Genetic Carrier Screening for the Public." Health Commun, 1–8. doi:10.1080/10410236.2016.1196515.

Jarren, Otfried, and Martina Vogel. 2011. "„Leitmedien" Als Qualitätsmedien: Theoretisches Konzept Und Indikatoren." In Krise Der Leuchttürme Öffentlicher Kommunikation: Vergangenheit Und Zukunft Der Qualitätsmedien, edited by Roger Blum, Heinz Bonfadelli, Kurt Imhof, and Otfried Jarren, 17–30. Wiesbaden: Verlag für Sozialwissenschaften.

Jebril, Nael, Erik Albæk, and Claes H. de Vreese. 2013. "Infotainment, Cynicism and Democracy: The Effects of Privatization Vs Personalization in the News." Eur J Comm 28 (2): 105–21. doi:10.1177/0267323112468683.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jones, Richard A. L. 2011. "Introduction: Public Engagement in an Evolving Science Policy Landscape." In Bennet and Jennings 2011, 1–14.

Kamenova, Kalina, Vardit Ravitsky, Spencer McMullin, and Timothy A. Caulfield. 2016. "Media Portrayal of Non-Invasive Prenatal Testing: A Missing Ethical Dimension." J Sci Comm 15 (02): A03. doi:10.22323/2.15020203.

Kamenova, Kalina, Amir Reshef, and Timothy A. Caulfield. 2014. "Angelina Jolie's Faulty Gene: Newspaper Coverage of a Celebrity's Preventive Bilateral Mastectomy in Canada, the United States, and the United Kingdom." Genet Med 16 (7): 522–28. doi:10.1038/gim.2013.199.

Kolb, Steffen. 2004. "Voraussetzungen für und gewinnbringende Anwendung von quasiexperimentellen Ansätzen in der kulturvergleichenden Kommunikationsforschung." In Einführung, Problematisierungen und Aspekte der Methodenlogik aus kommunikationswissenschaftlicher Perspektive. Vol. 1, edited by Werner Wirth, Edmund Lauf, and Andreas Fahr, 157–78. Forschungslogik und -design in der Kommunikationswissenschaft. Köln: Herbert von Halem.

———. 2005. Mediale Thematisierung in Zyklen: Theoretischer Entwurf und empirische Anwendung. Köln: Herbert von Halem.

———. 2015. Vielfalt Im Fernsehen: Eine Komparative Studie Zur Entwicklung Von TV-Märkten in Westeuropa. Forschungsfeld Kommunikation 35. Köln: Herbert von Halem.

Lebo, Patricia B., Franz Quehenberger, Lars-Peter Kamolz, and David B. Lumenta. 2015. "The Angelina Effect Revisited: Exploring a Media-Related Impact on Public Awareness." Cancer 121 (22): 3959–64. doi:10.1002/cncr.29461.

Lewis, Celine, Mahrufa Choudhury, and Lyn S. Chitty. 2015. "'hope for Safe Prenatal Gene Tests'. A Content Analysis of How the UK Press Media Are Reporting Advances in Non-Invasive Prenatal Testing." Prenat Diagn 35 (5): 420–27. doi:10.1002/pd.4488.

Locke, Simon. 2011. "Deficits and Dialogues: Science Communication and the Public Understanding of Science in the UK." In Bennet and Jennings 2011, 17–30.

Lynch, John, Ashley Parrott, Robert J. Hopkin, and Melanie Myers. 2011. "Media Coverage of Direct-to-Consumer Genetic Testing." J Genet Couns 20 (5): 486–94. doi:10.1007/s10897-011-9374-9.

Maier, Scott. 2010. "All the News Fit to Post? Comparing News Content on the Web to Newspapers, Television, and Radio." J Mass Commun Q 87 (3-4): 548–62. doi:10.1177/107769901008700307.

Mayring, Philipp. 2015. Qualitative Inhaltsanalyse: Grundlagen Und Techniken. 12 rev. ed. Weinheim: Beltz.

Metag, Julia, and Mike S. Schäfer. 2016. "Wissenschaftsbarometer Schweiz." Accessed January 27, 2020. https://wissenschaftsbarometer.ch/wp-content/uploads/2016/09/IPMZ_Wissenschaftsbarometer-1.pdf.

Miah, Andy. 2005. "Genetics, Cyberspace and Bioethics: Why Not a Public Engagement with Ethics?" Public Underst Sci 14 (4): 409–21. doi:10.1177/0963662505056616.

National Health Service. 2020. "About the NHS." Accessed January 27, 2020. https://www.nhs.uk/using-the-nhs/about-the-nhs/.

O'Mahony, Patrick, and Mike S. Schäfer. 2005. "The 'Book of Life' in the Press Comparing German and Irish Media Discourse on Human Genome Research." Soc Stud Sci 35 (1): 99–130. doi:10.1177/0306312705046542.

Patzelt, Werner J. 1997. Einführung in Die Politikwissenschaft: Grundriß Des Faches Und Studiumsbegleitende Orientierung. 3rd ed. Passau: Rothe.

Rinallo, Diego, and Suman Basuroy. 2009. "Does Advertising Spending Influence Media Coverage of the Advertiser?" J Marketing 73 (6): 33–46. doi:10.1509/jmkg.73.6.33.

Rowe, Gene, and Lynn J. Frewer. 2005. "A Typology of Public Engagement Mechanisms." Sci Technol Human Values 30 (2): 251–90. doi:10.1177/0162243904271724.

Schäfer, Mike S. 2009. "From Public Understanding to Public Engagement: An Empirical Assessment of Changes in Science Coverage." Sci Commun 30 (4): 475–505. doi:10.1177/1075547008326943.

Schwitzer, Gary, Ganapati Mudur, David Henry, Amanda Wilson, Merrill Goozner, Maria Simbra, Melissa Sweet, and Katherine A. Baverstock. 2005. "What Are the Roles and Responsibilities of the Media in Disseminating Health Information?" PLoS Med 2 (7): 576–82. doi:10.1371/journal.pmed.0020215.

Scott, William A. 2009. "Scott's Pi." In the Content Analysis Reader, edited by Klaus Krippendorff and Mary A. Block, 347–49. Thousand Oaks, Calif: SAGE Publications.

Smart, Andrew. 2003. "Reporting the Dawn of the Post-Genomic Era: Who Wants to Live Forever?" Sociol Health Illn 25 (1): 24–49. doi:10.1111/1467-9566.t01-1-00323.

Stilgoe, Jack, Simon Locke, and James Wilsdon. 2014. "Why Should We Promote Public Engagement with Science?" Public Underst Sci 23 (1): 4–15. doi:10.1177/0963662513518154.

Swiss Federal Council. 2004. "CC 810.12 Federal Act of 8 October 2004 on Human Genetic Testing (HGTA)." Accessed January 23, 2020. https://www.admin.ch/opc/en/classified-compilation/20011087/index.html.

———. 2017. "Direct Democracy." Accessed January 23, 2020. https://www.eda.admin.ch/aboutswitzerland/en/home/politik/uebersicht/direkte-demokratie.html.

Teune, Henry, and Adam Przeworski. 1970. The Logic of Comparative Social Inquiry. New York: Wiley-Interscience.

Weigold, Michael F. 2001. "Communicating Science: A Review of the Literature." Sci Commun 23 (2): 164–93. doi:10.1177/1075547001023002005.

Weingart, Peter. 2005. Die Wissenschaft Der Öffentlichkeit: Essays Zum Verhältnis Von Wissenschaft, Medien Und Öffentlichkeit. Weilerswist: Velbrück.

Weishaar, Heide, Lori Dorfman, Nicholas Freudenberg, Benjamin Hawkins, Katherine Smith, Oliver Razum, and Shona Hilton. 2016. "Why Media Representations of Corporations Matter for Public Health Policy: A Scoping Review." BMC Public Health 16:899. doi:10.1186/s12889-016-3594-8.

Wirth, Werner, and Steffen Kolb. 2004. "Designs and Methods of Comparative Political Communication Research." In Comparing Political Communication: Theories, Cases, and Challenges, edited by Frank Esser, 87–111. Communication, society, and politics. Cambridge: Cambridge University Press.

———. 2013. "Securing Equivalence: Problems and Solutions." In the Handbook of Comparative Communication Research, edited by Frank Esser and Thomas Hanitzsch, 469–85. International communication association (ICA) handbook series: Routledge.

Chapter 8: Media Coverage of Ethical Issues in Predictive Genetic Testing: A Qualitative Analysis

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8.1 Abstract

Background: Predictive genetic testing (PGT) raises many ethical issues and is of increasing interest to the general population. Mass media, especially newspapers, are the public's main source of information on this topic. **Methods**: We conducted a content analysis of British newspaper reporting, assessing which ethical issues were mentioned. The analysis was qualitative with semi-quantitative aspects. All articles about PGT published in *The Guardian* and the *Daily Telegraph* from 2011 to 2016 were included. **Results**: Most ethical issues discussed in the scientific and ethical literature are implicitly or explicitly covered in newspapers, but there was no discussion of incidental findings, and the possibility of false reassurance of a negative test result was mentioned only once. There are also important gaps regarding the multidimensional nature and complexity of many issues. *The Guardian* mentioned ethical issues more frequently than the *Daily Telegraph*. Most ethical issues were portrayed as first-person narratives. **Conclusions**: Ethical issues concern potential test users and society more than scientific background knowledge about such tests; therefore, more efforts should be taken to address these complex issues in a manner that is comprehensible for the lay public.

8.2 Introduction

Predictive genetic testing (PGT) raises many ethical issues which have been discussed at length in hundreds of papers in medical and ethics journals. Studies on the content and effects of media coverage of genetic testing have broadly tended to focus on scientific advances (Gerhards and Schäfer 2007; Henderson and Kitzinger 2007) and celebrities such as Angelina Jolie (Kamenova, Reshef, and Caulfield 2014; Borzekowski et al. 2014) who have used new technologies and spoken publicly about their experiences. Another topic that has been investigated is direct-to-consumer (DTC) genetic testing (Lynch et al. 2011; Vayena 2015), but there is generally little recent emphasis on the important ethical issues raised by PGT (Craig 2000).

PGT yields information regarding the likelihood that a person will develop a particular disease at some point in the future. Other kinds of genetic tests have been excluded from this analysis because they entail different specific ethical issues, such as diagnostic testing (which is used to rule out or confirm whether a patient has a particular suspected condition) or carrier testing (used to establish whether a person carries a gene mutation that could cause a genetic disease if he or she has a child with another carrier). Another excluded type is pharmacogenomic testing, where the response, effectiveness or dose of a patient's medication is personalized based on his or her genetic make-up.

PGT is offered both through clinicians and through DTC genetic testing companies. For the former, a medical indication is needed, and recipients are accompanied by medical specialists such as genetic counsellors throughout the testing process. The latter is often offered online, and recipients can send in a saliva sample themselves and receive test results directly, usually without personal medical counselling. Even though US legislation has restricted DTC genetic testing for medical purposes in the past (Yim and Chung 2014), certain conditions can now be disclosed to customers (U.S. Food and Drug Administration 2017, 2018). Unlike many other European countries, the UK does not require genetic counselling for PGT and has no restrictions on DTC genetic testing services. However, genetic counselling is provided when PGT is done in a clinical context (Kalokairinou et al. 2018). Given that people who use PGT are not ill, calling them "patients" does not seem appropriate. In the following, we will call those with a mutation-positive PGT result "affected individuals".

PGT has become more widespread in the last two decades, accompanied by much ethical discussion in the literature, which generally focuses much more on the problematic, harmful issues of PGT than on the beneficial ones. PGT is deemed beneficial when it enables preemptive medical treatment or leads to psychological relief (Lolkema et al. 2013). Knowledge about one's genetic risks can also confer benefit by enabling different decisions to be made about one's life, including reproductive decisions (Yarborough, Scott, and Dixon 1989; Wertz et al. 2003). In other cases, however, a mutation-positive test result can be devastating, particularly if that disease cannot be treated or prevented (Fulda and Lykens 2006; Hamilton, Lobel, and Moyer 2009). PGT might also offer false reassurance to mutation-negative people, with poor lifestyle choices resulting (Jackson, Goldsmith, and Skirton 2014). Harm can also be done to affected individuals by unnecessary preventive medical interventions (Evans, Skrzynia, and Burke 2001). Moreover, PGT frequently does not provide certainty but results in risk predictions, which may cause insecurity and anxiety (Wertz and Fletcher 1991; Evans, Skrzynia, and Burke 2001).

Disclosing genetic information to asymptomatic people is generally seen as respecting their autonomy, and some people assert their right to know certain genetic facts about themselves (Lolkema et al. 2013). It can, however, paradoxically also lead to reduced autonomy in the context of lack of voluntariness, lack of alternatives or regarding determination and authenticity (Huibers and van `t Spijker 1998; Millum 2014). Genetic counselling has a special emphasis with regards to PGT in order to improve understanding and enable informed consent (Marteau and Croyle 1998; Lolkema et al. 2013). In contrast, some people do not want to know anything about their own genetic disease risks, drawing upon their "right not to know" (Andorno 2004).

Employers and providers of health and life insurance (and mortgages) have in the past, discriminated against people because of genetic test results. While this is generally prohibited by legislation in most jurisdictions now, it remains an important ethical issue with regards to PGT (Harris, Winship, and Spriggs 2005). Decisions about publicly funding PGT also raise concerns of distributive justice in terms of resource allocation (American Society of Clinical Oncology 2003). Not everyone can access PGT easily, which raises issues of equality and justice (Ormond 2008).

Disclosing a genetic test to asymptomatic family members poses the ethical dilemma of respecting their right not to know or potentially preventing harm (Haupt 2018; Fulda and Lykens 2006; Lucassen and Parker 2010; Rothstein 2018; Elger, Michaud, and Mangin 2010). PGT for minors regarding adult-onset diseases is generally perceived as taking away children's autonomy regarding the right not to know (Mand et al. 2012; Harris, Winship, and Spriggs 2005; Caulfield et al. 2015; Elger 2010a; Millum 2014).

These ethical issues have a major impact both on societies and individuals (Miah 2005). With the rise of DTC genetic testing, the public is becoming aware of such testing. Therefore, including the public in the discourse about ethical issues is becoming not only preferable but a requirement. The function of public discourse is to empower the public to participate in decision-making regarding the future role of genetic testing (Weishaar et al. 2016) and to make informed decisions for themselves (Bergsma 2004). However, an informed discourse between the public and the research community is only possible when there is a general awareness of the topic and its ethical issues. Mass media play an important role in raising this awareness, and newspapers are a particularly common source of information for the public about complex scientific and medical issues, such as PGT (Garrett and Bird 2007; Schwitzer et al. 2005).

With this analysis, we thus aim to assess how two UK broadsheet newspapers have addressed the most important ethical issues about PGT. We will semiquantitatively compare which ethical aspects were prioritized in the newspapers, identify which aspects have been underrepresented or missing, and compare the two newspapers analyzed. We also make recommendations regarding how to improve media coverage about ethical issues in PGT in the future. More specifically, we aim to give answers to the following questions: Is newspaper coverage enabling individuals to make informed choices? Is newspaper coverage contributing to a fruitful public debate about societal aspects of PGT? This is, to our knowledge, the first attempt to assess how bioethical issues are portrayed in mass media, using an original methodological design.

8.3 Methods

We included all newspaper articles about PGT in two UK broadsheet newspapers: the left-liberal *The Guardian* and the right-conservative *Daily Telegraph*. Both have a comprehensive science section, represent good-quality journalism, are read nationwide and have wide coverage when compared to other UK broadsheet newspapers. We included articles published between 1 January 2011 and 31 December 2016, because we were interested in articles about Angelina Jolie's public statements that she had a preventive double mastectomy and hysterectomy (Jolie 2013; Jolie Pitt 2015) as well as articles before and after these events. The articles were selected through a keyword search in the online database Factiva (Dow Jones, New York, United States). We used the following search algorithm:

> ((*Angelina* or *Jolie*) AND (genet* OR gene OR effect* OR BRCA* OR mastectomy or ovar*)) OR BRCA* OR gene* test* OR Lynch syndrome* OR ((personalized medicine OR personalized health) AND (genet* or genom*)) OR ((((predictive OR preventive) AND (genet* OR genom*)) OR genetic* predispose*) AND test*)

Asterisks represent truncations in the Factiva search algorithm software. Articles were included for analysis if the topic predictive genetic or genomic testing on born humans, its social, economic, or political implications, or its consequences were the main focus of the article. The main focus was defined as being at least 20% of text in the article body. Articles about epigenetics, PGT before birth, and post-symptomatic genetic testing were excluded from analysis. Furthermore, we did not include articles about PGT on animals or plants; non-medical applications of PGT such as forensics, evolutionary genetics, or ancestry-search; or – because of the lack of relevant information – articles shorter than 100 words. The search algorithm and article selection were developed in a previous quantitative analysis (Zimmermann et al. 2019), during which an extensive, multi-step procedure resulted in the indicated search algorithm. The search algorithm aimed to capture all articles about PGT in the selected time period for the given newspapers, aiming to achieve maximal sensitivity. Specificity was obtained through the extensive selection process, which was conducted by two researchers separately and then compared. The search algorithm led to 691 articles after removal of duplicates, of which 99 articles matched our inclusion criteria (see Table D-1 in Appendix D for a list of these included articles).

We performed a deductive content analysis of the sampled newspaper articles (Elo and Kyngas 2008). The analysis was qualitative with semi-quantitative aspects, meaning that the ethical issues can be ranked in terms of their appearance frequency, but statistical analyses and defined numbering are not possible. In order to avoid over-interpretation, all articles were coded by two

researchers separately, but inter-coder reliability testing revealed that the coding was not reliably quantifiable. However, we aimed to capture every ethical issue mentioned in the data by systematically applying the codes; thus semi-quantitative analysis is possible.

To inform the development of our coding scheme, a literature review was conducted to ensure that all ethical issues concerning PGT that have been discussed in the medical and ethical literature were included. A PubMed search was conducted using the terms "ethics" and "genetic testing", the most highly-cited articles were read, and all ethical issues relating to PGT were listed. These articles were also used in a snowball approach, with references scanned to identify other relevant articles.

Using the codes established through the scientific literature review (see Table D-2 in Appendix D for the coding scheme), we then assigned the newspaper text passages to the codes. All articles were read by two bioethics researchers separately, and all text passages that include an ethical perspective were assigned to one of the four principles. Any thoughts, interpretations, or comments were noted for further analysis. Several codes were given to the same text passage if several ethical issues applied. Because we realized that these deductive codes did not always match the newspaper content, we added an inductive step, thereby identifying new (sub)topics and slightly reorganizing the codes according to our data. MaxQDA 2018 was used for coding and both qualitative and quantitative analyses. This study did not require IRB approval since it uses data previously published in British newspapers.

8.4 Results

A total of 99 articles were ultimately included for analysis. In 22 articles, no ethical statements were identified. The majority of text passages identified as ethically relevant were first-person narratives, mainly from affected individuals or experts. The principle of beneficence was the most commonly coded principle – we identified aspects related to beneficence in 64 of 99 analyzed articles. In contrast, the principle of nonmaleficence was only represented in 30 articles. Autonomy was another aspect that received important media attention; it was represented in 51 articles. The principle of justice was represented in 25 articles. In the following section, we qualitatively present how the different ethical issues were mentioned in the broadsheet newspapers, allocating them to ethical principles. Figures 8-1 and 8-2 provide a semi-quantitative analysis of the ethical issues, allowing the reader to get an impression on how much salience the ethical issues had in relation to each other.

8.4.1 Principle of beneficence

Saving lives by preventing disease

The medical benefit of PGT was the most salient topic in newspaper coverage. The main benefits mentioned were connected to the prevention of harm. Preventing diseases and deaths by identifying people with increased risks was often framed in connection with genetic research and public health aspects such as screening, but also with taking preventive measures such as surgery or medication. Three articles also mentioned the benefit of raising awareness regarding PGT and its consequences, in close connection to the principle of autonomy (Table 8-1). Psychological benefits mentioned in newspaper coverage were the feeling of reassurance, better coping through better understanding and the feeling of being able to change one's destiny. They were mainly mentioned by affected individuals talking about their experiences, but they were not as extensively discussed as medical benefits.

Preventing harm to future generations

Reproductive benefits were framed through the possibility of preventing harm to future generations, either by not having children due to a genetic risk or by testing them early in life or through preimplantation genetic testing, where embryos are genetically screened for certain genetic alterations during the process of in-vitro fertilization in order to avoid implanting an embryo with a disease-causing mutation.

Hope and belief in genetic research progress

Another common aspect was genetic research progress, which was framed as a benefit in media coverage. This aspect was not only mentioned in articles about genetic research but also expressed by affected individuals and from a societal point of view as hopes for a better future. The utility of PGT was also often connected to future applications and research (Table 8-1). A total of 13 articles mentioned the balance between risks and benefits, and the aspect of cost and cost-effectiveness was included in 5 of them.

8.4.2 Principle of nonmaleficence: anxiety, heritage and uncertainty

Psychological harm was mainly addressed through statements of affected individuals. They most often reported about anxiety and the distress of passing on the risk gene to their children (Table 8-2). In addition, psychological harm was discussed in connection with DTC genetic testing. While most statements concerned warnings about possible anxiety because of test results, DTC

Table 8-1: Ethical issues connected to the principle of beneficence mentioned in newspaper coverage.

Code	Expressions within the media articles	Example quote
Medical benefit	Harm prevention by preventing disease Harm prevention by identifying people at risk Harm prevention by genetic screening Harm prevention by raising awareness	"'Those [genetic test] results made it very easy for me to opt for a bilateral mastectomy – and it was a decision that ultimately saved my life.' [Clare Delaney, tested positive for inherited breast cancer gene]" (The Daily Telegraph, September 15, 2012) "In all, there is no doubt that lives were saved because of Jolie's decision to talk publicly about her surgery." (Pemberton, September 22, 2014)
Psychological benefit	Better coping through better understanding Relief when tested negative Being able to change your destiny	"Mr Anderson, who in the same test found out he was at a slightly higher than average risk of developing stomach cancer, says it was 'reassuring to know one way or the other.'" (Ensor, April 11, 2015)
Reproductive benefit	Preventing harm from future generations (Not) having children due to genetic risk	"'If I do find out I have the gene, I'll have my eggs tested before having a child. […] Our family has had to live with this horrible disease for generations, but we'll be the ones who'll change things for the future. We're going to make sure that the terrible difficulties our parents and grandparents had to live with will never touch the lives of our children and grandchildren. I feel quite proud of that.'" [Megan Smith, 16, daughter of a Huntington's Disease patient] (Moorhead, September 22, 2012)
Genetic research progress	More harm prevention in the future through research progress Genetics will revolutionize healthcare	"Experts said last night that the findings were a 'promising' and exciting prospect, which could in future help protect women." (Donnelly, June 27, 2014)
Utility	Balancing risks and benefits Cost-benefit assessment	"'Although the cost of a full genome sequence is getting to be very affordable, robust interpretation of what it means for an individual lags behind [...] There's a lot of promise about tailoring treatments, or modifying lifestyles, depending on particular genetic variation, but to date, with a few exceptions, the clinicial utility of genome sequencing is limited.' [Anneke Lucassen, professor of clinical genetics at Southampton University]" (Sample, December 29, 2011)

company speakers were sometimes cited, stating that anxiety among DTC test users has not been severe (Robinson, January 12, 2016; Brennan, June 13, 2016). Another aspect that was prone to causing psychological harm (according to media coverage) was testing for susceptibility of non-preventable or non-treatable diseases such as Alzheimer's or Huntington's disease. The aspect of uncertainty regarding PGT, while being significant in the scientific literature, was

only marginally discussed in media coverage. In some statements, the uncertainty was generalized to medical outcomes or risks in general. Only a few statements were about the uncertainty a positive predictive genetic test result specifically brings to its recipients.

Aspects of medical harm mentioned in newspaper articles included unnecessary preventive treatments and their side-effects. These aspects were mostly mentioned in connection to the Angelina Jolie case. Medical harm was not a very common aspect compared to medical benefit (Figure 8-1), and coverage focused on preventive treatments following the genetic test, not the predictive genetic test itself.

8.4.3 Principle of respect for autonomy

"Knowledge is power"

The newspapers picked up the issue of knowledge and understanding by citing medical experts mentioning the importance of thinking carefully about the consequences before taking a test,

Table 8-2: Ethical issues connected to the principle of nonmaleficence mentioned in newspaper coverage.

Code	Expressions within the media articles	Example quote
Psychological harm	Passing on genes to children Anxiety connected to test result Anxiety connected to DTC genetic testing Testing for diseases lacking preventive options	"To find out my daughter was a carrier too, it was like my world was crumbling around me." (Brennan, June 13, 2016)
		"'It's like being told you're going to be involved in a car crash, but you don't know when it will hit. You know it is going to happen, but you can't do anything about it,' says Amy Burton." (Brennan, June 13, 2016)
		"Despite 23andMe's careful use of language and explanation, there is an understandable concern that this type of genetic testing could cause inappropriate harm simply through people worrying excessively or becoming neurotic over these small increases in risk." (Gibbs, October 02, 2015)
Uncertainty	Incomplete knowledge Not certain whether disease develops	"The risk of someone who is positive for BRCA mutation developing breast cancer is estimated at around 60 per cent. It's an agonising decision for women who find themselves in this situation. No one can say for sure whether they will be in the lucky 40 per cent." (Pemberton, January 28, 2013)
Medical harm	Genetic tests: false reassurance Preventive treatments: side effects Unnecessary preventive treatments	"Both operations are major undertakings with inherent risk in the surgery as well as potential long-term consequences." (Donnelly, June 27, 2014)

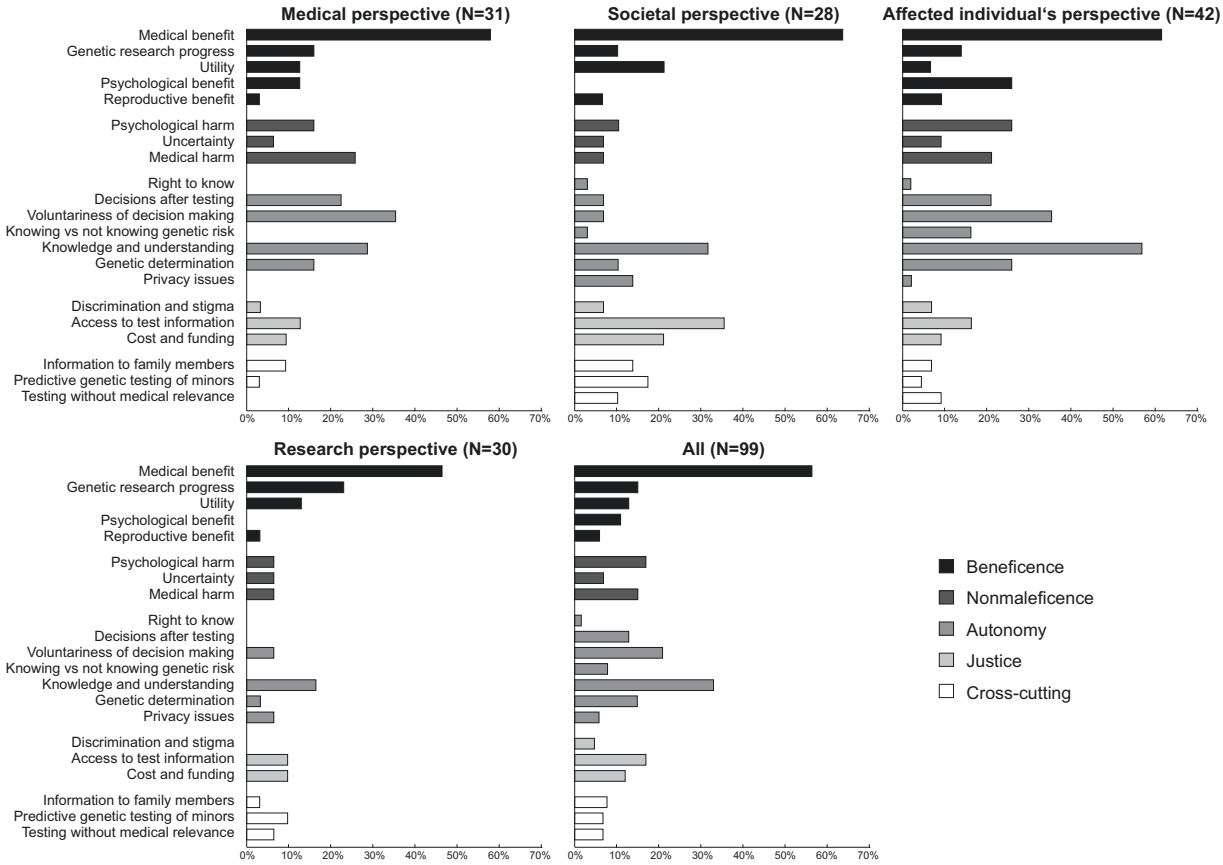

Figure 8-1: Percentage of articles where ethical issues appeared (at least once). N=number of articles included in the analysis. One article can contain several perspectives. Perspectives where obtained by summarizing topics identified and published in Zimmermann et al. (2018). Medical perspective = Medical use of PGT + Disclosure of disease risk + Preventive treatment; societal perspective = Legal/economic aspects + ethical aspects + Public health + DTC genetic testing (2 articles); research perspective = Genetic data + Research; affected individuals' perspective = Public debates + Case stories + DTC genetic testing (5 articles). Articles have been double-checked for suitability of these perspectives.

and of seeking medical advice before taking preventive surgery (Table 8-3). Genetic counselling was mentioned a few times in this context. DTC genetic testing was also mentioned in the context of understanding genetic test information and getting proper information, with both positive (participants understand the testing well) and negative assessments (some information should not be provided without professional counselling). The Angelina Jolie case triggered media to mention extensively the aspects of awareness-raising and informed choice regarding PGT and its possible preventive consequences. The statements were predominantly positive towards this awareness-raising.

The tension between knowing and not knowing genetic risks

Ambivalence regarding the reasons in favour of or against taking a test was mentioned in affected individuals' statements and mainly connected to a genetic predisposition to untreatable

Table 8-3: Ethical issues connected to the principle of respect for autonomy mentioned in newspapers.

Code	Expressions within the media articles	Example quote
Knowledge and understanding	Awareness-raising Genetic information is powerful Getting proper information prior to and after testing Information through counselling Informed choice Understanding test information	Experts hail Jolie for going public on the private affair of her life-saving treatment: Actor raises awareness of test for defective gene (Pilkington, Meikle, and Ridley, May 15, 2013b)
		"With the BRCA gene a woman's risk of developing cancer changes with age – it is very important to have someone like a genetic counsellor to talk through the consequences with." [Katherine Taylor, CEO of charity Ovarian Cancer Action] (Brennan, June 13, 2016)
		For Burton, knowledge is, indeed, power: "There are days when I wish I hadn't signed up to the trial, but I wouldn't rewind time. I think I'm better being informed. At least this way I feel better prepared and more in control of my life." [Amy Burton, genetically at risk for diabetes] (Brennan, June 13, 2016)
		"'If you know what your genetic likelihoods are you can have a better planned experience of your time on Earth.' [Philip Hodson, UK Council for Psychotherapy]" (Carey, March 07, 2015)
Right to know	People have a right to know their genetic risks	"She [Anne Wojcicki, head of 23andme] believes people have a right to know about their genetic risks, so they can make informed lifestyle decisions."(Ensor, April 11, 2015)

diseases, i.e., Alzheimer's or Huntington's disease. A few statements by DTC companies explicitly mentioned the right to know (Ensor, April 11, 2015). Lack of alternatives was perceived by affected individuals, but the media focused mainly on the situation after a positive test result was received. The voluntariness of decision making was mentioned in connection to both the decision regarding whether to have preventive surgery and the decision of whether to test.

More choice or inevitable determination?

The question of whether predictive genetic test results were determining people's lives, thereby decreasing autonomy by preventing self-authentic decisions due to uncertain knowledge about the future, was discussed ambiguously. One position represented in four articles was that PGT does indeed diminish the autonomy of its users in this way (Table 8-1). In more articles, however,

Table 8-3 (continued).

Code		Expressions within the media articles	Example quote
Decision making	Knowing vs not knowing genetic risks	Difficult decision: tension between knowing or not knowing genetic risk Reasons to (not) do genetic testing	"'Sometimes I can hardly bear to just wait and see what happens,' says Carrol. 'I think, I've got to know - I've got to find out whether my children have this. But then I think, if they tested positive, how would I begin to handle that? It's the most horrific cloud to have to live under.'" (Moorhead, June 02, 2015)
	Voluntariness of decision making	Difficulty to decide for or against preventive surgeries Celebrities' influence on affected individuals' decisions Emphasis that decisions must be taken individually --> testing minors Hope that advances make decisions easier in the future	"Then the woman asked for something that Moalem, a Toronto-based American physician and geneticist, couldn't give her. "She said to me: 'I want to test my children but I don't want to tell them the results.' I said: 'My hands are tied. In most countries, you can't test children like that - it's taking away their autonomy.'" (Jeffries, April 05, 2014)
	Decisions after testing	Decisions about consequences after testing positive Perceived lack of alternatives after testing positive	"I've made the decision to have the mastectomy sooner rather than later as my looming 30th birthday was becoming a bit of a timebomb." (Hirst, August 13, 2015)
Determination and authenticity		Decreased autonomy: predictive genetic tests determine people's life vs Increased autonomy: proactive choices, empowerment	"It is a world inhabited by 'previvors' and 'post-humans': men and women who have been screened for genetic vulnerabilities or created with altered genetic propensities. Illness might progressively vanish, but so might identity. [...] Chance would become mitigated, but so, inevitably, would choice." (Mukherjee, May 21, 2016)
Privacy issues - data storage		Data storage Access by third parties (insurance, employer, banks)	"As the costs of genetic testing plummet and as it continues to get easier to do, we'll face more of these situations," says Moalem. "To hack or not to hack into the genome is the question we'll increasingly be faced with." (Jeffries, April 05, 2014)

the opposite was stated, specifically that PGT actually increased the autonomy of its users, particularly in connection to Angelina Jolie case. Privacy issues were mentioned in the context of the use of genetic data and often framed as an important open issue that needs discussion regarding how to handle access to genetic information in the future in order to respect the privacy of individuals.

8.4.4 Principle of justice

Fear of discrimination and stigma

The discriminatory or stigmatizing misuse of genetic test information by employers, health or life insurances, or for mortgages was mostly framed as a threat for people with a positive genetic test result (Table 8-4). This was despite the fact that such discrimination is forbidden in the UK (Murray–West, April 06, 2013).

Measuring cost-effectiveness of PGT applications

Costs of predictive genetic tests and their funding through public healthcare systems were not a central topic. Media mentioned the cost-effectiveness of PGT and preventive treatments for both society and individuals in six articles. Another aspect mentioned was cost coverage for genetic testing and follow-up tests by insurance, especially regarding future scenarios when genetic testing becomes more prominent.

Equality of access: a challenge for the NHS

Some 16 articles mentioned the issue of equality of access to testing and preventive treatment. Unequal access to certain genetic screening tests throughout the UK was the main issue in several articles. Other articles mentioned that distributive justice of genetic testing was promoted

Table 8-4: Ethical issues connected to the principle of justice mentioned in newspaper coverage.

Code	Expressions within the media articles	Example quote
Discrimination and stigma	Discrimination by insurances Stigma attached to male breast cancer	"'Deciding whether to have the test is really huge because it has massive implications if the test is positive […]. It can make it difficult to get a mortgage or life insurance and cause employment problems.'" [son of a Huntington's disease patient] (Moorhead, September 22, 2012)
Costs and funding	Cost-effectiveness of genetic testing Cost coverage by the public healthcare system Patenting: companies are making money	"This treatment [a preventive drug for breast cancer] is potentially not just cost–effective but cost–saving to the NHS […]" (Collins, June 25, 2013)
Access by recipients	Unequal access to genetic screening programs Unequal access because of missing cost coverage Requirements to get a test offered	"'It is my hope that they, too, will be able to get gene tested, and that if they have a high risk they, too, will know they have strong options,' she [Angelina Jolie] writes, while acknowledging the issues of financial access that prevent too many women from getting tested and treated."(Freeman, May 15, 2013a)

by Angelina Jolie; in particular, she advocated to make testing affordable for everybody. Accessibility has been framed as increased when tests get cheaper or are covered by health insurance.

8.4.5 Cross-cutting issues

Information provision to family members about genetic risks

Whether and how family members should be informed about a relative's positive genetic test result is a highly discussed topic in the scientific community. The ethical dilemma about the duty to warn versus respecting the tested person's privacy has not been picked up by the media. Instead, they frame cascade testing and screenings as a purely beneficial procedure that should be promoted (Table 8-5). One article mentioned the possible negative implications of a genetic test for all family members regarding possible discrimination.

PGT of minors infringes their autonomy

Testing minors was framed mainly as potentially taking away their autonomy to be tested before they can decide for themselves. There were two articles emphasizing benefits of testing children, both in a research context: one showing good results for preventing heart attacks, and another sequencing sick's children's genomes to help future children.

Pros and cons of DTC genetic testing

The issue of PGT without medical indication was mainly framed in the context of DTC genetic testing. Benefits were mentioned mainly by DTC companies, stating, e.g., that DTC genetic testing empowered people and made them engage with genetics. It was also mentioned that DTC genetic testing improved access due to its low price. Potential harm often was brought up by experts and officials, who expressed concern about a possible lack of understanding and anxiety after receiving PGT results without professional counselling. It was furthermore stated that DTC genetic testing information was misleading and unreliable.

8.4.6 Neglected aspects in media coverage

In general, we found that most ethical issues around PGT discussed in the scientific community were also implicitly or explicitly represented in newspaper coverage of the two included newspapers. However, the issue of incidental findings was completely absent from our newspaper sample, even though it has great ethical implications in relation to PGT according to scientific literature. The duty to warn family members of affected individuals about potential harm also was marginalized. Merely one article mentioned the risk of false reassurance of a negative test

result, and another one the risk of false-positive results. The issue of patenting a genetic test was not a central issue in our data either. Two articles mentioned it as a critical issue, one of them not only from a justice but also from an autonomy perspective.

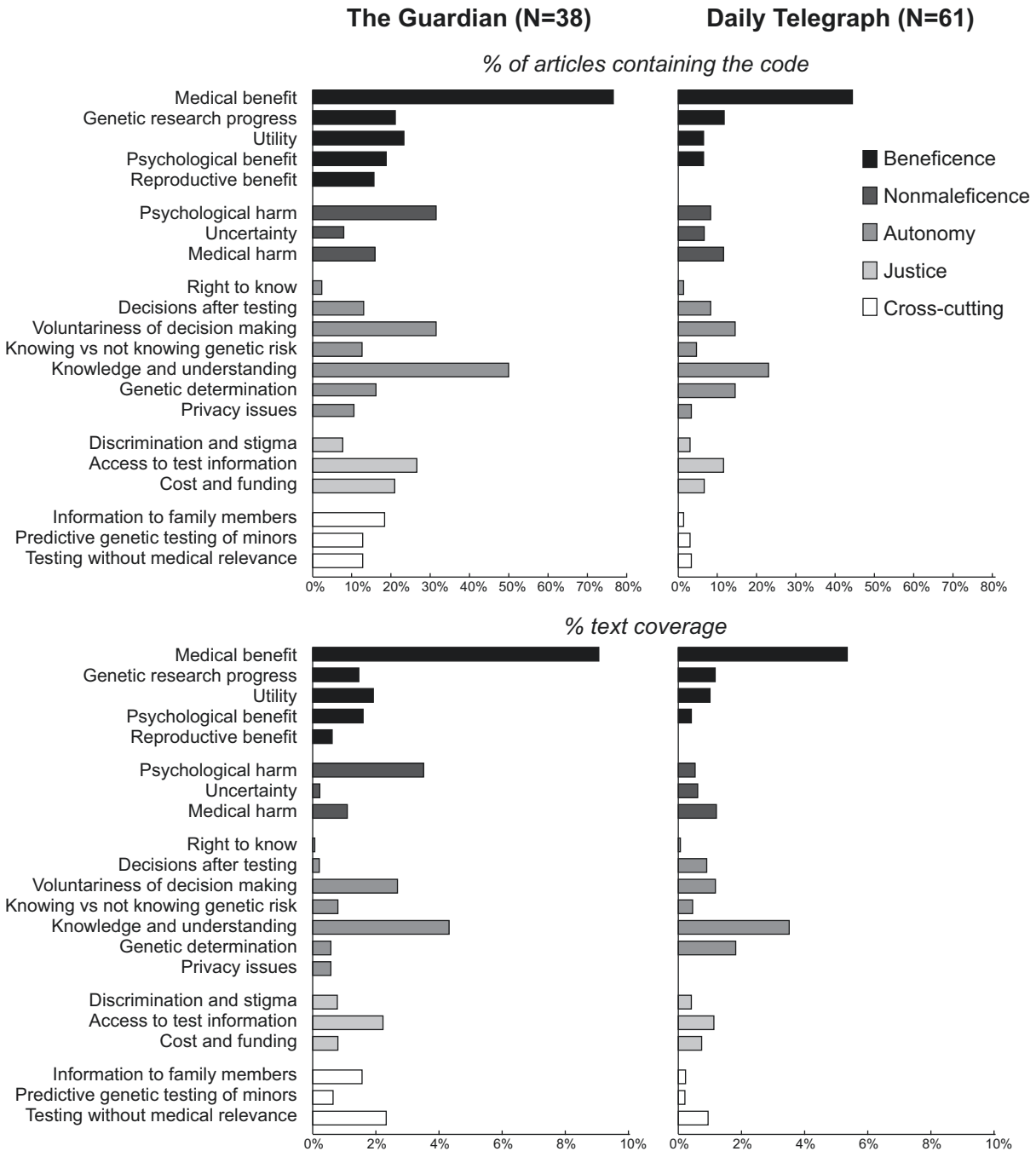

Figure 8-2: Comparison between the left-liberal *The Guardian* and the right-conservative *Daily Telegraph*.
Two indicators were used to measure the extent of coverage for each ethical issue: the percentage of articles where an ethical issue was mentioned at least once; and the percentage of text coverage of each code, the total being all text of the articles. N = number of articles included in the analysis.

8.4.7 Comparison between the *Daily Telegraph* and *The Guardian*

Figure 8-2 shows a comparison between the two analyzed newspapers. Even though *The Guardian* published fewer articles about PGT (38 versus 61 for the *Daily Telegraph*), there were generally more codes given per article, a tendency true for all ethical issues. *The Guardian* focused considerably more on medical benefits and psychological harm and had more articles, including the information given to family members.

Table 8-5: Issues connected to cross-cutting ethical issues mentioned in newspaper coverage.

Code	Expressions within the media articles	Example quote
Information to family members about genetic risks	Confidentiality issues Benefits of screening whole families Duty to warn family members	"'People don't realize they have made it more difficult for their family members to get insurance.' [geneticist Dr. Sharon Moalem]" (Jeffries, April 05, 2014)
		"'[…] Cascade screening is an effective way of finding FH patients and thus saving lives. This is preventive medicine.'" [Steve Humphries, professor of cardiovascular genetics at University College London] (Campbell, January 23, 2013)
		"Do you have an ethical duty to tell other family members?" (Robinson, January 12, 2016)
Testing minors	Taking away autonomy by testing children Identifying at-risk minors early for suitable treatment	"'We see genetics as being the future of medicine. From the time we are born we will know how at risk we are of disease. I think this will happen within my lifetime,' said Prof Bowcock. 'Obviously there are ethical problems that run alongside that, particularly for parents. But it would mean many diseases could be caught early and treated.' (Knapton, January 10, 2014)
		"She said to me: 'I want to test my children but I don't want to tell them the results.' I said: 'My hands are tied. In most countries, you can't test children like that - it's taking away their autonomy.'" [geneticist Dr. Sharon Moalem]" (Jeffries, April 05, 2014)
PGT without medical indication	DTC benefits: empowerment of individuals, better access, engagement DTC risks: lack of understanding, anxiety, misleading information	'DNA isn't something scary or all about the disease, which is why we try to celebrate the long-tail of humanity – why is it some people like coriander and other fun things – rather than DNA is just about cancer or Alzheimer's,' said Wojcicki [head of 23andme]. (Gibbs, October 02, 2015)
		"Despite 23andMe's careful use of language and explanation, there is an understandable concern that this type of genetic testing could cause inappropriate harm simply through people worrying excessively or becoming neurotic over these small increases in risk." (Gibbs, October 02, 2015)

8.5 Discussion

Most ethical issues were covered by newspapers and were relevant and relatable for the recipients because they were mainly transmitted through first-person narratives.

However, they were not all covered to the same extent, raising some ethical issues that we mention below. The case of Angelina Jolie pushed the aspect of autonomy regarding PGT in media coverage – it was Angelina's narrative to take control over one's life, to explore options, and to make informed decisions, and media picked up this narrative extensively after the actress' statements (Jolie, May 13, 2013; Jolie Pitt, March 25, 2015). Over the last few decades, there has been a general shift within healthcare from medical paternalism, with more of an emphasis on beneficence and nonmaleficence, towards a greater focus on respect for autonomy, mirroring the increasing emphasis in Western society on individualism and "consumer choice". This is in line with the finding from an analysis of the empirical scientific literature (Elger 2010b) that the focus on beneficence is still present, but the perception is that granting patients autonomy benefits them: knowing one's genetic predisposition and making decisions based on that knowledge is seen to create psychological benefit as compared to the uncertainty of not knowing (Elger 1998; Wiggins et al. 1992). Newspaper coverage related to informing family members about inheritable genetic risks mostly supports this as well: Respecting the autonomy of those who do not want to know was mentioned rarely, and the benefits of knowing genetic risks running in the family were emphasized.

Issues related to nonmaleficence (potential harms related to PGT) were mentioned less than those concerning beneficence in newspaper coverage, indicating a rather positive assessment of PGT and its consequences. This finding is supported by a quantitative study on the same data material, showing that UK media coverage was predominantly positive (Zimmermann et al. 2019). In particular, the benefit of being able to prevent diseases and save lives by PGT was very salient. In contrast, medical harm caused by PGT was not very highly emphasized, even though the harm resulting from medical interventions after PGT is an important issue in the ethical and medical literature. This aspect is underrepresented in media coverage. In addition, media often connected the risk of psychological harm to the testing procedure: there is a risk of psychological harm attached to PGT without professional counselling. The fact that these psychological issues also arise in clinical settings and that genetic counsellors are taking a lot of effort and research to address and investigate these issues (Vadaparampil et al. 2007; Houfek et al. 2015) is not reflected in newspaper coverage, indicating a potential bias.

Not surprisingly, articles with a research perspective mentioned fewer ethical issues than articles featuring affected individuals', societal, or medical perspectives. Mostly, these articles focused on the progress of scientific research, which was predominantly framed as beneficial for future patients and society.

8.5.1 Is newspaper coverage facilitating informed decision-making?

With the increasing spread of PGT, be it DTC or in a clinical setting, making informed choices becomes more important for laypeople on an individual level. One central issue here is the decision regarding whether to have a predictive genetic test. This decision is very individual, and much debate has focused on non-directive counselling, which aims to let affected individuals make their own decisions (Pennacchini and Pensieri 2011; Aston 1998). It was implied by media that decision-making about whether to do a predictive genetic test or not was personal and needed consideration, but the essential elements that may play a role in this decision-making process were rarely discussed. For preventable diseases, newspaper articles only referred to cases where people such as Angelina Jolie had already made a decision. Moreover, the decision not to receive a genetic test result was only connected to diseases that were untreatable and unpreventable, such as inherited Alzheimer's disease or Huntington's disease, potentially leaving readers with the wrong impression that deciding against PGT is otherwise not really an option that is often taken. This finding leads us back to the issue of a perceived lack of alternatives (Huibers and van `t Spijker 1998). At least for diseases where there is some medical benefit attached to PGT, the newspaper narrative implies that there is no real choice.

8.5.2 Is newspaper coverage contributing to a fruitful public debate about societal aspects of PGT?

There were considerable differences in the treatment of ethical issues between the two newspapers analyzed, with the left-liberal *The Guardian* mentioning ethical issues more frequently, despite having fewer articles on this topic. This might be because the right-conservative *Daily Telegraph* published a higher ratio of research-related articles that were neutral and thus likely not to treat any ethical issues. *The Guardian* also had a higher ratio of articles that contained ambivalent statements. It is also possible that *The Guardian*'s coverage is more in-depth, and thus reflects ethical issues more. Whether it is generally true that media coming from a left-liberal political spectrum reflect more on ethical issues needs to be confirmed with other topics and other media.

Some ethical issues with a potentially high impact on society were only framed from an individualistic, but not a societal perspective. Thus, even though we found references to these issues, they were not discussed on a societal meta-level. For example, decision-making aspects were mentioned rarely on the meta-level. Furthermore, the prevention of discrimination is an important and current issue for society and stakeholders (Abel et al. 2005) but was not very salient from a societal perspective. In the UK, the discussion about this justice issue remains mainly confined to experts. In order to reflect the public's point of view in future policymaking and to enable people to be aware of and protect themselves against discrimination, more effort needs to be made to discuss this issue publicly. In contrast, issues about equal access and public funding were more frequently discussed from a societal perspective.

The aspect of determination through PGT was quite explicitly discussed in media coverage on several occasions, even though this issue is complex and far from self-explanatory or intuitive. In addition, coverage about this issue was ambiguous, with some statements claiming that PGT was "[…] far from deterministic" (Ensor, April 11, 2015), while others drew a rather pessimistic picture of a future where life decisions are determined by genetic risk information (Mukherjee, May 21, 2016). This issue is not only a personal, but also a societal one, and a public debate about this issue is crucial if the public should be included in the decision making process on what role predictive genetic tests should play in our healthcare systems in the future. Our data illustrate that it is possible to discuss even such complex issues in newspaper coverage, and we encourage more coverage on this aspect in the future.

It is particularly important that conflicts between ethical principles are publicly discussed because they often lead to dilemma situations where there is no easy right or wrong answer. To achieve good solutions both on a societal and individual level, it is beneficial to include the public in the process of solving such dilemmas. Unfortunately, our analysis showed that media often do not explicitly explain these ethical dilemmas. For the dilemma related to the disclosure of information to family members, the media mainly describe this as being beneficial for the family members. Only rarely was the underlying ethical dilemma involving the right not to know touched upon, and it was never explicitly and concisely explained to readers in our sample. Similarly, the issue of incidental findings, which illustrates a dilemma between the benefits of knowing a genetic risk and the freedom of making informed autonomous decisions on the one hand, with the potential psychological harm and right not to know on the other, has not been mentioned at all.

The dilemma surrounding PGT for children has been discussed more explicitly in several articles (Knapton, January 10, 2014; Jeffries, April 05, 2014). Also, the issues around PGT without medical indication were discussed ambiguously in connection with the pros and cons of DTC genetic testing. Ethical dilemmas such as disclosure to family members, incidental findings, the right not to know versus the right to know PGT information, and ethical problems such as discrimination, equality of access, and coverage of PGT and subsequent preventive measures are not sufficiently represented even though they are becoming more important for policymaking.

8.5.3 Limitations

Our analysis included all articles published in The *Daily Telegraph* and *The Guardian* about PGT and its consequences 2011-2016. Even though both newspapers are of national and international relevance, are broadsheet newspapers prone to provide high-quality journalism, and have a high number of readers, they represent only part of the broad British media landscape. Many recipients nowadays receive information via online news portals, TV, or radio, which we did not include for this analysis. Given that the media landscape can look considerably different from country to country, the analysis needs to be performed in other settings to make it comparable. Moreover, we chose a qualitative methodology because we wanted to get enough depth in our analysis to grasp details, and the often implicit nature of media mentioning ethical issues also made it difficult to have reliable quantitative measures for some variables. This means that our findings cannot be fully generalizable, even though we aimed to reduce subjective bias by having two researchers conduct the analysis separately before reaching consensus. Because of the systematic application of our deductively derived codes, we were able to apply a semi-quantitative analysis on the frequency of code appearance.

8.6 Conclusion

Overall, our results suggest that media coverage of PGT taken collectively does cover most of the ethical issues identified in the scientific and ethical literature. However, the differential focus on particular topics paints a rather rosy picture of the technology and its application. Specifically, articles preferentially mentioned potential medical benefit over potential harm and focused on psychological harm rather than physical harm. Where psychological harm was mentioned, it tended to be in the context of DTC. There were gaps in coverage of key issues concerning autonomy (right not to know and incidental findings) and little emphasis on justice-related issues such as discrimination. This is quite a biased, prescriptive picture that tends to depict PGT as

something that should normally be done. The failure to provide a detailed, balanced picture for newspaper readers is ironic given the emphasis on knowledge, understanding, and autonomy in some of the articles (and the omission of incidental findings and right not to know as an issue particularly). It is ironic in the sense that, if newspapers seek to inform readers fully as their stories suggest potential users of PGT should be informed, the lack of balance indicates that they are failing to do so. If the right to know is mentioned and the right not to know is not, this creates an imbalanced sketch of rights and thus of the ethical landscape.

Our analysis is qualitative and contains semi-quantitative elements, and only concerned two newspapers. It is, to our knowledge, the first study that assessed how media treat ethical issues of a biomedical topic, and we suggested a new methodological design. There was a clear preponderance of statements regarding beneficence and autonomy, and the lesser focus nonmaleficence and justice. It is concerning that such a bias exists even in serious newspapers known for high-quality journalism in which readers trust the quality of information. Taken together, these mean that the take-home message from much of the coverage is that PGT can enhance autonomy and save lives. This is true, but there are caveats, and those caveats seem to be underreported. To some extent, the narrative concerning PGT seems biased towards positive aspects, thus encouraging readers towards using PGT.

If newspaper coverage of ethical issues in PGT is biased, why might this be the case? It may be because careful consideration and contextualization are less interesting to readers, and more positive stories sell newspapers (Caulfield 2005).

Alternatively, it might simply be difficult to communicate complex ethical issues in the limited format of a newspaper article. A cynic might even suggest that PGT is profitable, and newspapers generally serve the capitalist model by encouraging consumerism.

Finally, is it ethically problematic for newspapers to under-report the ethical downsides of PGT? Journalists have a duty to report facts accurately, and while newspapers may be (generally) getting the science right, they are not yet reporting ethical issues properly. This is an important ethical issue because the current perspective of much coverage emphasizes the positive ethical issues and neglects more challenging ones like unnecessary surgery, incidental findings, and the right not to know. These are important aspects to consider for society –probably more important than exactly how a genetic test actually works (Miah 2005). Of course, in the age of the internet, people can use Google to find out about genetics and related issues by themselves. But the internet is also full of misinformation, and the fact that facts can be obtained elsewhere does not excuse journalists from the duty to present an objective perspective. However, journalists

and others working news media should not shoulder this burden alone. Doctors, scientists, and bioethicists also have a duty to engage more with journalists and the public about ethical issues related to PGT. Some of these ethical issues are complex, and if the public is to understand them, experts will need to be involved in helping to explain them.

8.7 References

Abel, Elizabeth, Sharon D. Horner, Diane Tyler, and Sheryl A. Innerarity. 2005. "The Impact of Genetic Information on Policy and Clinical Practice." Policy Polit Nurs Pract 6 (1): 5–14. doi:10.1177/1527154404272143.

American Society of Clinical Oncology. 2003. "American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility." J Clin Oncol 21 (12): 2397–2406. doi:10.1200/JCO.2003.03.189.

Andorno, Roberto. 2004. "The Right Not to Know: An Autonomy Based Approach." J Med Ethics 30 (5): 435-9; discussion 439-40. doi:10.1136/jme.2002.001578.

Aston, Robert. 1998. "Genetic Counselling Must Be Non-Directive." BMJ 317 (7150): 82. doi:10.1136/bmj.317.7150.82.

Bergsma, Lynda J. 2004. "Empowerment Education." Am Behav Sci 48 (2): 152–64. doi:10.1177/0002764204267259.

Borzekowski, Dina L. G., Yue Guan, Katherine Smith, Lori H. Erby, and Debra L. Roter. 2014. "The Angelina Effect: Immediate Reach, Grasp, and Impact of Going Public." Genet Med 16 (7): 516–21. doi:10.1038/gim.2013.181.

Brennan, Sharon. 2016. "Are DIY Gene -Testing Kits a Good Idea??" *The Guardian*, June 13.

Campbell, Denis. 2013. "Blood Screening That Is Preventing Heart Attacks - but Not in England: NHS Elsewhere in Britain Is Tracking down and Treating People with Inherited Condition." *The Guardian*, January 23.

Carey, Tanith. 2015. "Should You Worry About Your Genes?" The *Daily Telegraph*, March 7.

Caulfield, Timothy A. 2005. "Popular Media, Biotechnology, and the Cycle of Hype the Mass Media's Influence on Health Law and Policy Symposium." Houston J Health Law Policy 5: 213-233.

Caulfield, Timothy A., Pascal Borry, Maeghan Toews, Bernice S. Elger, Henry T. Greely, and Amy McGuire. 2015. "Marginally Scientific? Genetic Testing of Children and Adolescents for Lifestyle and Health Promotion." J Law Biosci 2 (3): 627–44. doi:10.1093/jlb/lsv038.

Collins, Nick. 2013. "Breast Cancer Drugs Offered to Women at High Risk." The *Daily Telegraph*, June 25.

Craig, David A. 2000. "Ethical Language and Themes in News Coverage of Genetic Testing." J Mass Commun Q 77 (1): 160–74. doi:10.1177/107769900007700112.

Donnelly, Laura. 2014. "Women Offered Hope of a Blood Test That Can Predict the Risk of Breast Cancer Years Ahead." The *Daily Telegraph*, June 27.

Elger, Bernice S. 1998. Médecine Prédictive Et Décisions Procréatives Et Prénatales. Chêne-Bourg: Médecine & Hygiène.

———. 2010a. "Ethical, Legal and Social Issues in the Genetic Testing of Minors." In Handbook of Genomics and the Family: Psychosocial Context for Children and Adolescents, edited by Kenneth P. Tercyak, 485–521. Boston: Springer.

———. 2010b. Le Paternalisme Médical: Mythe Ou Réalité? Aspects Philosophiques Et Empiriques D'un Phénomène Persistant. Chêne-Bourg: Médecine & Hygiène.

Elger, Bernice S., Katarzyna Michaud, and Patrice Mangin. 2010. "When Information Can Save Lives: The Duty to Warn Relatives About Sudden Cardiac Death and Environmental Risks." Hastings Cent Rep 40 (3): 39–45. doi:10.1353/hcr.0.0254.

Elo, Satu, and Helvi Kyngas. 2008. "The Qualitative Content Analysis Process." J Adv Nurs 62 (1): 107–15. doi:10.1111/j.1365-2648.2007.04569.x.

Ensor, Josie. 2015. "'after Watching My Father Lose His Memory, I Had to Know My Fate.'" The *Daily Telegraph*, April 11.

Evans, James P., Cécile Skrzynia, and Wylie Burke. 2001. "The Complexities of Predictive Genetic Testing." BMJ 322 (7293): 1052–56. doi:10.1136/bmj.322.7293.1052.

Freeman, Hadley. 2013a. "'she Has Challenged Not Just Her Own Public Image but Also the Wearisome Cliche of How a Woman Considered Sexy Talks About Her Body'." *The Guardian*, May 15.

Fulda, Kimberly G., and Kristine Lykens. 2006. "Ethical Issues in Predictive Genetic Testing: A Public Health Perspective." J Med Ethics 32 (3): 143–47. doi:10.1136/jme.2004.010272.

Garrett, Jinnie M., and Stephanie J. Bird. 2007. "Ethical Issues in Communicating Science." Sci Eng Ethics 6 (4): 435–42. doi:10.1007/s11948-000-0001-7.

Gerhards, Jürgen, and Mike S. Schäfer. 2007. "Hegemonie der Befürworter. Der mediale Diskurs über Humangenomforschung." Soz Welt 58 (4): 367-395.

Gibbs, Samuel. 2015. "DNA-Screening Test 23andMe Launches in UK After US Ban." *The Guardian*, October 2.

Hamilton, Jada G., Marci Lobel, and Anne Moyer. 2009. "Emotional Distress Following Genetic Testing for Hereditary Breast and Ovarian Cancer: A Meta-Analytic Review." Health Psychol 28 (4): 510–18. doi:10.1037/a0014778.

Harris, Marion, Ingrid Winship, and Merle Spriggs. 2005. "Controversies and Ethical Issues in Cancer-Genetics Clinics." Lancet Oncol 6 (5): 301–10. doi:10.1016/S1470-2045(05)70166-2.

Haupt, Laura. 2018. "Rescue via Genetic Findings." Hastings Cent Rep 48 (3): 2. doi:10.1002/hast.844.

Henderson, Lesley, and Jenny Kitzinger. 2007. "Orchestrating a Science 'Event': The Case of the Human Genome Project." New Genet Soc 26 (1): 65–83. doi:10.1080/14636770701218175.

Hirst, Olivia. 2015. "Why a Double Mastectomy Before the Age of 30 Can Be a Laughing Matter." *The Guardian*, August 13.

Houfek, Julia F., Brigette S. Soltis-Vaughan, Jan R. Atwood, Gwendolyn M. Reiser, and G. B. Schaefer. 2015. "Adults' Perceptions of Genetic Counseling and Genetic Testing." Appl Nurs Res 28 (1): 25–30. doi:10.1016/j.apnr.2014.03.001.

Huibers, Alex K., and Adriaan van 't Spijker. 1998. "The Autonomy Paradox: Predictive Genetic Testing and Autonomy: Three Essential Problems." Patient Educ Couns 35 (1): 53–62. doi:10.1016/S0738-3991(98)00083-4.

Jackson, Leigh, Lesley Goldsmith, and Heather Skirton. 2014. "Guidance for Patients Considering Direct-to-Consumer Genetic Testing and Health Professionals Involved in Their Care: Development of a Practical Decision Tool." Fam Pract 31 (3): 341–48. doi:10.1093/fampra/cmt087.

Jeffries, Stuart. 2014. "Family: How Much Do You Want to Know? If You Could Manipulate Your Child's Future - Would You? Stuart Jeffries Meets Geneticist Dr Sharon Moalem." *The Guardian*, April 5.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jolie Pitt, Angelina. 2015. "My Children Won't Have to Say 'Mom Died of Ovarian Cancer'." The *Daily Telegraph*, March 25.

Kalokairinou, Louiza, Heidi C. Howard, Santa Slokenberga, Eva Fisher, Magdalena Flatscher-Thöni, Mette Hartlev, Rachel E. van Hellemondt et al. 2018. "Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape." J Community Genet 9 (2): 117–32. doi:10.1007/s12687-017-0344-2.

Kamenova, Kalina, Amir Reshef, and Timothy A. Caulfield. 2014. "Angelina Jolie's Faulty Gene: Newspaper Coverage of a Celebrity's Preventive Bilateral Mastectomy in Canada, the United States, and the United Kingdom." Genet Med 16 (7): 522–28. doi:10.1038/gim.2013.199.

Knapton, Sarah. 2014. "Genetic Screening from Birth to Identify Disease Risks 'Within 40 Years'." The *Daily Telegraph*, January 10.

Lolkema, Martijn P., Christa G. Gadellaa-van Hooijdonk, Annelien L. Bredenoord, Peter Kapitein, Nancy Roach, Edwin Cuppen, Nine V. Knoers, and Emile E. Voest. 2013. "Ethical, Legal, and Counseling Challenges Surrounding the Return of Genetic Results in Oncology." J Clin Oncol 31 (15): 1842–48. doi:10.1200/JCO.2012.45.2789.

Lucassen, Anneke, and Michael Parker. 2010. "Confidentiality and Sharing Genetic Information with Relatives." Lancet 375 (9725): 1507–9. doi:10.1016/S0140-6736(10)60173-0.

Lynch, John, Ashley Parrott, Robert J. Hopkin, and Melanie Myers. 2011. "Media Coverage of Direct-to-Consumer Genetic Testing." J Genet Couns 20 (5): 486–94. doi:10.1007/s10897-011-9374-9.

Mand, Cara, Lynn Gillam, Martin B. Delatycki, and Rony E. Duncan. 2012. "Predictive Genetic Testing in Minors for Late-Onset Conditions: A Chronological and Analytical Review of the Ethical Arguments." J Med Ethics 38 (9): 519–24. doi:10.1136/medethics-2011-100055.

Marteau, Theresa M., and Robert T. Croyle. 1998. "Psychological Responses to Genetic Testing." BMJ 316 (7132): 693–96. doi:10.1136/bmj.316.7132.693.

Miah, Andy. 2005. "Genetics, Cyberspace and Bioethics: Why Not a Public Engagement with Ethics?" Public Underst Sci 14 (4): 409–21. doi:10.1177/0963662505056616.

Millum, Joseph. 2014. "The Foundation of the Child's Right to an Open Future." J Soc Philos 45 (4): 522–38. doi:10.1111/josp.12076.

Moorhead, Joanna. 2012. "A Cruel Inheritance: The Smiths Have a Family History of Huntington's Disease - an Incurable Neuro- Degenerative Disorder That Has Changed Life for Them All." *The Guardian*, September 22.

———. 2015. "Doctors Aim for Single Test to Detect Risk of Four Cancers in Women." *The Guardian*, June 2.

Mukherjee, Siddhartha. 2016. "'i Should Come with a Letter of Warning …." The *Daily Telegraph*, May 21.

Murray–West, Rosie. 2013. "Could New Cancer Tests Make You Uninsurable?" The *Daily Telegraph*, April 6.

Ormond, Kelly. 2008. "Medical Ethics for the Genome World: A Paper from the 2007 William Beaumont Hospital Symposium on Molecular Pathology." J Mol Diagn 10 (5): 377–82. doi:10.2353/jmoldx.2008.070162.

Pemberton, Max. 2013. "Breast Cancer Is Still a Risky Business." The *Daily Telegraph*, January 28.

———. 2014. "Should It Be Angelina's Job to Educate Us About Health?" The *Daily Telegraph*, September 22.

Pennacchini, M., and C. Pensieri. 2011. "Is Non-Directive Communication in Genetic Counseling Possible?" Clin Ter 162 (5): e141-144.

Pilkington, Ed, James Meikle, and Sarah Ridley. 2013b. "Experts Hail Jolie for Going Public on the Private Affair of Her Life-Saving Treatment: Actor Raises Awareness of Test for Defective Gene: Screening Detects Risk of Developing Breast Cancer." *The Guardian*, May 15.

Robinson, Ann. 2016. "DNA-Testing Kit 23andme: Patient-Powered Healthcare or Just Confusing?" *The Guardian*, January 12.

Rothstein, Mark A. 2018. "Reconsidering the Duty to Warn Genetically at-Risk Relatives." Genet Med 20 (3): 285-290. doi:10.1038/gim.2017.257.

Sample, Ian. 2011. "Patients' Genomes to Be Stored with Medical Records: US Pilot Study Hopes to Improve Drug Choices: Cost of Sequencing Now Similar to Single Gene Test." *The Guardian*, December 29.

Schwitzer, Gary, Ganapati Mudur, David Henry, Amanda Wilson, Merrill Goozner, Maria Simbra, Melissa Sweet, and Katherine A. Baverstock. 2005. "What Are the Roles and Responsibilities of the Media in Disseminating Health Information?" PLoS Med 2 (7): 576–82. doi:10.1371/journal.pmed.0020215.

U.S. Food and Drug Administration. 2017. "FDA Allows Marketing of First Direct-to-Consumer Tests That Provide Genetic Risk Information for Certain Conditions." Accessed January 23, 2020. https://www.fda.gov/news-events/press-announcements/fda-allows-marketing-first-direct-consumer-tests-provide-genetic-risk-information-certain-conditions.

———. 2018. "FDA Authorizes, with Special Controls, Direct-to-Consumer Test That Reports Three Mutations in the BRCA Breast Cancer Genes." Accessed January 23, 2020. https://www.fda.gov/news-events/press-announcements/fda-authorizes-special-controls-direct-consumer-test-reports-three-mutations-brca-breast-cancer.

Vadaparampil, Susan T., Cheryl A. Miree, Crystal Wilson, and Paul B. Jacobsen. 2007. "Psychosocial and Behavioral Impact of Genetic Counseling and Testing." Breast Disease 27:97–108. doi:10.3233/bd-2007-27106.

Vayena, Effy. 2015. "Direct-to-Consumer Genomics on the Scales of Autonomy." J Med Ethics 41 (4): 310–14. doi:10.1136/medethics-2014-102026.

Weishaar, Heide, Lori Dorfman, Nicholas Freudenberg, Benjamin Hawkins, Katherine Smith, Oliver Razum, and Shona Hilton. 2016. "Why Media Representations of Corporations Matter for Public Health Policy: A Scoping Review." BMC Public Health 16:899. doi:10.1186/s12889-016-3594-8.

Wertz, Dorothy C., and John C. Fletcher. 1991. "Privacy and Disclosure in Medical Genetics Examined in an Ethics of Care." Bioethics 5 (3): 212–32. doi:10.1111/j.1467-8519.1991.tb00161.x.

Wertz, Dorothy C., John C. Fletcher, Kare Berg, and WHO Human Genetics Programme. 2003. "Review of Ethical Issues in Medical Genetics: Report of Consultants to WHO." Accessed January 27, 2020. https://apps.who.int/iris/handle/10665/68512.

Wiggins, Sandi, Patti Whyte, Marlene Huggins, Shelin Adam, Jane Theilmann, Maurice Bloch, Samuel B. Sheps, Martin T. Schechter, and Michael R. Hayden. 1992. "The Psychological Consequences of Predictive Testing for Huntington's Disease. Canadian Collaborative Study of Predictive Testing." N Engl J Med 327 (20): 1401–5. doi:10.1056/NEJM199211123272001.

Yarborough, Mark, Joan A. Scott, and Linda K. Dixon. 1989. "The Role of Beneficence in Clinical Genetics: Non-Directive Counseling Reconsidered." Theor Med Bioeth 10 (2): 139–49. doi:10.1007/BF00539879.

Yim, Seon-Hee, and Yeun-Jun Chung. 2014. "Reflections on the US FDA's Warning on Direct-to-Consumer Genetic Testing." Genomics & informatics 12 (4): 151–55. doi:10.5808/GI.2014.12.4.151.

Zimmermann, Bettina M., Noah Aebi, Steffen Kolb, David Shaw, and Bernice S. Elger. 2019. "Content, Evaluations and Influences in Newspaper Coverage of Predictive Genetic Testing: A Comparative Media Content Analysis from the United Kingdom and Switzerland." Public Underst Sci 28 (3): 256-274. doi:10.1177/0963662518816014.

Chapter 9: To what extent should the public be informed about genetic testing?

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9.1 Abstract

Genetic testing is becoming increasingly relevant for the public, but medical, social and ethical issues are often complex and sometimes difficult to understand for laypeople. Informing the public empowers individuals to assess the usefulness of direct-to-consumer genetic tests realistically and enables them to participate in public debates about genetics. However, too much information can lead to information overload, misconceptions and stress. Thus, we argue that, while informing the public about genetic testing is important, public health authorities need to prioritize the most important messages and provide reliable and up-to-date information that is accessible to the public. Mass media are not appropriate as predominant information sources because of their conflicts of interest and inaccuracy in reporting genetics.

9.2 The public relevance of genetic testing

Genetic testing is becoming more relevant to the public for three reasons. First, with recent technological advances, such as next-generation sequencing (Bahassi and Stambrook 2014) and the genome-editing tool Crispr-Cas9 (Zhang, Wen, and Guo 2014), genetic research is making important progress in collecting clinically relevant evidence that improves and broadens the possibilities of clinically meaningful genetic testing. Second, Angelina Jolie's statements about having preventive surgery twice because of an increased genetic risk of developing breast and ovarian cancer (Jolie, May 13, 2013; Jolie Pitt, March 25, 2015) has increased public awareness and demonstrates the interest of laypeople in such tests (Borzekowski et al. 2014). Third, direct-to-consumer genetic testing companies advertise a variety of genetic tests directly to the public, but they are usually not scientifically reliable or clinically useful (Covolo et al. 2015).

Understanding genetic testing is complex because of the many different applications. Within medicine, diagnosing diseases, predicting future disease risks and determining individualized drug regimens are common applications of genetic testing (Kamps et al. 2017). Prenatal genetic testing is another important area of application (Vermeesch, Voet, and Devriendt 2016), and each domain has different difficulties, significance, and ethical issues that need to be considered. Prenatal and predictive genetic examinations, for instance, often only give results in terms of risks, with no definite answers (Evans, Skrzynia, and Burke 2001). Thus, anticipating the consequences of a test result is especially complex. Genetic testing also affects family members, because detected genetic variants are heritable if they concern the germline. This leads to potential social and ethical issues, such as how to disseminate information to family members while respecting their right to know, but also their right not to know (Juth 2014) genetic information about themselves or family members.

Moreover, genetic testing can be analyzed on different levels: targeted analyses look at specific regions in the genome whose functions are well understood; in contrast, genome-wide analyses look at genetic variations throughout the whole genome, which increases the risk of incidental findings[1] and variants of unknown significance[2] (Machini et al. 2014). Direct-to-consumer genetic testing companies often only sequence a small portion of the genome and use a standardized algorithm for analysis, but such analyses can be misleading because they are not individualized and do not take all relevant scientific knowledge into account. From a medical perspective, they are useless more often than not (Covolo et al. 2015).

9.3 Benefits of an informed public

Enhancing the public's understanding of and engagement with genetics and its ethical implications (Miah 2005; Zimmermann, Elger, and Shaw 2019) enhances individual autonomy since it offers members of the public the opportunity to make informed choices regarding genetic testing. It benefits those who are eligible for special preventive measures and potentially prevents disease if pathogenic genetic variants are detected early (at least in some cases, like hereditary breast/ovarian cancer or Lynch syndrome). It also benefits those interested in direct-to-consumer genetic testing because it enables them to appraise the limits of these tests. Moreover, only an informed public can contribute fruitfully to debates relevant to policy-making issues, such as the allocation of financial resources to preventive genetic testing or priority setting in the clinical context. Therefore, we argue that it is an ethical obligation for public health authorities to inform the public of the complexities of genetic testing, including issues associated with clinical usefulness, and ethical and social issues.

However, there is a moral dilemma regarding the nature and amount of information that it is optimal for the public to receive: on the one hand, information increases autonomy and potentially prevents harm, but on the other, too much information can be harmful. We argue that a reflective equilibrium needs to be established to find the correct balance between respecting and enhancing people's autonomy and preventing them from harm when informing the public about genetics.

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1 Incidental findings, also often called secondary findings, refer to the detection of genetic variants that are linked to different diseases from those that were originally anticipated. Ethical problems that arise from such findings include when to report them and how to obtain informed consent (see (Mackley et al. 2017) for a systematic review on this issue).

2 Variants of uncertain significance (VUS) are detected genetic variants whose influence on disease risk or progression is unknown. In such cases, the test result cannot give the tested individual or relatives a definitive "positive" or "negative" answer (see (Greenblatt 2015)for more information).

9.4 How to inform the public

Considering the complexity of genetic testing and the significant variation between members of the public in terms of education and prior knowledge, perceptions and attitudes, the optimal information channel would be individual and personal genetic counselling. However, it will be difficult to provide enough personnel and financial resources to provide this if a large proportion of the population is interested in genetic counselling. Information campaigns, mass media coverage, and social media initiatives are more cost-effective options to provide information to the public. However, these are impersonal and allow only limited interaction between the sender and the receiver of information. Moreover, mass media follow their own rules. They cover topics that interest their readers and tend to sensationalize and exaggerate the meaning or implication of genetics (Marcon, Bieber, and Caulfield 2018; Zimmermann et al. 2019). They are also prone to financial conflict of interests and it is unclear how stakeholders are influencing media coverage "behind the curtain" (e.g., Bajo, Bigelli, and Raimondo 2019; Thomas et al. 2018). Public health authorities are just one stakeholder competing for mass media coverage.

We thus need more independent information campaigns and platforms that are not subject to such substantial conflict of interests. Public health authorities should enable such initiatives through funding and make sure that conflicts of interests are limited as much as possible.

9.5 The more information, the better?

While informing the public is an ethical duty of health authorities, an excess of such information might cause harm, such as stress, anxiety, and unhealthy behaviour. First, individuals might have information overload, meaning the inability to process the amount of information and/or access relevant information. Information overload is associated with information avoidance and fatalistic attitudes towards disease prevention, leading to stress and unhealthy behaviour (Jensen et al. 2014). Second, because of the impossibility of directly clarifying misunderstandings and misperceptions in public communication, it is challenging to provide information that is perceived in the intended way. The framing of information largely influences how it is perceived and interpreted by its recipients (Chong and Druckman 2007). By exposing the public to unfiltered information about genetic testing, we risk leaving the interpretation and framing to the recipient, which might lead to misconceptions. An additional complication is that the aforementioned fact that "the public" as a target audience is a fuzzy concept and includes individuals with a large variety of characteristics regarding age, culture, prior knowledge, and attitudes.

In summary, providing detailed and unfiltered information to the public bears the risk of

information overload and misperceptions, which might leave people insecure and unable to make informed decisions. Thus, we need to establish the extent to which the public should be educated about genetic testing and develop a concept of communication that puts this into practice.

9.6 Set priorities and avoid conflicts of interest

At the moment, there seems to be too much information on the one hand and too little on the other. People dig through the vast amount of information available on the World Wide Web and have difficulty finding what is relevant for them (Ramírez and Arellano Carmona 2018; Klerings, Weinhandl, and Thaler 2015). Any effort to educate the public through mass media, therefore, need to fulfil the following minimal requirements: (1) it needs to be salient and easily accessible for everybody; (2) it needs to be evidence-based and correct but easy to understand; (3) it needs to address existing misperceptions and exaggerated claims; (4) it needs to be condensed to the most relevant information. This is generalizable to any health-related issue, but regarding genetic testing, we think the most relevant point is to raise awareness of its complexity without explaining it in details: making people aware that there are different applications of genetic testing and that they have different psychological and medical consequences for both the tested individual and family members. Moreover, we think it is important to make people aware of the lack of reliability and quality control of direct-to-consumer genetic testing.

We think it is important to reach a consensus within the public health community on what priorities health communication should focus on in order to avoid information overload while also providing relevant and high-quality information. We also consider it useful to develop a publicly funded information platform, which focuses not only on medical and scientific but also on ethical and social issues relating to genetic testing and is tailored to enable members of the public to make informed decisions on this topic, especially regarding direct-to-consumer genetic testing. This information platform should be informed by experts but led by an independent body to minimize conflicts of interest. If our recommendations are followed, the public will receive balanced and helpful information about genetic testing.

9.7 References

Bahassi, El M., and Peter J. Stambrook. 2014. "Next-Generation Sequencing Technologies: Breaking the Sound Barrier of Human Genetics." Mutagenesis 29 (5): 303–10. doi:10.1093/mutage/geu031.

Bajo, Emanuele, Marco Bigelli, and Carlo Raimondo. 2019. "Ownership Ties, Conflict of Interest, and the Tone of News." Eur Financ Manag, 1–19. doi:10.1111/eufm.12237.

Borzekowski, Dina L. G., Yue Guan, Katherine Smith, Lori H. Erby, and Debra L. Roter. 2014. "The Angelina Effect: Immediate Reach, Grasp, and Impact of Going Public." Genet Med 16 (7): 516–21. doi:10.1038/gim.2013.181.

Chong, Dennis, and James N. Druckman. 2007. "Framing Theory." Annu Rev Polit Sci 10 (1): 103–26. doi:10.1146/annurev.polisci.10.072805.103054.

Covolo, Loredana, Sara Rubinelli, Elisabetta Ceretti, and Umberto Gelatti. 2015. "Internet-Based Direct-to-Consumer Genetic Testing: A Systematic Review." J Med Internet Res 17 (12): e279. doi:10.2196/jmir.4378.

Evans, James P., Cécile Skrzynia, and Wylie Burke. 2001. "The Complexities of Predictive Genetic Testing." BMJ 322 (7293): 1052–56. doi:10.1136/bmj.322.7293.1052.

Greenblatt, Marc S. 2015. "Sequence Variants of Uncertain Significance: What to Do When Genetic Test Results Are Not Definitive." Surg Oncol Clin N Am 24 (4): 833–46. doi:10.1016/j.soc.2015.06.009.

Jensen, Jakob D., Nick Carcioppolo, Andy J. King, Courtney L. Scherr, Christina L. Jones, and Jeff Niederdieppe. 2014. "The Cancer Information Overload (CIO) Scale: Establishing Predictive and Discriminant Validity." Patient Educ Couns 94 (1): 90–96. doi:10.1016/j.pec.2013.09.016.

Jolie, Angelina. 2013. "My Medical Choice." The New York Times, May 13.

Jolie Pitt, Angelina. 2015. "My Children Won't Have to Say 'Mom Died of Ovarian Cancer.'" The Daily Telegraph, March 25.

Juth, Niklas. 2014. "The Right Not to Know and the Duty to Tell: The Case of Relatives." J Law Med Ethics 42 (1): 38–52. doi:10.1111/jlme.12117.

Kamps, Rick, Rita D. Brandão, Bianca J. van den Bosch, Aimee D. C. Paulussen, Sofia Xanthoulea, Marinus J. Blok, and Andrea Romano. 2017. "Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification." Int J Mol Sci 18 (2): 308. doi:10.3390/ijms18020308.

Klerings, Irma, Alexandra S. Weinhandl, and Kylie J. Thaler. 2015. "Information Overload in Healthcare: Too Much of a Good Thing?" Z Evid Fortbild Qual Gesundhwes 109 (4-5): 285–90. doi:10.1016/j.zefq.2015.06.005.

Machini, Kalotina, Jessica Douglas, Alicia Braxton, Judith Tsipis, and Kate Kramer. 2014. "Genetic Counselors' Views and Experiences with the Clinical Integration of Genome Sequencing." J Genet Couns 23 (4): 496–505. doi:10.1007/s10897-014-9709-4.

Mackley, Michael P., Benjamin Fletcher, Michael Parker, Hugh Watkins, and Elizabeth Ormondroyd. 2017. "Stakeholder Views on Secondary Findings in Whole-Genome and Whole-Exome Sequencing: A Systematic Review of Quantitative and Qualitative Studies." Genet Med 19 (3): 283–93. doi:10.1038/gim.2016.109.

Marcon, Alessandro, Mark Bieber, and Timothy A. Caulfield. 2018. "Representing a "Revolution": How the Popular Press Has Portrayed Personalized Medicine." Genet Med 20 (9): 950–56. doi:10.1038/gim.2017.217.

Miah, Andy. 2005. "Genetics, Cyberspace and Bioethics: Why Not a Public Engagement with Ethics?" Public Underst Sci 14 (4): 409–21. doi:10.1177/0963662505056616.

Ramírez, A. S., and Kimberly Arellano Carmona. 2018. "Beyond Fatalism: Information Overload as a Mechanism to Understand Health Disparities." Soc Sci Med 219:11–18. doi:10.1016/j.socscimed.2018.10.006.

Thomas, Jackson, Gregory M. Peterson, Erin Walker, Julia K. Christenson, Melissa Cowley, Sam Kosari, Kavya E. Baby, and Mark Naunton. 2018. "Fake News: Medicines Misinformation by the Media." Clin Pharmacol Ther 104 (6): 1059–61. doi:10.1002/cpt.1199.

Vermeesch, Joris R., Thierry Voet, and Koenraad Devriendt. 2016. "Prenatal and Pre-Implantation Genetic Diagnosis." Nat Rev Genet 17 (10): 643–56. doi:10.1038/nrg.2016.97.

Zhang, Feng, Yan Wen, and Xiong Guo. 2014. "CRISPR/Cas9 for Genome Editing: Progress, Implications and Challenges." Hum Mol Genet 23 (R1): R40-6. doi:10.1093/hmg/ddu125.

Zimmermann, Bettina M., Noah Aebi, Steffen Kolb, David Shaw, and Bernice S. Elger. 2019. "Content, Evaluations and Influences in Newspaper Coverage of Predictive Genetic Testing: A Comparative Media Content Analysis from the United Kingdom and Switzerland." Public Underst Sci 28 (3): 256-274. doi:10.1177/0963662518816014.

Zimmermann, Bettina M., Bernice S. Elger, and David Shaw. 2019. "Media Coverage of Ethical Issues in Predictive Genetic Testing: A Qualitative Analysis." AJOB Empirical Bioethics 10 (4): 250–64. doi: 10.1080/23294515.2019.1670275.

Chapter 10: Discussion

This dissertation aimed to examine what information about genetic testing should be given to individuals and the public to foster and facilitate informed genetic testing decision-making, and how such information is best transmitted. These objectives were empirically investigated with an interview study and a newspaper content analysis on predictive genetic testing. By doing so, it also aimed to explore how newspaper content analyses can inform bioethics research.

Information needs in the genetic testing decision-making process differ importantly between at-risk individuals (chapter 4). Prior knowledge is an advantage for understanding concepts necessary for genetic testing decision-making, for identifying and finding missing information. However, besides factual knowledge, personal life philosophy, decision-making style and social relations influence genetic testing decision-making importantly (chapter 3).

Genetic counselling is the most important source of information. Adequate alternatives are difficult to find for at-risk individuals in the German-speaking part of Switzerland, especially for those with little prior knowledge. At-risk individuals did not consider mass media a reliable information source; they either did not recall reading about the topic in mass media or described coverage as tendentious and populistic (chapter 4). Indeed, we found little coverage about predictive genetic testing in Switzerland 2011-2016 as compared to the UK (chapter 7). Triggers for newspaper coverage were the Angelina Jolie case and political discussions, for instance regarding preimplantation genetic diagnoses or direct-to-consumer genetic testing in Switzerland (chapters 6 and 7). Swiss coverage included a variety of stakeholders and evaluations. In contrast, newspaper coverage in the UK was positively biased: For instance, UK newspapers described predictive genetic testing for medically actionable variants as purely beneficial (chapter 8). This bias is problematic, as the interview study revealed that there are good reasons to refuse genetic testing for actionable variants (chapter 3). The comparative newspaper content analysis confirmed expected differences in media portrayal of genetic testing due to different cultures and legislations. Ethical issues related to predictive genetic testing were underrepresented in press coverage of both countries (chapter 7), even though they are of high societal and individual relevance (chapter 8).

The following sections discuss the results with particular emphasis on practical recommendations. Chapter 10.1 elaborates on the extent of information needed for informed decision-making regarding predictive genetic testing and the opportunities and limits of mass media communication in that context. In chapter 10.2, alternatives to genetic counselling for obtaining information regarding genetic testing are discussed. Chapter 10.3 focuses on the potential role of newspaper content analyses in bioethics. Limitations and implications for further research are presented in chapter 11.4. Chapter 10.5 covers the conclusion of this dissertation.

10.1 The public level: public education and public debates

10.1.1 Informed decision-making and the extent of public education

As introduced in chapter 1.3.1, an autonomous decision should be made intentionally, with understanding and with few or no controlling influences [1] (Faden and Beauchamp 1986). Based on the results presented in chapter 3, I argue that self-reflection should be an additional requirement for informed genetic testing decision-making. Self-reflection is commonly connected to philosophical discussions about autonomous persons (Christman 2018). Also White, L. (2018) emphasises the importance of self-reflection concerning medical decision-making. Faden and Beauchamp, however, criticise that this makes the requirements for autonomous actions too complicated (Faden and Beauchamp 1986, 264). Still, decision-making in predictive genetic testing differs from other medical decision-making, as the individual decides whether to take the test or not without any direct recommendation from the genetic counsellor.[2] Moreover, in the case of predictive genetic testing, the decision of whether to take a test usually is not as urgent as in other medical settings. Therefore, decision-makers for predictive genetic testing should be encouraged to reflect on how a disease risk would influence their life and how predictive genetic testing is in line with their life philosophy.

To fulfil the remaining two conditions for autonomous decision-making, intentionality and understanding, people need information. Prior knowledge about genetics supports people's information-seeking behaviour (chapter 3). While some people are highly educated and might even have formal medical or scientific training, the majority of the population has no such qualifications and requires more information. Public education can contribute to the improvement of prior knowledge among the general population; therefore many authors have noted the importance of informing the public about genetics (e.g., Condit 2010; Marzuillo et al. 2013; McBride et al. 2010; de Vries, H. et al. 2005).

1 Because family plays a vital role in genetic testing decision-making, social influences are unavoidable in that context and acceptable as long as they do not lead to coercion or undue pressure. Chapter 5 discusses this issue in detail.

2 This nondirectiveness was an important principle in genetic counselling (Aston 1998). However, its appropriateness and usefulness has been questioned (Weil, J. et al. 2006; Pennacchini and Pensieri 2011). Still, Swiss law and the recent German genetic counselling guidelines adhere to this principle (Swiss Federal Council 2004; Deutsche Gesellschaft für Humangenetik e.V. and Berufsverband Deutscher Humangenetiker e.V. 2018).

By contrast, more information does not necessarily make decisions more informed (Lynöe and Hoeyer 2005, see also chapter 1.3.1). Humans have limited capacity to process information (Khaleel et al. 2020; Chae, Lee, C.-j., and Jensen 2016). Moreover, messages are interpreted differently depending on how they are framed (Bunnik, Schermer, and Janssens 2014) and might not be remembered adequately (Kahneman 1994). That is especially true for inherently complex topics such as predictive genetic testing with its medical, scientific and psychosocial dimensions (Djurdjinovic and Peters, J. 2017). Thus, more information is *not* always better (see also Beauchamp and Childress 2013; Manson and O'Neill 2007).

Accordingly, a prioritisation of information is necessary, which is valid for both private and public information channels. In private information channels, this prioritisation can be individualised, and the receiver can influence it by asking questions. In public information channels, prioritisation must be more standardised. Before giving practical recommendations of this prioritisation in chapter 10.1.3, the next section discusses the role of mass media in informing the public about predictive genetic testing.

10.1.2 Mass media as information channels

Mass media, here represented by broadsheet newspapers,[3] function as an information channel and as an arena for public debates (Peters, H. 1994; Kamber and Imhof 2011). The first function of mass media as information channels includes (1) *informing* people about genetic testing applications and their implications for society (Weingart and Schulz 2014), and (2) *educating* people to guide meaningful decision-making and prevent harmful behaviour. While information leaves the interpretation to the recipients, education through mass media takes directives and tells people what they should or should not do[4] (Schwitzer et al. 2005; Peters, H. 1994). The arena function of mass media is crucial for democratic societies (Luhmann 2009, 118). They enable, for instance, the scientific and the public sphere to communicate and discuss with each other (Franzen, Weingart, and Rödder 2012). That includes other relevant actors as well, for example, stakeholders from politics and economics. Mass media have an additional function to inform members of the public so that they can follow and participate in these public debates.

In the here-presented studies, broadsheet newspapers serve this arena function well by giving these stakeholders a stage to present their interests and views regarding predictive genetic testing

3 See chapter 2.2.1 for a justification of this representation.

4 For example, Swiss newspaper coverage about predictive genetic testing emphasised the dangers of direct-to-consumer genetic test and the narrative discouraged people from buying it, whereas UK newspaper coverage was more balanced in this regard (see also chapter 3).

(chapter 7). However, they did not sufficiently fulfil their information function. While our newspaper content analyses showed more distinguished media portrayal than it was the case in previous studies (Gerhards and Schäfer 2006), they represented ethical issues in a simplistic manner (chapter 8). This is not a satisfactory information function to foster informed decision-making for predictive genetic testing.

Instead of blaming newspapers that they have not served their information function, I consider this an inherent feature of mass media coverage. Mass media are not neutral. They are a societal system themselves and have their own rules, interests and biases (Luhmann 2009, 128–29). As such, they are themselves a player in the public discourse arena. Thus, media coverage is neither objective nor independent (Weingart and Schulz 2014).[5] Instead, mass media inherently tend to report biased information on predictive genetic testing. For instance, they overemphasise risks, create exaggerated expectations or neglect an issue entirely. Therefore, instead of criticising media for their often tendentious reporting about genetics (Caulfield and Condit 2012), it should be acknowledged that mass media might not be the most objective and neutral information source.[6] Carrying this idea further, the next section provides practical recommendations on how mass media are most efficiently used to foster informed decision-making.

10.1.3 Prioritisation of information dissemination through mass media

As indicated, the results suggest that mass media are quite successfully serving the arena function by letting different stakeholder have their say (chapter 7). However, they might be biased information channels and should not be the primary information source for individual decision-making regarding predictive genetic testing.[7] Therefore, I suggest to prioritise the arena function and prioritise mass media communication that fosters public debates about societal issues related to predictive genetic testing. These recommendations are for science journalists and researchers involved in science communication, but they should by no means lead to censorship or mass media control. The independence of mass media is crucial for democratic societies.

5 This is also supported by the theory of news values (Badenschier and Wormer 2012), the gatekeeping theory (Shoemaker 2002) and the time development analyses in chapters 6 and 7. At the same time, the effects of media coverage on individual recipients are complex and manifold (Früh and Schönbach 2005; Scheufele and Tewksbury 2007), which creates space for misconceptions and prejudice (see also chapter 1.3.3).

6 Alternative public information sources are discussed in chapter 10.1.4.

7 While, in a clinical setting, genetic counselling usually is the main information source, mass media could indeed be one of the most important information source of direct-to-consumer genetic testing users.

While science communicators can try and place these messages, mass media still decide themselves what they publish.

Issues relevant to society and policy should have top priority as such information prepares individuals to follow and participate in public debates and political decision-making (Figure 10-1). It fosters public engagement with predictive genetic testing regarding its societal relevance. Political decision making is crucial for democracies, notably Switzerland.[8] Current examples include genetic discrimination, equality of access, implications of sharing genetic data, genetic determinism and the potential future influence of the increasing use of predictive genetic testing on healthcare systems and societies.

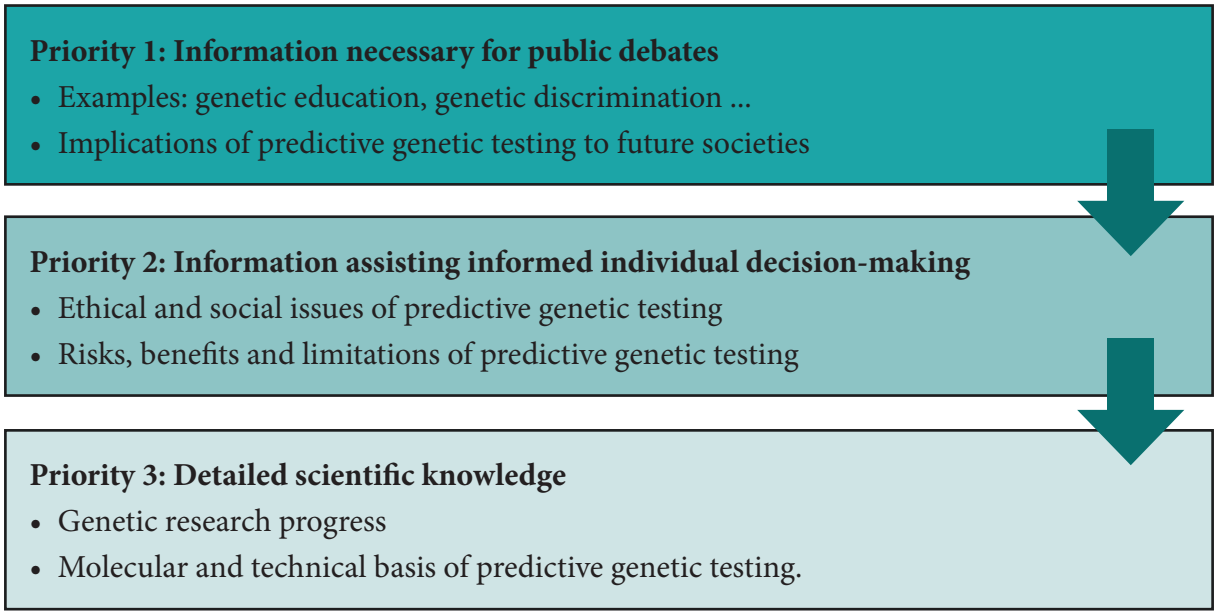

Figure 10-1: Recommendations for priorities of science communication regarding predictive genetic testing.

The second priority is information relevant to individual decision-making (see also Condit 2010). This includes ethical and social issues as well as information about risks, benefits and limitations of predictive genetic testing. A survey study identified a knowledge gap regarding such issues (Haga et al. 2013). At the same time, science communicators should strive to counteract existing tendentious media portrayal to provide more balanced public information. For instance, UK newspapers framed predictive genetic testing for actionable variants as purely beneficial, but some might want to decide against predictive genetic testing, even if actionable, because of their life philosophy (chapter 3.4). Media should cover this aspect as well.

8 Switzerland is a direct democracy, which means people can vote directly on policy issues. The public thus has to be informed in detail on factual and technical aspects of voting issues.

While factual and practical information about predictive genetic testing is essential and relevant, it is only the third priority. For instance, some authors suggested focusing media coverage on types and symptoms of hereditary cancers, what to do if hereditary cancer was suspected and gene-environment interactions (de Vries, H. et al. 2005; Smerecnik et al. 2008; Corbellini 2004). I suggest using alternative public information channels for such information.

10.1.4 Information platforms and school education

To make informed decisions about predictive genetic testing, people need holistic and decision-neutral information. Professional and independent information platforms can provide interested members of the public with the opportunity to find relevant and high-quality information adapted to the legal and cultural contexts. Such information platforms are more efficient than mass media in providing balanced and comprehensive information regarding the medical and scientific facts, psychosocial, legal and ethical issues and different applications of predictive genetic testing. They provide opportunities to foster exchange with interested representatives of the public, to do research and to include different formats of information transmission, such as text, video, and social media. They can also serve as platforms for e-learning tools and electronic decision aids.

The main challenges regarding such platforms are financing and responsibilities. While, in my opinion, public health authorities should finance these platforms, they should be set up and maintained by an interdisciplinary expert committee. It should include geneticists, ethicists and communication specialists and should reflect on potential conflicts of interest when defining the content of the platform. Future efforts should first and foremost focus on implementing such platforms.

No such platform is available so far in the German language.[9] The platform SantéPerSo (Fondation Leenaards and Médecine et Hygiène 2019) provides a comparable platform for personalised medicine, targeting the French-speaking part of Switzerland. It provides educative articles, presents ethical and societal issues related to personalised medicine, illustrates medical applications and introduces current research topics. Moreover, it serves as an exchange platform between

9 Existing information platforms in the German-speaking part of Switzerland do not fulfill the criteria of independency, professionalism, holism and neutrality. The foundation GENSUISSE provides information but is financed from pharmaceutical industry. The association biorespect, by contrast, runs an information platform distinctly against genetic testing (biorespect 2020). Swiss university hospitals and the Swiss cancer league provide some information, but this information is difficult to find for non-experts and usually focussing on genetic used in standard clinical practice in Switzerland today.

experts and the public, as it includes opinion pieces and a Q&A section. Numerous regional experts from various disciplines write articles. A private foundation finances the platform.

Another essential aspect is school education. Basic knowledge about genetics is part of primary school education in many cantons in Switzerland (e.g., Amt für Volksschulen Kanton Basel-Landschaft 2019; Deutschschweizer Erziehungsdirektoren-Konferenz 2016; Bildungsdirektion des Kantons Zürich 2017). Still, applied genetic knowledge with societal impacts, such as direct-to-consumer and predictive genetic testing, should be included as well – even though it may prove politically challenging to implement such new contents on a national level in a federalist system. This issue, however, goes beyond the scope of this dissertation.

In summary, public education is essential for informed decision-making if individual information channels are not available. However, information should be prioritised because too much information can lead to misconceptions and information overload. While information platforms should transmit holistic knowledge, science communicators should focus their efforts on societal and ethical issues of predictive genetic testing, including risks, benefits and limitations. The next chapter will now move to an individual level and elaborate on genetic counselling in the context of scarce resources.

10.2 The individual level: personal counselling in the context of scarce resources

For many individuals considering predictive genetic testing in a clinical setting in Switzerland, genetic counselling is the most important source of information (chapter 4.4). Genetic counselling is standard clinical practice for predictive genetic testing and even required by Swiss law (Swiss Federal Council 2004). Because of the increasing demand, Switzerland needs more personnel trained to provide genetic counselling,[10] but the following section outlines reasons for also considering other alternatives. Based on the findings of this dissertation, I then suggest how information could be provided based on individual information needs.

10.2.1 Alternatives to personal genetic counselling

The scientific community has been discussing and testing alternatives to genetic counselling: Genetic counselling in groups (Benusiglio et al. 2017), counselling via video conferencing, with the help of e-education and online decision aids (Reumkens et al. 2019; Adam et al. 2018; Otten

10 See chapter 1.4.1.

et al. 2016b; Wolfe et al. 2015) or via social media (Moore, Matthews, and Cohen, L. 2018). Artificial Intelligence can further support genetic counselling, for instance, using chatbots or advanced risk assessment tools (Gordon, Babu, and Laney 2018). In most approaches, one of the alternatives replaced only the initial information part, followed up by shortened personal counselling, where patients could ask remaining questions. This personal part was either standard part of the information procedure or upon the individuals' request, depending on the genetic testing application.

Users and healthcare professionals positively evaluated these alternatives: Knowledge scores and psychological outcomes were comparable to traditional genetic counselling (Buchanan, Rahm, and Williams 2016). However, no intervention study using such an alternative has been performed in Switzerland. It is thus necessary to test such alternatives in the Swiss context and to investigate the attitudes of medical professionals and patients.

This is important because, first, alternatives might be more flexibly accessible, allowing individuals to find information at any time during the decision-making process. At the time of genetic counselling, the decision-making process is often already ongoing (chapters 3 and 4), but acquiring knowledge before the actual decision-making process can facilitate informed decision-making. It enables individuals to contextualise information efficiently and to identify relevant knowledge gaps during the genetic counselling session.

Second, direct-to-consumer genetic testing usually does not include genetic counselling. It falls within the individual's responsibility to acquire the knowledge necessary for informed decision-making. Even if the current draft of the revised Swiss Human Genetic Testing Act (HGTA) proposes a ban for medical applications of direct-to-consumer genetic testing (Swiss Federal Council 2017), people can still purchase these tests online. A legal ban might result in neglecting public debates on the subject. To independently inform costumers of direct-to-consumer genetic testing, public information needs to be available.

A third reason to think about alternatives to personal genetic counselling is the increasing burden for healthcare systems. While preventing diseases might reduce healthcare costs in some cases, installing unlimited preventive screening would, in the medium term, bring the finances of healthcare systems to their limits (Severin et al. 2015). Considering alternatives to genetic counselling is one option for proactively rationalising available resources. The recommendation presented in the next section contributes to this discussion.

10.2.2 Stratifying people according to their information needs

I suggest offering people different pathways of decision support according to their individual information needs. An electronic stratification algorithm suggests these individual information pathways (Figure 10-2), which is implemented in the form of an online recommendation system with a clearly defined decision tree. The stratification aims to maximise informed decision-making by accounting for individual information needs with a constraint condition regarding the use of limited healthcare resources. It addresses the future information bottleneck regarding genetic counselling.

Implementation of the stratification algorithm

People differ importantly in their decision-making process: they have different starting points, different information needs and different mindsets regarding predictive genetic testing decision-making (chapters 3-5; Case 2007; Etchegary et al. 2009; Hamilton and Bowers 2007). The algorithm will use these variables to determine the individual's information pathway. Relevant variables include, for instance, genetic literacy, decision-making style, educational background, present psychological state, the family situation regarding genetic testing, life philosophy, and personal preferences. Another critical variable is the testing condition. Predictive genetic testing for to-date medically nonactionable diseases (e.g. Huntington's disease) or untargeted genome-wide analyses (e.g. for early-onset rare diseases) might lead to a higher information need. These variables could, for instance, be collected through electronic questionnaires to be filled out by the patient and the referring medical doctor. If infrastructure allows it, it could be potentially useful to include data from electronic health records to save resources to fill out these online questionnaires. Based on these input variables, the stratification algorithm then suggests an individualised information pathway.

If the algorithm identifies a high information need, it refers to the individual directly to personal genetic counselling (pathway 1). The genetic counsellor will carefully assess open questions and take as much time as necessary to explain predictive genetic testing in detail, according to the individual's needs and knowledge gaps. The genetic counsellor and the at-risk individual will then together determine the next steps of the decision-making process.

A medium need (pathway 2) indicates that the individual requires targeted education. The individuals will first finalise an e-education task tailored towards these particular information needs. Knowledge gaps will be addressed using their preferred information channel: online counselling, group counselling or personal counselling. By identifying the fields of interest, the

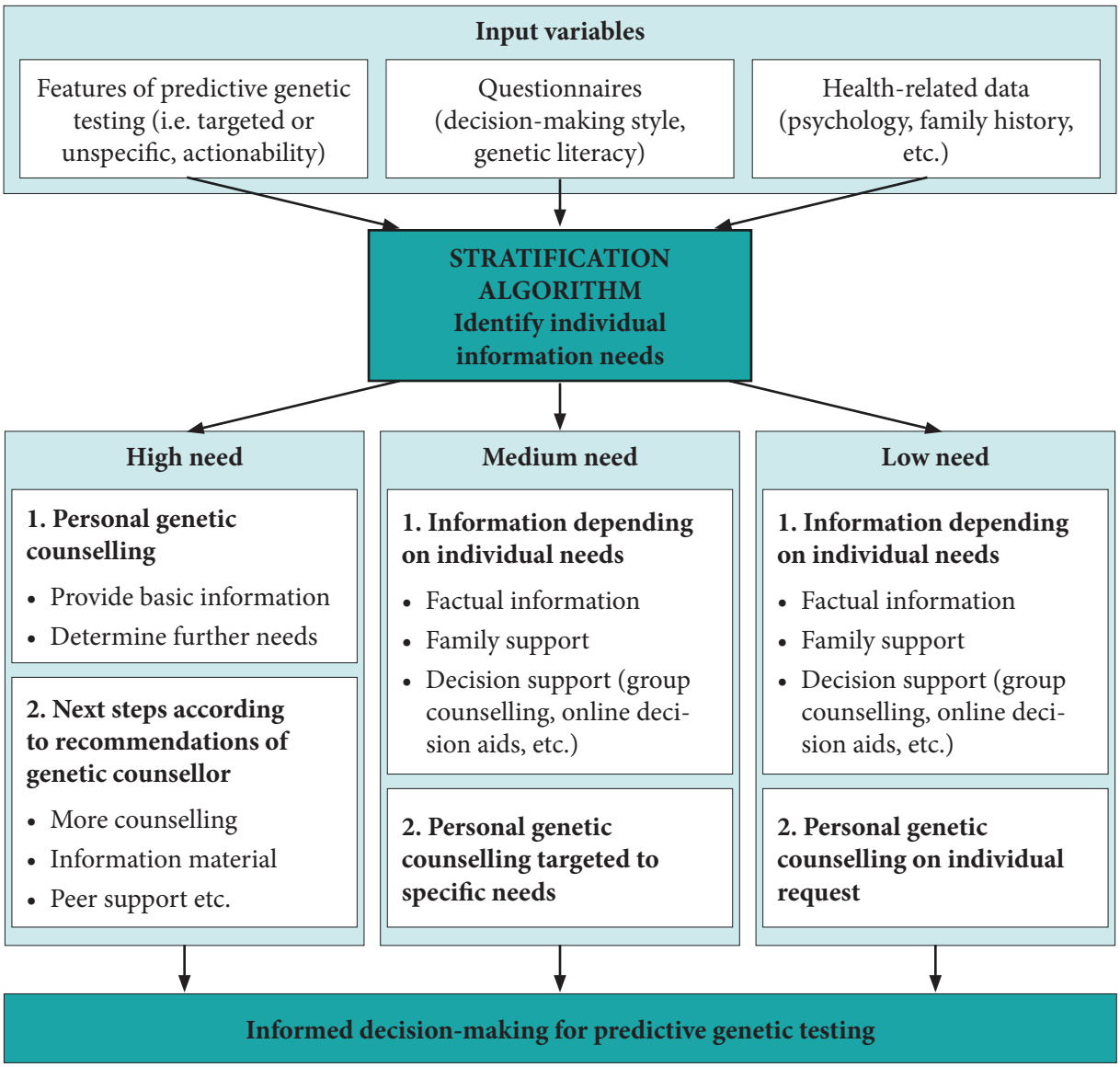

Figure 10-2: Recommendations for individual predictive genetic testing decision support to maximise informed decision-making.
Individualise information pathways according to people's needs.

algorithm optimises these interventions to the at-risk individual's information requirements. However, individuals always have the option to contact a specialist if they are unsatisfied with the process or the information obtained.

Pathway 3, finally, is for individuals with low information needs. They already have high genetic literacy, are conscious about the medical, social and ethical consequences of a genetic test and request a straightforward genetic analysis (for instance, a genetic test for a pathogenic BRCA variant already diagnosed in a family member). The recommendation system will refer them to an e-education platform, where they can find additional information and can contact a genetic counsellor for remaining questions.

Challenges

Many challenges must be overcome before such a stratification system is useful for practice. First, extensive research is necessary: the existing empirical evidence needs to be confirmed and complemented. Quantitative research tailored to the context of pre-test genetic counselling will identify an exhaustive list of variables relevant for stratification. The stratification algorithm will also need to be validated to make sure all individuals get the level of support necessary for informed decision-making. Since algorithms are based on statistics, a small error margin is accepted, but this is accounted for by making additional personal genetic counselling available upon request.

Moreover, the outcome variable "informed decision-making" must be operationalised for this specific context. In previous studies, informed decision-making was evaluated based on knowledge, attitudes and behaviour (Marteau, Dormandy, and Michie 2001). Still, that score should be reevaluated for the context of predictive genetic testing, as the results from chapter 3 indicate that individuals might make informed decisions differing from their attitudes towards genetic testing.

A second challenge of this approach is its social acceptance. While non-personal alternatives to genetic counselling have been introduced long ago in the United States (Cohen, S. et al. 2013), they are implemented to a lesser degree in Europe, with substantial regional differences. The main barriers to using video-conferencing mentioned by European healthcare professionals are lack of money or reimbursement and lack of interest from the employees (Otten et al. 2016a). Thus, the main obstacles are systemic or personal ones. It will thus be critical to convince healthcare professionals to be part of the research in this area. Additionally, people must be transparently informed about this stratification approach. Otherwise, some individuals might feel unjustly treated if they find out they receive less personal support than others.

Third, more practical challenges concern the financing and management of the system. Optimally, the whole system, including questionnaires, are online-based but run by an official stakeholder. The detailed implementation is highly depending on existing healthcare structures and the country's bureaucratic organisation. It might be useful and synergetic to combine it with the implementation of the information platform, as suggested in chapter 10.1.4.

Ethical objections

The main aim of the stratification system is to maximise beneficence for everybody by individualised decision support. Still, potential ethical issues are related to this stratification model.

First, errors in the input data or the algorithm might mistakenly classify a person to the low-need pathway when the person, in reality, has high information needs. That could leave such individuals overwhelmed and with no professional help. The system foresees additional support and genetic counselling upon request from the individual to avoid such cases. Additionally, the platform should nudge people towards asking for additional counselling in case of doubts.

Second, it might be considered paternalistic to treat people unequally based on presumed decision support. While the system excludes nobody from personal counselling, it is mandatory in the high-need pathway and optional on the other pathways. Those classified in the high-need pathway might feel worse off if they do not see the benefits of genetic counselling. They might see it as a hurdle to predictive genetic testing others do not have. Thus, the stratification system involves a certain level of selective paternalism: Those whom the system considers having a high information need must undergo genetic counselling. By contrast, those with a medium or low information need are more independent in their choice of decision aids. In general, the system requires individuals to obtain information in order to take a genetic test, even if they do not want to. In my opinion, this is morally justified as it aims to support people making informed, autonomous and reflected decisions on predictive genetic testing.

In summary, I propose to individualise decision support for predictive genetic testing according to people's information needs. The algorithm determines these information needs depending on self-assessment questionnaires, the genetic testing application, genetic literacy and relevant health-related data. However, before such a stratification system is implementable, challenges include the collection of empirical evidence, social acceptance and practical issues, such as financing and administration. The next chapter now moves to a more abstract level, elaborating on how newspaper content analyses can be useful for bioethics research.

10.3 Newspaper content analyses in bioethics

Newspaper content analyses are interesting for bioethicists in various ways because they reliably describe how newspapers portray topics with ethical relevance to the public.[11] This descriptive dimension is, first, useful for pilot studies in research projects aiming to investigate the societal perspective of a phenomenon. For instance, in chapter 6, a quantitative screening explored what aspects of medical genetics recent newspaper reports had covered and how they framed it. Such information can be used for further analyses and inform, for instance, comparative newspaper content analyses (chapter 7; O'Mahony and Schäfer 2005), survey studies (Carducci et al. 2011;

11 See chapter 2.2.1.

Lynch et al. 2008), interview studies (Haran and Kitzinger 2013, 214) or experimental designs (King, Schneer, and White, A. 2017; Dunlop, Wakefield, and Kashima 2010).

Second, media content analyses are valuable indicators of public engagement with bioethical issues (Etchegary et al. 2015; chapter 7). As Gerhards and Schäfer (2009) propose, public engagement is expressed by having various stakeholders involved in the public discourse and represented in media coverage, evaluations are divers, and the debate is open to differing perspectives. This embeds science in society and allows for a two-sided exchange.

A third meaningful application of the description of the current newspaper coverage is to derive recommendations on how it *should be* from an ethical perspective. That includes concrete, practical recommendations, as demonstrated in chapter 8, and can serve as an example to link empirical and normative bioethics.[12] Since the reflection and reporting of ethical issues in newspaper coverage is a complex phenomenon, it might be fruitful to emphasise qualitative analyses and complement them with quantitative elements (see chapter 8).

Fourth, newspaper content analyses allow the investigations of social phenomena, for example, the Human Genome Project (Nerlich, Dingwall, and Clarke 2002; Tambor et al. 2002) or the Angelina Jolie effect (Sabel and Dal Cin 2016; Kamenova, Reshef, and Caulfield 2014). Longitudinal analyses of newspaper coverage to identify causes and triggers for coverage peaks help to identify such triggers for newspaper coverage (see chapter 6), following the issue attention cycle model (Kolb 2005).[13] Researchers can use such dynamics to enforce solutions to scientifically discovered issues with societal relevance; for instance, the missing information dissemination about predictive genetic testing.

In conclusion, even though newspaper content analyses cannot replace survey studies and can, from a bioethics perspective, mainly provide descriptive results, they are meaningful in various ways to assess public debates. As bioethical inquiries often investigate issues of societal relevance, newspaper content analyses can meaningfully contribute to assessing the current situation. The following chapter will declare the limitations and implications for further research.

12 See chapter 2.3 for a more detailed discussion about this issue.

13 See chapters 1.3.3 and 6.3.1 for a more detailed introduction.

10.4 Limitations and implications for further research

10.4.1 Limitations

This dissertation has several limitations. This section first addresses the limitations of the specific projects and then declares limitations concerning the overall dissertation. In the interview study, the sample size was small, and those refusing predictive genetic testing were underrepresented. The findings are only limitedly generalisable and need to be validated in other settings. However, the presented qualitative inquiry presents data for hypotheses for quantitative validation studies. Moreover, qualitative analyses require subjective interpretation, but we strived to overcome personal bias through team discussion, reflective memo writing and other established measures (see chapters 3, 4, 5 and 8). That also applies to the newspaper content analysis in chapter 8, where low reliability allowed for only limited quantitative analyses.

The comparative newspaper content analysis on predictive genetic testing (chapter 7) also had low sample sizes; thus, statistical analysis was not informative for all variables. Moreover, it only covered broadsheet newspapers, which reflect only part of the public debate. The pilot newspaper screening presented in chapter 6 included only few quantitative variables. Thus, the conclusions drawn from this analysis were limited in scope, but the study provided fruitful suggestions for further research. Media content analyses, in general, do not provide direct evidence for public attitudes.

More generally, this dissertation focused on predictive genetic testing, excluding other relevant applications, such as pharmacogenomics or prenatal genetic testing. In the newspaper content analyses, predictive genetic testing was defined broader than in the interview study, including direct-to-consumer genetic testing and research on potential new predictive tests. In contrast, the interview study was restricted to predictive genetic testing for genetic cancer predisposition in a clinical setting, and cancer-affected individuals were included, too, as they tested for the predisposition of other cancer types. These differences in the scope of the studies were considered when interpreting the results.

Another limitation concerns the link between the interview study and media analysis. It could have been meaningful to inform the newspaper content analyses based on the results from the interview study, especially regarding the Swiss context. However, the explorative nature of the interview study and the schedule did not allow waiting for the results. Nevertheless, other studies informed our newspaper content analyses (Schäfer 2009; Gerhards and Schäfer 2007), and I demonstrated in the discussion of this dissertation how these two project parts could be meaningfully interlinked.

Similarly, the work might have benefitted from delving into normative implications at an earlier stage. As explained in chapter 2, the normative research questions only arouse during the process of data analysis. Formulating the normative aims in advance would have made data collection more tailored. At the same time, explorative research includes discovering new research questions during the process of analysis and the normative conclusions are meaningful.

The stratification system is a new idea concerning future challenges. It is challenging to use it in the current system, and a lot of scientific and political efforts are necessary to implement such a system into practice in an ethical and meaningful way. Still, as the information bottleneck will be a challenging issue to solve, it is crucial to take a forward-looking perspective and discuss possible alternatives proactively.

Finally, all practical recommendations specifically apply to predictive genetic testing. Applications to other areas are conceivable but should be assessed thoroughly. The next section provides insights into future research projects that can follow on from this dissertation.

10.4.2 Further research

Various research projects following the results of this dissertation are conceivable. First, this dissertation assessed information dissemination of predictive genetic testing for informed decision-making from the media, the patients' and the healthcare professionals' perspective. A population survey and assessing the perception of other relevant stakeholders, such as policymakers, health insurances or the pharmaceutical and biotech industry might give additional valuable insights and should be included in future studies.

Second, research on customers of direct-to-consumer genetic testing regarding their information-seeking behaviour, information needs and information source is needed. They might differ from at-risk individuals interested in predictive genetic testing in a clinical setting. Besides, it would be interesting to critically assess how informed and reflected their decision to buy a genetic test online is.

A third follow-up study concerns the quantification of the here-presented qualitative results. Patient questionnaires or population-wide surveys are tools to investigate the predictive genetic testing decision-making process and the level of information people want and need for decision-making quantitatively. Regarding the stratification of people according to their information needs (chapter 10.2.2), it is crucial to operationalise informed decision-making for such survey studies. Additionally, quantitative studies are necessary to implement this stratification algorithm, including data collection for input variables, validation studies and studies on attitudes towards such an approach.

Finally, another relevant research project concerns healthcare professionals' attitudes towards genetic counselling decision support and alternatives to genetic counselling in underexamined countries, such as Switzerland. That will identify practical challenges and might reveal alternatives that have so far not been considered. Qualitative interview studies, focus groups or surveys are the most suitable format to start such an analysis.

10.5 Conclusion

This dissertation investigated the phenomenon of informed decision-making for predictive genetic testing from the at-risk individuals' and the mass media's perspective. The empirical evidence in this dissertation is limited to predictive genetic testing, excluding prenatal and pharmacogenomic testing, and focuses mainly on the context of Switzerland, with particular emphasis on the German-speaking region.

The status quo in Switzerland, where at-risk individuals usually have low prior knowledge and heavily rely on personal genetic counselling, is not promising for the future: First, the scientific progress and infiltration of genetics in healthcare and society will lead to public debates regarding reimbursement issues, status of preventive medicine and implications for future generations. Due to the complexity of these topics, people need to be equipped with basic knowledge. While mass media are useful to create awareness, occasionally inform on topics of interest and provide a forum for public debates, professional online information material and school education should provide in-depth and accessible information. Second, with the increasing number of individuals becoming eligible for predictive genetic testing, limited personal resources will become a pressing matter, especially in the German-speaking part. In light of this resource shortage, the findings of the presented work provide insights on how to save resources by supplying directed decision support to individuals based on their specific information needs.

This dissertation further illustrated how newspaper content analyses could be useful for empirical bioethics research. Several fields of applications are conceivable: first, the content analysis itself can focus on how newspapers portray ethical issues to the public. Second, it can describe how newspaper coverage currently is, and make recommendations on how it *should be* from an ethical perspective. Third, newspaper content analyses might be useful in preparation for an experimental design regarding their effects on participants. Fourth, newspaper content analyses provide the possibility to explore a so far underresearched phenomenon efficiently and reliably from a societal perspective and use these insights to inform studies involving members of the public, such as surveys.

In summary, this dissertation aimed to investigate what information at-risk individuals and the public should receive regarding predictive genetic testing, and how such information should be transmitted. While all relevant information should be made available on professional online platforms, mass media coverage should primarily focus on information relevant to public debates and raise awareness about ethical and social issues and the limits, risks and benefits of predictive genetic testing. While personal genetic counselling standard clinical practice, personal resources are limited and, thus, alternatives need to be implemented. E-education and personalised decision-support can provide useful tools in this respect. The findings of this dissertation fill a research gap regarding predictive genetic testing information dissemination and informed decision-making in Switzerland, sheds light on how Swiss newspapers report about predictive genetic testing as compared to the United Kingdom, and provides practical recommendations on how to improve information dissemination to at-risk individuals and the public.

10.6 References

Adam, Shelin, Patricia H. Birch, Rachel R. Coe, Nick Bansback, Adrian L. Jones, Mary B. Connolly, Michelle K. Demos, Eric B. Toyota, Matthew J. Farrer, and Jan M. Friedman. 2018. "Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions." J Genet Couns 28:10–17. doi:10.1007/s10897-018-0281-1.

Amt für Volksschulen Kanton Basel-Landschaft. 2019. "Lehrplan Volksschule Basel-Landschaft: Erläuterung 'Stoffinhalte Und Themen' 3. Zyklus Biologie." Accessed December 25, 2019. https://www.baselland.ch/politik-und-behorden/direktionen/bildungs-kultur-und-sportdirektion/bildung/handbuch/unterricht/stundentafeln-lehrplaene/volksschule/biologie-3-zyklus/dokumente-biologie-3-zyklus/erlaeuterung-biologie.pdf/@@download/file/Erlaeuterung_Biologie.pdf.

Aston, Robert. 1998. "Genetic Counselling Must Be Non-Directive." BMJ 317 (7150): 82. doi:10.1136/bmj.317.7150.82.

Badenschier, Franziska, and Holger Wormer. 2012. "Issue Selection in Science Journalism: Towards a Special Theory of News Values for Science News?" In Rödder, Franzen, and Weingart 2012, 59–86.

Beauchamp, Tom L., and James F. Childress. 2013. Principles of Biomedical Ethics. 7th ed. New York: Oxford University Press.

Benusiglio, Patrick R., Marina Di Maria, Leila Dorling, Anne Jouinot, Antoine Poli, Sophie Villebasse, Marine Le Mentec, Beatrice Claret, Diane Boinon, and Olivier Caron. 2017. "Hereditary Breast and Ovarian Cancer: Successful Systematic Implementation of a Group Approach to Genetic Counselling." Fam Cancer 16 (1): 51–56. doi:10.1007/s10689-016-9929-x.

Bildungsdirektion des Kantons Zürich. 2017. "Lehrplan Für Die Volksschule Des Kantons Zürich Auf Der Grundlage Des Lehrplans 21." Accessed December 25, 2019. https://zh.lehrplan.ch/index.php?code=a|6|2|8|0|3.

biorespect. 2020. "Gen-Test.Info." Accessed January 29, 2020. https://gen-test.info/.

Buchanan, Adam H., Alanna K. Rahm, and Janet L. Williams. 2016. "Alternate Service Delivery Models in Cancer Genetic Counseling: A Mini-Review." Front Oncol 6:120. doi:10.3389/fonc.2016.00120.

Bunnik, Eline M., Maartje H. N. Schermer, and A. C. J. W. Janssens. 2014. "Naming and Framing in Genomic Testing." Trends Mol Med 20 (2): 63–65. doi:10.1016/j.molmed.2013.11.001.

Carducci, Annalaura, Simona Alfani, Manuela Sassi, Alessandra Cinini, and Andrea Calamusa. 2011. "Mass Media Health Information: Quantitative and Qualitative Analysis of Daily Press Coverage and Its Relation with Public Perceptions." Patient Educ Couns 82 (3): 475–78. doi:10.1016/j.pec.2010.12.025.

Case, Donald O. 2007. Looking for Information: A Survey of Research on Information Seeking, Needs, and Behavior. 2nd ed. Library and Information Science. London.

Caulfield, Timothy A., and Celeste M. Condit. 2012. "Science and the Sources of Hype." Public Health Genom 15 (3-4): 209–17. doi:10.1159/000336533.

Chae, Jiyoung, Chul-joo Lee, and Jakob D. Jensen. 2016. "Correlates of Cancer Information Overload: Focusing on Individual Ability and Motivation." Health Commun 31 (5): 626–34. doi:10.1080/10410236.2014.986026.

Christman, John. 2018. "Autonomy in Moral and Political Philosophy." In the Stanford Encyclopedia of Philosophy, edited by Edward N. Zalta. Spring 2018: Metaphysics Research Lab, Stanford University.

Cohen, Stephanie A., Monica Marvin, Bronson D. Riley, Hetal S. Vig, Julie A. Rousseau, and Shanna L. Gustafson. 2013. "Identification of Genetic Counseling Service Delivery Models in Practice: A Report from the NSGC Service Delivery Model Task Force." J Genet Couns 22 (4): 411–21. doi:10.1007/s10897-013-9588-0.

Condit, Celeste M. 2010. "Public Understandings of Genetics and Health." Clin Genet 77 (1): 1–9. doi:10.1111/j.1399-0004.2009.01316.x.

Corbellini, Gilberto. 2004. "Genetic Risk, Medical Education, Public Understanding of Genetics and Evolutionary Medicine: The Challenges of Genetic Counselling for Complex Disorders." Topoi 23 (2): 187–93. doi:10.1007/s11245-004-5379-4.

de Vries, Hein, Ilse Mesters, Hermanna van de Steeg, and Cora Honing. 2005. "The General Public's Information Needs and Perceptions Regarding Hereditary Cancer: An Application of the Integrated Change Model." Patient Educ Couns 56 (2): 154–65. doi:10.1016/j.pec.2004.01.002.

Deutsche Gesellschaft für Humangenetik e.V., and Berufsverband Deutscher Humangenetiker e.V. 2018. "S2k-Leitlinie Humangenetische Diagnostik Und Genetische Beratung." medgen 30 (4): 469–522. doi:10.1007/s11825-018-0223-1.

Deutschschweizer Erziehungsdirektoren-Konferenz. 2016. "Lehrplan Für Die Volksschule Des Kantons Basel-Stadt." Accessed December 25, 2019. https://bs.lehrplan.ch/index.php?code=a|6|2|8|0|3.

Djurdjinovic, Luba, and June Peters. 2017. "Special Issue Introduction: Dealing with Psychological and Social Complexity in Genetic Counseling." J Genet Couns 26 (2): 1–4. doi:10.1007/s10897-017-0080-0.

Dunlop, Sally M., Melanie Wakefield, and Yoshihisa Kashima. 2010. "Pathways to Persuasion: Cognitive and Experiential Responses to Health-Promoting Mass Media Messages." Commun Res 37 (1): 133–64. doi:10.1177/0093650209351912.

Etchegary, Holly, Jane Green, Patrick Parfrey, Catherine Street, and Daryl Pullman. 2015. "Community Engagement with Genetics: Public Perceptions and Expectations About Genetics Research." Health Expect 18 (5): 1413–25. doi:10.1111/hex.12122.

Etchegary, Holly, Fiona Miller, Sonya deLaat, Brenda J. Wilson, June C. Carroll, and Mario Cappelli. 2009. "Decision-Making About Inherited Cancer Risk: Exploring Dimensions of Genetic Responsibility." J Genet Couns 18 (3): 252–64. doi:10.1007/s10897-009-9218-z.

Faden, Ruth R., and Tom L. Beauchamp. 1986. A History and Theory of Informed Consent. With the assistance of N. M. P. King. New York: Oxford University Press.

Fondation Leenaards, and Médecine et Hygiène. 2019. "Santé Personnalisée & Société." Accessed December 23, 2019. https://www.santeperso.ch/.

Franzen, Martina, Peter Weingart, and Simone Rödder. 2012. "Exploring the Impact of Science Communication on Scientific Knowledge Production: An Introduction." In Rödder, Franzen, and Weingart 2012, 3–16.

Früh, Werner, and Klaus Schönbach. 2005. "Der Dynamisch-Transaktionale Ansatz III: Eine Zwischenbilanz." Publizistik 50 (1): 4–20. doi:10.1007/s11616-005-0115-7.

Gerhards, Jürgen, and Mike S. Schäfer. 2006. Die Herstellung einer öffentlichen Hegemonie: Humangenomforschung in der deutschen und der US-amerikanischen Presse. Wiesbaden: Verlag für Sozialwissenschaften.

———. 2007. "Hegemonie der Befürworter. Der mediale Diskurs über Humangenomforschung." Soz Welt 58 (4): 367-395.

———. 2009. "Two Normative Models of Science in the Public Sphere: Human Genome Sequencing in German and US Mass Media." Public Underst Sci 18 (4): 437–51. doi:10.1177/0963662507082891.

Gordon, Erynn S., Deepti Babu, and Dawn A. Laney. 2018. "The Future Is Now: Technology's Impact on the Practice of Genetic Counseling." Am J Med Genet C 178 (1): 15–23. doi:10.1002/ajmg.c.31599.

Haga, Susanne B., William T. Barry, Rachel Mills, Geoffrey S. Ginsburg, Laura Svetkey, Jennifer Sullivan, and Huntington F. Willard. 2013. "Public Knowledge of and Attitudes Toward Genetics and Genetic Testing." Genet Test Mol Biomarkers 17 (4): 327–35. doi:10.1089/gtmb.2012.0350.

Hamilton, Rebekah J., and Barbara J. Bowers. 2007. "The Theory of Genetic Vulnerability: A Roy Model Exemplar." Nurs Sci Q 20 (3): 254–64. doi:10.1177/0894318407303127.

Haran, Joan, and Jenny Kitzinger. 2013. "Human Genetics and Cloning in the Media: Mapping the Research Field." In Handbook of Genetics and Society: Mapping the New Genomic Era, edited by Paul Atkinson, Peter Glasner, and Margaret Lock. Abington: Routledge.

Kahneman, Daniel. 1994. "New Challenges to the Rationality Assumption." J Inst Theor Econ 150 (1): 18–36.

Kamber, Esther, and Kurt Imhof. 2011. "Qualitätsmedien Sind Wissensvermittler." In Krise Der Leuchttürme Öffentlicher Kommunikation: Vergangenheit Und Zukunft Der Qualitätsmedien, edited by Roger Blum, Heinz Bonfadelli, Kurt Imhof, and Otfried Jarren, 131–50. Wiesbaden: Verlag für Sozialwissenschaften.

Kamenova, Kalina, Amir Reshef, and Timothy A. Caulfield. 2014. "Angelina Jolie's Faulty Gene: Newspaper Coverage of a Celebrity's Preventive Bilateral Mastectomy in Canada, the United States, and the United Kingdom." Genet Med 16 (7): 522–28. doi:10.1038/gim.2013.199.

Khaleel, Israa, Barbara C. Wimmer, Gregory M. Peterson, Syed T. R. Zaidi, Erin Roehrer, Elizabeth Cummings, and Kenneth Lee. 2020. "Health Information Overload Among Health Consumers: A Scoping Review." Patient Educ Couns 103 (1): 15–32. doi:10.1016/j.pec.2019.08.008.

King, Gary, Benjamin Schneer, and Ariel White. 2017. "How the News Media Activate Public Expression and Influence National Agendas." Science 358 (6364): 776–80. doi:10.1126/science.aao1100.

Kolb, Steffen. 2005. Mediale Thematisierung in Zyklen: Theoretischer Entwurf und empirische Anwendung. Köln: Herbert von Halem.

Luhmann, Niklas. 2009. Die Realität der Massenmedien. 4th ed. Neue Bibliothek der Sozialwissenschaften. Wiesbaden: Verlag für Sozialwissenschaften.

Lynch, John, Jennifer Bevan, Paul Achter, Tina M. Harris, and Celeste M. Condit. 2008. "A Preliminary Study of How Multiple Exposures to Messages About Genetics Impact on Lay Attitudes Towards Racial and Genetic Discrimination." New Genet Soc 27 (1): 43–56. doi:10.1080/14636770701843634.

Lynöe, Niels, and Klaus Hoeyer. 2005. "Quantitative Aspects of Informed Consent: Considering the Dose Response Curve When Estimating Quantity of Information." J Med Ethics 31 (12): 736–38. doi:10.1136/jme.2005.012013.

Manson, Neil, and Onora O'Neill. 2007. Rethinking Informed Consent in Bioethics. Cambridge: Cambridge University Press.

Marteau, Theresa M., Elizabeth Dormandy, and Susan Michie. 2001. "A Measure of Informed Choice." Health Expect 4 (2): 99–108. doi:10.1046/j.1369-6513.2001.00140.x.

Marzuillo, Carolina, Corrado de Vito, E. D'Andrea, Annalisa Rosso, and Paolo Villari. 2013. "Predictive Genetic Testing for Complex Diseases: A Public Health Perspective." An International Journal of Medicine 107 (2): 93–97. doi:10.1093/qjmed/hct190.

McBride, Colleen M., Deborah J. Bowen, Lawrence C. Brody, Celeste M. Condit, Robert T. Croyle, Marta Gwinn, Muin J. Khoury et al. 2010. "Future Health Applications of Genomics: Priorities for Communication, Behavioral, and Social Sciences Research." Am J Prev Med 38 (5): 556–65. doi:10.1016/j.amepre.2010.01.027.

Moore, Rebekah A., Anne L. Matthews, and Leslie Cohen. 2018. "How Might the Genetics Profession Better Utilize Social Media." J Genet Couns. doi:10.1007/s10897-018-0215-y.

Nerlich, Brigitte, Robert Dingwall, and David D. Clarke. 2002. "The Book of Life: How the Completion of the Human Genome Project Was Revealed to the Public." Health 6 (4): 445–69. doi:10.1177/136345930200600403.

O'Mahony, Patrick, and Mike S. Schäfer. 2005. "The 'Book of Life' in the Press Comparing German and Irish Media Discourse on Human Genome Research." Soc Stud Sci 35 (1): 99–130. doi:10.1177/0306312705046542.

Otten, Ellen, Erwin Birnie, Anneke Lucassen, Adelita V. Ranchor, and Irene M. van Langen. 2016a. "Telemedicine Uptake Among Genetics Professionals in Europe: Room for Expansion." Eur J Hum Genet 24 (2): 157–63. doi:10.1038/ejhg.2015.83.

Otten, Ellen, Erwin Birnie, Adelita V. Ranchor, and Irene M. van Langen. 2016b. "Telegenetics Use in Presymptomatic Genetic Counselling: Patient Evaluations on Satisfaction and Quality of Care." Eur J Hum Genet 24 (4): 513–20. doi:10.1038/ejhg.2015.164.

Pennacchini, M., and C. Pensieri. 2011. "Is Non-Directive Communication in Genetic Counseling Possible?" Clin Ter 162 (5): e141-144.

Peters, Hans P. 1994. "Mass Media as an Information Channel and Public Arena." Risk 5: 241.

Reumkens, Kelly, Marly H. E. Tummers, Joyce J. G. Gietel-Habets, Sander M. J. van Kuijk, Cora M. Aalfs, Christi van Asperen, Margreet G. E. M. Ausems et al. 2019. "Online Decision Support for Persons Having a Genetic Predisposition to Cancer and Their Partners During Reproductive Decision-Making." J Genet Couns 28 (3): 533–42. doi:10.1002/jgc4.1056.

Rödder, Simone, Martina Franzen, and Peter Weingart, eds. 2012. The Sciences' Media Connection - Public Communication and Its Repercussions. Sociology of the sciences A yearbook. Dordrecht: Springer.

Sabel, Michael S., and Sonya Dal Cin. 2016. "Trends in Media Reports of Celebrities' Breast Cancer Treatment Decisions." Ann Surg Oncol 23 (9): 2795–2801. doi:10.1245/s10434-016-5202-7.

Schäfer, Mike S. 2009. "From Public Understanding to Public Engagement: An Empirical Assessment of Changes in Science Coverage." Sci Commun 30 (4): 475–505. doi:10.1177/1075547008326943.

Scheufele, Dietram A., and David Tewksbury. 2007. "Framing, Agenda Setting, and Priming: The Evolution of Three Media Effects Models." J Commun 57 (1): 9–20. doi:10.1111/j.1460-2466.2006.00326.x.

Schwitzer, Gary, Ganapati Mudur, David Henry, Amanda Wilson, Merrill Goozner, Maria Simbra, Melissa Sweet, and Katherine A. Baverstock. 2005. "What Are the Roles and Responsibilities of the Media in Disseminating Health Information?" PLoS Med 2 (7): 576–82. doi:10.1371/journal.pmed.0020215.

Severin, Franziska, Pascal Borry, Martina C. Cornel, Norman Daniels, Florence Fellmann, Shirley Victoria Hodgson, Heidi C. Howard et al. 2015. "Points to Consider for Prioritizing Clinical Genetic Testing Services: A European Consensus Process Oriented at Accountability for Reasonableness." Eur J Hum Genet 23 (6): 729–35. doi:10.1038/ejhg.2014.190.

Shoemaker, Pamela J. 2002. "A New Gatekeeping Model." In McQuail's Reader in Mass Communication Theory, edited by Denis McQuail, 253–59. London: SAGE Publications.

Smerecnik, Chris M. R., Ilse Mesters, Nanne K. de Vries, and Hein de Vries. 2008. "Educating the General Public About Multifactorial Genetic Disease: Applying a Theory-Based Framework to Understand Current Public Knowledge." Genet Med 10 (4): 251–58. doi:10.1097/GIM.0b013e31816b4ffd.

Swiss Federal Council. 2004. "CC 810.12 Federal Act of 8 October 2004 on Human Genetic Testing (HGTA)." Accessed January 23, 2020. https://www.admin.ch/opc/en/classified-compilation/20011087/index.html.

———. 2017. "Draft of the Revised Human Genetic Testing Act." Accessed January 23, 2020. https://www.bag.admin.ch/dam/bag/de/dokumente/biomed/genetischeuntersuchung/aktuelle-rechtsetzungsprojekte/entwurf-gumg-2017.pdf.download.pdf/entwurf-gumg-2017.pdf.

Tambor, Ellen S., Barbara A. Bernhardt, Joann Rodgers, Neil A. Holtzman, and Gail Geller. 2002. "Mapping the Human Genome: An Assessment of Media Coverage and Public Reaction." Genet Med 4 (1): 31. doi:10.1097/00125817-200201000-00006.

Weil, Jon, Kelly Ormond, June Peters, Kathryn Peters, Barbara B. Biesecker, and Bonnie LeRoy. 2006. "The Relationship of Nondirectiveness to Genetic Counseling: Report of a Workshop at the 2003 NSGC Annual Education Conference." J Genet Couns 15 (2): 85–93. doi:10.1007/s10897-005-9008-1.

Weingart, Peter, and Patricia Schulz. 2014. "Einleitung: Das schwierige Verhältnis zwischen Wissenschaft, Öffentlichkeit und Medien." In Wissen - Nachricht - Sensation: Zur Kommunikation zwischen Wissenschaft, Öffentlichkeit und Medien, edited by Peter Weingart and Patricia Schulz, 9–18. Weilerswist: Velbrück.

White, Lucie. 2018. "The Need for Authenticity-Based Autonomy in Medical Ethics." HEC Forum 30 (3): 191–209. doi:10.1007/s10730-017-9335-2.

Wolfe, Christopher R., Valerie F. Reyna, Colin L. Widmer, Elizabeth M. Cedillos, Christopher R. Fisher, Priscila G. Brust-Renck, and Audrey M. Weil. 2015. "Efficacy of a Web-Based Intelligent Tutoring System for Communicating Genetic Risk of Breast Cancer: A Fuzzy-Trace Theory Approach." Med Decis Making 35 (1): 46–59. doi:10.1177/0272989X14535983.

Appendices

Appendix A: Interview study

A-1 Interview guide patient interviews[1]

1. When did you first get in touch with genetic testing? Why did you go to genetic counselling?
2. How was your decision-making process regarding genetic testing?
   - What were the reasons for your decision?
   - Which factors influenced your decision?
   - Who did you exchange your thoughts with during the decision-making process?
   - Was your decision more of a gut or a head decision?
3. How did you feel during the testing process?
   - Who were your exchange partners during this time?
   - How did you deal with the uncertainty?
4. Do you experience or fear positive or negative consequences due to your genetic test?
   - If so, what are these consequences?
   - How do you deal with the test result?
   - How do you deal with the consequences? Do you have specific strategies?
5. With whom do you talk about genetic testing and its consequences?
   - Have other family members also had themselves tested?
   - Who can or do you not want to talk to about it? Why?
   - Can you talk openly about the subject?
6. If an at-risk individual would ask for your advice regarding genetic testing, what would you reply?
7. Do you associate something negative/positive with genetic testing? *(ask for an opposite position than previously displayed by the participant)*
8. Where did you find information about genetic testing?
   - What were your expectations of the genetic consultations?

1 Please note: the interview guide was adapted in the course of the study according to findings. Vignettes were introduced after six interviews. Vignette 2 was removed from the interview guide after interview 11. Question 7 was introduced after interview 11. The other changes concerned the follow-up questions that were asked depending on the participant's reactions to the main questions (bold).

- Did you obtain information about the genetic test from other sources in addition to genetic counselling? If so, where? What kind of information?
- Would you have liked to have more alternative sources of information?
- Did you hear/read about genetic testing in the media?
- What do you think about the presentation of genetic tests in the media?
- What were your information needs? What interested you most?

9. Is there anything else on the subject that you would like to add?
10. Finally, may I ask you for some demographic information?
    - How old are you?
    - What is your profession? What is your education?
    - If not already mentioned: What is your civil status? Do you have siblings? Do you have children?
    - Do you live in the city or in the countryside?

Hypothetical vignettes

Vignette 1: Imagine there is a genetic test that can determine an increased risk of 40% for a possible outbreak of a severe, fatal hereditary disease. The exact time of onset of the disease cannot be determined. Unfortunately, there are no preventive measures for this disease and treatment is only possible symptomatically (to alleviate suffering). You have already lost one parent to this disease.

- Would you want to do a genetic test or not? Why (not)?
- What if the risk only increased by 10-20%? Or by 50-80%? Would there be a percentage threshold for you as to when the test is still useful?
- What if the outbreak could be predicted to be 100%?

Vignette 2: Imagine a friend of yours should undergo a genetic test. If the test result is positive, the risk of getting the disease would rise to 40% and there would be six-monthly check-ups to detect the disease as early as possible. Your friend, however, has been planning a two-year trip around the world for a long time, and the preventive examinations would not be feasible during this period. In your opinion, should your friend have the genetic test anyway?

Vignette 3: Imagine your family doctor telling you about a genetic test that can tell you how high your personal genetic risk is of contracting a widespread disease - for example, diabetes, high blood pressure or a heart attack. This risk is also influenced by personal behaviours, such as diet, exercise, etc. Would you like to take such a test? Who would you recommend this test to? What are the advantages and disadvantages of this test?

A-2 Interview guide expert interviews

Thank you for taking the time and speaking with me! To complement the patient interviews, I would like to ask you, as an expert, some additional questions that arose during the patient interviews. This interview will take approximately 60 minutes and is confidential. Only the analyzing team and I have direct access to the tape. All the mentioned names and places will be encrypted in the transcript. In publications, care is taken to ensure that no statements can be traced back to your institution (and thus indirectly to you as a person) unless you explicitly agree. Some questions are rather technical, but I am also interested in your personal opinion regarding improvement suggestions or problems.

Do you agree to this interview? Do you have any remaining questions before we start?

1. We found that certainty and uncertainty play an important role in genetic testing. Do you remember the last genetic genetic consultation where this was discussed?
    - What uncertainties do you face?
    - How do you convey certainty?
2. From your perspective, how do patients cope with the information provided during the genetic consultation?
    - Do you experience some patients being overwhelmed?
    - How do you assess the understanding of your patients after the consultations regarding risk assessment?
    - When you are under the impression that patients do not fully understand the concept of risk, how do you deal with it?
    - Do patients frequently ask for recommendations? How do you handle them?
3. How often do you experience that patients are still undecided whether to take the test or not when coming to the first genetic consultation?

4. What concerns do patients address during the genetic consultation? Have patients ever mentioned to you that they find it difficult to talk to others about it?
    - With whom?
    - What do you advise your patients to do in this case?
5. Are there patients concerned about discrimination?
    - What do you tell these patients?
    - How important do you think is the risk of discrimination in the case of diagnosed HBOC or Lynch Syndrome?
    - What kind of discrimination have you experienced with your patients?
6. Do you remember patients who regretted their decision after testing?
    - Why?
    - How did you and these patients cope with this situation?
    - Does such an incident influence future genetic consultations?
7. In your opinion, who should be responsible for genetic counselling?
8. How is the information dissemination to family members handled?
9. Is it common to conduct BRCA genetic testing in cancer patients without genetic counselling? How does the cooperation between oncologists and geneticists look like?
10. Do you see patients that were counselled by medical doctors not specialized in genetics? How do you handle these situations?
11. How standardized is the referral procedure? Where do you see room for improvement?
12. What is the indication for panel testing in the case of HBOC?
13. Are there any statistics on how many people make use of genetic counselling and how many then decide not to take a test? If not: what do your estimate?
14. How often do you get questions from patients after the consultations?
15. Which information aids do you use in your genetic consultations?
16. Do individuals who have been tested positive perceive themselves as patients or as healthy individuals in the case of predictive testing? What is your impression?
17. What do you think about predictive genetic testing for nonactionable diseases?
18. What do you think about predictive genetic testing for diseases with high environmental influences, for instance for cardiovascular diseases or diabetes

A-3 Study information sheet

Attitudes, reasons and information strategies of people undergoing genetic counselling in Switzerland. Genetic testing of inherited cancer risk.

Project organization and responsibility: Prof Bernice Elger, Institute for Biomedical Ethics, University of Basel.

Researcher and Co-responsibility: Bettina Zimmermann, Doctoral student

Dear Sir or Madam

We would like ask you to take part in a research project. Below the project will be presented to you in detail.

Aim of the project

With this project we aim to assess the attitudes, reasons and information strategies of people undergoing genetic counselling for testing of inherited cancer risk.

Selection of participants

People undergoing genetic counselling at the university hospitals in Basel or Bern are eligible to participate in the study. Participants have to be between 18 and 65 years of age, speak German, French or English and should not be pregnant at the time of participation. Whether you decide to take a genetic test or not is not an issue for participation.

General information about the project

Genetic sciences have progressed significantly in the last two decades. Thus, genetic testing has become more frequent and broader in applications in clinical practice. It will be possible to test more and more people for more and more genetic diseases in the near future. This may lead to better prevention of and cure of diseases, but also to difficult decisions. An explicit, effective and sustainable information strategy can help affected people, because they can facilitate decision making in favour of or against learning certain genetic knowledge. As a first step, we aim to analyse the attitudes, reasons and information strategies of potential users of genetic testing today.

With this study we aim to gain insights and knowledge so far unknown in Switzerland. To do so we will interview up to 25 people who have had a genetic counselling session. The project runs from May 2017 until December 2018 and is executed in accordance with Swiss law. The responsible ethics committee has examined and approved the project.

Study process

If you are interested in participating in this study, please contact the interviewer Bettina Zimmermann via email or phone. This contact is non-binding and does not correspond to consent for participation. You will be then orally informed about the study details. If after that you accept to participate in an interview, you will sign the informed consent form at end of this document. Bettina Zimmermann will then make an appointment with you for the interview. The interview will take place at the hospital where you had the genetic counselling.

The interview will take about one hour and will be conducted personally. It will be recorded and later on transcribed and encrypted. The encrypted transcript will be analysed by our research team, interpreted and compared with other interviews.

Benefit of participation

Some participants in such interview studies find it helpful to formulate and reflect their opinion in the context of an interview. The results we get from yours and other participants' interviews are also important for other affected people thinking about genetic testing in the future, as the field of application for genetic testing is enlarging constantly. Knowing information strategies and reasons for decision making of users of genetic testing can help to improve the health and information systems. The results will be interesting for all stakeholders involved: hospitals, general practitioners, medical specialists, politicians, insurances and many more.

Rights of participants

You participate in this study on your own free will. You do not have to justify your decision if you decide not to participate or with to withdraw your consent at a later stage. Your medical care is guaranteed independently of your decision to participate. If you have questions regarding participation and the project you may ask the responsible team anytime.

Duties of participants

As participant we would ask you for being on time for the arranged interview. If you change your mind and would like to withdraw your consent to give an interview (which is possible anytime and without any consequences) please inform the interviewer Bettina Zimmermann (via email or phone, contact details see below).

Risks

Participating in this project will only put you at the small risk of some mental stress because of the interview. Stressful questions at the interview can be skipped anytime and the interview can even be interrupted or stopped if necessary.

Results

If you wish we will inform you about the results of the study as soon as the analysis is completed. We aim to have first results in autumn 2018. We would be very interested in your feedback, and whether you can recognise your experience in the results. The results of this study have nothing to do with your personal genetic analysis.

Confidentiality of data

In this project we interview you about your personal and medical history. Only very few people (Bettina Zimmermann and her supervisors) will have access to your unencrypted interview data. Other people involved in the analysis will only see the encrypted transcription of your interview. Encrypted means that all data that could identify you (like names, places, date of birth etc.) will be replaced by a code. People that do not know the code key can thus not identify you as a person. All researchers involved in the project are bound to professional discretion. The code key is placed only at the Institute of Biomedical Ethics, University of Basel, Bernoullistrasse 28, 4056 Basel. As a participant you have the right to access your data at all times. The interview recording as well as the encrypted transcript will be secured and be kept for a maximum of five years. After that, all data will be destroyed.

It is possible that the responsible ethics committee will retrospectively assess the quality of this study project. For that, the project leader may have to disclose your interview data for these purposes.

Withdrawal of consent

You can stop at any time and withdraw from participating in this study project. You can furthermore demand the destruction of the audio tape of your interview in case of resignation. If an encrypted transcript is already existent it may be used for analysis. Please note that in the transcript, personal data is encrypted and only researchers involved in the project are allowed to access the transcript.

Compensation

No compensation is foreseen for the participation in this study project. Travel expenses caused by the study participation will be refunded.

Liability

If the participation in this study project causes you any harm, the institution inducing the projet and responsible for the study execution is liable. The requirements for the procedure are regulated by law.

Funding

This study project is funded by the Institute for Biomedical Ethics for the University of Basel.

Contact

In case of questions, uncertainties or emergencies in the framework of this study project you may refer to these contacts anytime.

Execution of the study project:

Bettina Zimmermann, doctoral student

bettina.zimmermann@unibas.ch

+41 61 207 17 85

Supervision and responsible institution:

Dr. David Shaw, supervision (david.shaw@unibas.ch)

Prof. Bernice Elger, head of institute (b.elger@unibas.ch)

University of Basel

Institute for Biomedical Ethics, IBMB

Bernoullistrasse 28, 4056 Basel

+41 61 207 17 86

http://ibmb.unibas.ch

Informed consent form

Written informed consent for participation in a study project

Please read this form carefully. Please ask if you have any questions or have some uncertainties.

BASEC number (after submission):	2017-00316
Title of the study (wissenschaftlich und Laiensprache):	Attitudes, reasons and information strategies of people undergoing genetic counselling for inherited cancer risk
Responsible institution (project lead with address):	Institute for Biomedical Ethics, University of Basel Prof. Bernice Elger, head of institute Dr. David Shaw, supervisor Bettina Zimmermann, co-responsible and executor Bernoullistrasse 28, 4056 Basel
Study location:	University hospital Basel / Institute for Biomedical Ethics
Project leader at the place of study:	Prof. Bernice Elger
Participant: Name and surname in block letters: date of birth:	☐ female ☐ male

- I was informed in written and oral form by the signing responsible person about the purpose and the procedure of the study, about possible advantages and disadvantages as well as about possible risks.
- I participate in this project of my own free will and accept the content of the written study information above. I have had enough time to make my decision.
- My questions related to the participation in this study project have been answered. I keep the study information and get a copy of my written informed consent.
- I agree that my genetic counsellor (medical doctor) can be informed about my participation in that project if necessary.
- I agree that the responsible experts of the project lead and the responsible ethics committee can review my unencrypted data for examination and controlling under strict confidentiality.
- I can withdraw my consent anytime without any justification without having any disadvantages in my medical treatement/care.
- The liability insurance of the institution will cover possible harm.

Place, date	Signature participant

Confirmation of signing responsible person

I hereby confirm that I have explained the essence, purpose and importance of the project to this participant. I assure I will fulfil all obligations in the context of this project according to the applicable law. If I at any point in time during the study I learn about aspects that might influence the willingness of the participant to participate in this study project I will inform the participant immediately.

Place, date	Signature participant
	Name and surname of the informing responsible person Bettina Zimmermann Signature of responsible

Appendix B: Public discourse about medical genetics in Switzerland

B-1 Search algorithms[2]

German sampling

First round

Gentest* OR genetisch* Test* OR genetisch* Krankheit* OR genetisch* Erkrankung* OR Angelina Jolie Effekt* OR Angelina-Jolie-Effekt* OR Angelina-Effekt* OR Angelina-Gen* OR BRCA OR vererbbar* Brustkrebs OR Lynch Syndrom* OR Lynch-Syndrom* OR familiär* adenomatöse* Polyposis OR vererbbar* Darmkrebs OR vererbbar* Krebsveranlagung* OR CRISPR OR Genomeditierung OR Genom* editier* OR Genom-Editierung* OR Präimplantationsdiagnostik OR Biobank* OR (selten* Krankheit* AND (genet* OR Gendefekt* OR gentechni*)) OR personalisiert* Medizin OR personalisiert* Gesundheit OR genomisch* Medizin or PID not ("pid=" OR primäre* Immundefizienz* OR potenzialinduzierte* Degradation*)

Second round

((Angelina Jolie* AND (genet* OR BRCA*)) OR BRCA1 OR BRCA2) OR (genetische* Untersuchung* or GUMG) OR (Genom-Sequenzierung* OR Genomsequenzierung* OR DNA-Sequenzierung*) OR (Gentherapie* OR Gen-Therapie* OR Genmodifikation* OR Gen-Modifikation* OR Humangenetik*) OR (Big data AND genet*) NOT (Gentest* OR genetisch* Test* OR genetisch* Krankheit* OR genetisch* Erkrankung* OR Angelina Jolie Effekt* OR Angelina-Jolie-Effekt* OR Angelina-Effekt* OR Angelina-Gen* OR BRCA OR vererbbar* Brustkrebs OR Lynch Syndrom* OR Lynch-Syndrom* OR familiär* adenomatöse* Polyposis OR vererbbar* Darmkrebs OR vererbbar* Krebsveranlagung* OR CRISPR OR Genomeditierung OR Genom* editier* OR Genom-Editierung* OR Präimplantationsdiagnostik OR Biobank* OR (selten* Krankheit* AND (genet* OR Gendefekt* OR gentechni*)) OR personalisiert* Medizin OR personalisiert* Gesundheit OR genomisch* Medizin)

(Angelina Jolie* and (Mastektomie or Eierstöcke or Eierstock)) not ((Angelina Jolie* AND (genet* OR BRCA*)) OR BRCA1 OR BRCA2 OR Angelina Jolie Effekt* OR Angelina-Jolie-Effekt* OR Angelina-Effekt* OR Angelina-Gen* OR BRCA)

2 Algorithm used in Factiva; for LexisNexis the truncation sign (*) was replaced by (!), the truncation sign for the LexisNexis search engine.

Third round

(Pränataldiagnostik or ((pränatal or vorgeburtlich*) and genet*) or NIPT) not Präimplantationsdiagnostik

French sampling

First round

test* génétique* OR maladie* génétique* OR effet* Angelina Jolie OR l'effet Angelina Jolie OR BRCA OR cancer* du sein hérédita* OR syndrome de Lynch OR polypos* adénomateu* familia* OR cancer* du côlon hérédita* OR cancer* colorecta* hérédita* OR predisposition* hérédita* au cancer* OR CRISPR OR édition* génomique* OR edition* du génome OR diagnostic génétique préimplantatoire OR diagnostic préimplantatoire OR DGP OR biobanque* OR (maladie* rare* AND (génom* OR génét*)) OR médecine personnalisée OR santé personnalisée OR médecine génomique

Second round

(Angelina Jolie* AND (génét* OR génom* OR BRCA*) OR BRCA*) OR (enquête* génétique* OR LAGH OR analyse génétique humain) OR (séquençage* génom* or séquençage* ADN) OR (thérapie* génique* OR modification* génom* OR maladie* orpheline* OR maladie* génétique* orpheline*) (Angelina Jolie* AND (génét* OR génom* OR BRCA*) OR BRCA*) OR (Big data AND (génét* OR génom*)) NOT (Angelina Jolie* AND (génét* OR génom* OR BRCA*) OR BRCA*) NOT (test* génétique* OR maladie* génétique* OR effet* Angelina Jolie OR l'effet Angelina Jolie OR BRCA OR cancer* du sein hérédita* OR syndrome de Lynch OR polypos* adénomateu* familia* OR cancer* du côlon hérédita* OR cancer* colorecta* hérédita* OR predisposition* hérédita* au cancer* OR CRISPR OR édition* génomique* OR edition* du génome OR diagnostic génétique préimplantatoire OR diagnostic préimplantatoire OR DGP OR biobanque* OR (maladie* rare* AND (génom* OR génét*)) OR médecine personnalisée OR santé personnalisée OR médecine génomique)

(Angelina Jolie* and (mastectomie or ovaire*)) not (Angelina Jolie* AND (génét* OR génom* OR BRCA*) OR BRCA* OR effet* Angelina Jolie OR l'effet Angelina Jolie OR BRCA)

Third round

(diagnostic prénatal* or (prénatal* and génét*) or NIPT) not (diagnostic préimplantatoire or diagnostic génétique préimplantatoire)

B-2 Codebook

Identification

Name of coder (codername)

1 Bettina

2 Fabian

Date of registration – day (reg_day)

Date of registration – month (reg_month)

Date of registration – year (reg_year)

Article number (articlenumber)
State number given to the article (4-digit number)

Article title (title)
Copy-paste the title of the article

Keyword(s) (keyword)
Separate by ";" if several in the same article

Author of the article (author)

Newspaper columns (column)

0 not applicable

1 Title/front / editorial

2 Politics / Switzerland / national

3 Feuilleton / Style / Literature / Media / Art / nonfiction / Education / Society / culture

4 Economics / Business / Finances / Investment / Money

5 Science / Technique

6 Opinion / reader's letters

7 Local news

8 Other (Sports / Cars / Travel / Others)

9 Magazine

10 News

11 Analyse (TA) / Hintergrund / Temps fort

12 International

13 People

14 Health

99 not specified

Length of article

No of words in the article (words)

Word count incl. titles, subtitles and textboxes, but not subtext of pictures. If a wordcount is already given (which is the case for articles sampled from LexisNexis and Factiva), please use your own word count, since those are overestimating the real article length.

Date of publication

Publication date day (pub_day): DD

Publication date month (pub_month): MM

Publication date year (pub_year): YYYY

Medium

Language (lang)

1	German	4	Italian
2	French	5	Other
3	English		

Type of medium (medium-type)

1	Boulevard	7	Magazine health
2	Cost-free press	8	News agency
3	Daily press	9	Other
5	Sunday press	999	Unclear or unknown
6	Magazine general		

Name of medium (medium-name)

1x	**Boulevard**	**6x**	**Magazines**
11	Blick	62	Beobachter
12	Le Matin	7x	Health-related magazines
2x	**Free press**	71	KTipp/Gesundheitstipp
21	20 Minuten	**8x**	**Agency**
23	20 minutes	81	SDA de
3x	**Daily press**	82	SDA fr
31	Tages-Anzeiger	999	Unclear/unknown
32	Neue Zürcher Zeitung		
37	Tribune de Genève		
38	Le temps		

Topic of article

Main focus of the article (focus1-3)

Concerning genetics, what was the main focus of the article? If several foci apply to an article, they are graded the following: (1) Mentioned in the title; (2) Mentioned in subtitle/lead; (3) Mentioned in the first paragraph; (4) Which focus got more space in the article (more word counts accounted to that topic). If (4) applies, the passages accounted to the respective focus topic will be marked with a color, and the words of these marked parts will be counted (use word count in Word). Please safe the marked document in a separate file!

Note: The categories here are deductive; but if there is a topic not present in the list below, please state the topic briefly in the open-text variable ("other"). They are used in another round of coding, when these new categories will be synthesized and embedded into the already existing context.

Color code: brown = excluded. Orange = main topic. Yellow = open category, please specify. Blue = added after first round of coding. Green = added after second round of coding.

Code	Category	Explanation
0000	**EXCLUDED**	STOP CODING
0100	**Genetic diseases in general**	Articles about genetic diseases, not further differentiable in subsequent categories
0101	Hereditary breast and ovarian cancer / BRCA	If genetic testing for HBOC (i.e. BRCA), please state it here and NOT in section 3
0102	Hereditary colorectal cancer / Lynch syndrome / FAP	If genetic testing for hCRC (i.e.Lynch), please state it here, NOT in sect. 3
0103	Other hereditary cancer	Please specify
0104	None-hereditary cancer	Heritance not specifically mentioned (e.g. breast cancer in general)
0105	Metabolic disorders	
0106	Huntington's disease	
0107	Connective tissue disorders	
0108	Alzheimer's disease	
0109	Movement disorder	Dystrophy, ataxia, dystonia
0110	Mental disabilities	
0111	Albinism	
0112	Osteogenesis imperfecta	
0113	Eye diseases	Retinoblastoma, retinopathy…
0114	Thalassemia	
0115	X-linked disorders	
0116	Chromosomal disorders	i.e. Down syndrome
0117	Viral diseases	Prion, HIV
0118	Economic factors	i.e. insurance coverage
0199	Other syndromes	despite rare diseases (please specify)
0200	**Rare diseases in general**	Articles about rare diseases, not further differentiable
0201	Case story of one rare disease	
0202	Genetic testing of rare diseases	
0203	Insurance coverage information	Problems, coverage etc.
0204	Background information on rare diseases in general	Numbers, statistics

Code	Category	Explanation
0205	Donations, financial support	Of research and affected families
0206	Economic factors/industry	
0207	Sensibilization of the public	"Konzept seltene Krankheiten" of the government, events…
0208	Research	New findings, use rare diseases as study example…
0299	Other (please specify)	
0300	**Genetic testing in general**	Articles about genetic testing, not further differentiable
0301	Present possible applications of genetic testing on born humans	Please state what applications (for testing before birth, please check section 5, for basic research section 11)
0302	Future possible applications of genetic testing on born humans	Please state what applications; also for new research findings not implemented in clinic yet (for testing before birth, please check section 5, for basic research section 11)
0303	Direct-to-costumer genetic testing for born humans	online, in pharmacies, freely available, private use, not via MDs; Lifestyle genetic testing
0304	Economic factors of genetic testing for born humans	i.e. profit, test costs – public health
0305	Insurance coverage of genetic testing in general	
0306	---	
0307	Ethical issues of genetic testing for born humans	e.g. used by insurances
0308	GUMG revision	GUMG = Gesetz für Genetische Untersuchungen am Menschen (political discussion)
0309	General information about genetic testing for born humans	About procedures to get a test, or one specific test etc…
0310	Public opinion on genetic testing for born humans	
0311	Research for genetic testing for born humans	Applied! For basic research see section 11
0350	Ancestry	Paternity test, identify or find relatives… (for medical cases, see 0301)
0351	Genetic testing in animals and food	Also plants included; for GMO see section 0607
0352	Forensics	Crime, identify dead people
0353	Evolution	e.g. Ötzi, population-genetics analysis of very old genetic material
0399	Other (please specify)	Other subtheme of genetic testing; please specify
0400	**Personalized medicine in general**	Articles about personalized medicine, not further differentiable in subsequent categories
0401	Background information on what personalized medicine means	Definition, impact
0402	Genetic testing for personalized medicine	Role of genetic testing in personalized medicine
0403	Personalized medicine for cancer treatment	

Code	Category	Explanation
0404	---	
0405	Applications/treatments of PM	Other than cancer treatment
0406	Challenges and ethical aspects of PM	
0407	Economics related to PM	Pharma/biotech industry
0408	---	
0409	Research/science for PM	Funding, topics, findings
0410	Social implications of PM	Impact on society
0411	Swiss Personalized Health Network initiative	Choose this if this initiative is specifically stated
0499	Personalized medicine for other implications	please specify
0500	**Preimplementation genetic diagnostics (PGD) in general**	Articles about PGD, not further differentiable in subsequent categories. In section 5, all articles about testing/treatment of the unborn human beings are collected
0501	PGD: Vote-related: first vote (June 2015)	Directly referring to the vote
0502	PGD: Vote-related: referendum	Directly referring to the referendum
0503	PGD: Vote-related: second vote (June 2016)	Directly referring to the vote
0504	Technical information on PGD	Explanation: What is PGD; more than one sentence!
0505	Swiss political discussions/decisions on PGD (not directly vote-related)	e.g. discussion before vote was set, or details after discussion
0506	PGD applications	i.e. genetic testing of embryos
0507	PGD case story	
0508	PGD legislation in other countries	Also comparison in-between the countries
0509	Ethical considerations of PGD	
0510	Discussion on health insurance coverage on PGD	
0551	Prenatal genetic testing	Other than PGD
0552	Genetic reproductive medicine	Not prenatal testing; other applications
0553	Reproductive medicine in general	Non-genetic
0599	Other (please specify)	
0600	**Genome editing / CRISPR in general**	Articles about genome editing, not further differentiable
0601	Emergence/presence of the possibility to do genome editing	Informative
0602	Explanation of methods for genome editing	In detail, e.g. with metaphors (“gene scissor”)
0603	Use of CRISPR on human embryo (Chinese team; UK)	Related to the event that Chinese researchers manipulated the genome of a human embryo; or to the UK allowing
0604	Implications ethical of genome editing in general	Mentioning any ethical issues

Code	Category	Explanation
0605	Implications medical / technical of genome editing in general	Mentioning medical/technical implications (not method explanation!)
0606	Crispr inventors/researchers	Info on personalities, prices, who invented (patent dispute)
0607	Applications of genome editing in plants/food; GMOs	
0608	Economic factors of Crispr	Economy, income due to Crispr
0609	Research on genome editing	
0699	Other (please specify)	
0700	**Angelina Jolie in connection to genetics**	(article about Angelina Jolie in connection to genetics, not more specified)
0701	Information about Angelina's decision / letter / statement in general	Event that Angelina Jolie published a statement that she had a mutated BRCA, mastectomy, hysterectomy
0702	Information on mastectomy and/ or hysterectomy in the context of Angelina's history	mentioned in the same context (can also be implicit, e.g. date close to a statement)
0703	Presence of the Angelina-Jolie-Effect	e.g. higher test rates due to the announcements
0704	Discussion about Angelina's decision (ethical / opinion / evaluation)	e.g. reader's opinion
0799	Other (please specify)	
0800	**Biobank in general**	
0801	Emergence of biobanks	i.e. new biobank funded
0802	Use of biobanks	What are biobanks here for?
0803	Implications of biobanks (ethically, technically, medically)	
0899	Other (please specify)	
0900	**Completely different topic (please specify)**	
1001	Data sharing	Both individual data and research data
1002	Data protection	
1003	Digital health	Wearables, electronic medical records etc.
1101	Economics of genetics	i.e. gene patenting – research/theory-based (not mainly application based)
1102	Education in genetics	Educating medical students, or public events, etc….
1103	Philosophy/ethics of genetics	
1201	Stem cell transplantation	
1202	Gene therapy applications	
1301	Bioinformatics for sequencing	e.g. variant identification
1302	Next generation sequencing	In research, technical (applied see 311)
9999	Unclear or unknown	

Other focus of article (focus99)

See list above, if there are more than three foci present in the article, please state here with open text which they are (leave blanc if not applicable) or specify with open text all other topics mentioned in the article.

B-3 Supplementary Figures

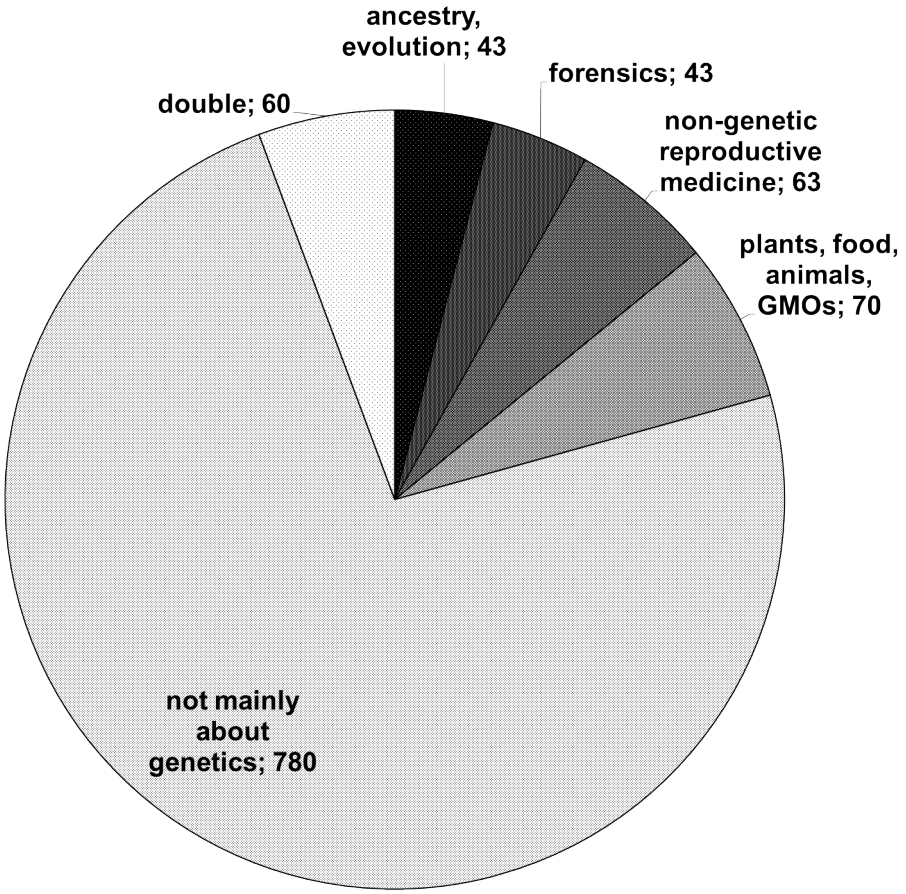

Figure B-1: Categories excluded from analysis.
Numbers after semicolon indicate the number of articles assigned to these categories.

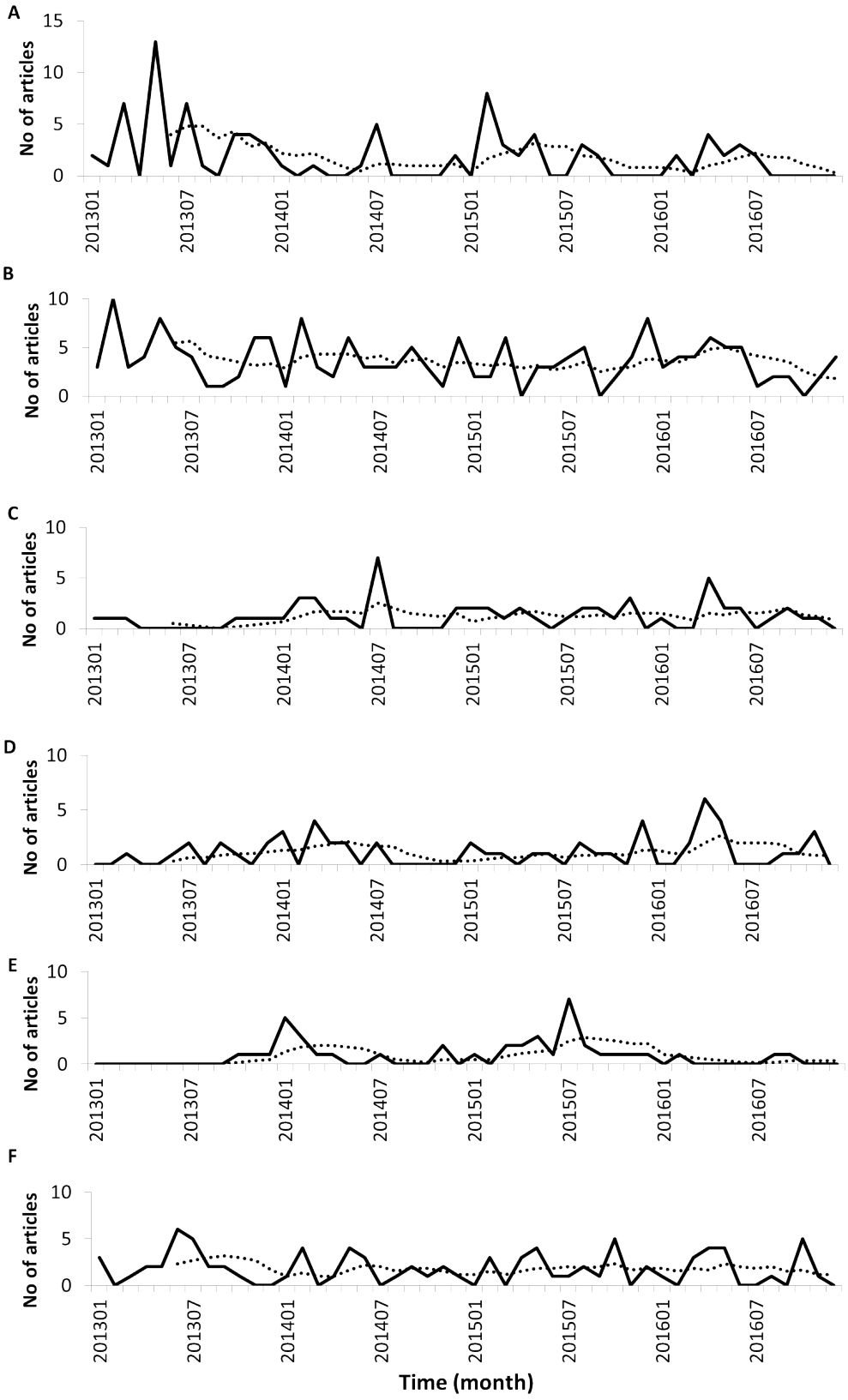

Figure B-2: Development of topics over time.
The presented data was normalized using the Article Count Ratio (ACR), meaning the ratio between the number of articles of a topic at one time point (one month) divided by the total amount of articles of the topic. A) All topics; B) Genetic testing of born humans (GTBH); C) Human Genetic Diseases (HGD); D) Personalized Medicine (PM); E) Big Data in medical genetics & health (BD); F) Gene Therapy (GTh); G) Genetic Research (GRe).

Appendix C: Newspaper coverage of predictive genetic testing in Switzerland and the UK

C-1 Search algorithms

German

((*Angelina* or *Jolie*) AND (genet* OR Gen OR BRCA* OR Mastektomie or Eierstöcke or Eierstock OR Effekt*)) * OR BRCA* OR Gentest* OR genetisch* Test* OR genetisch* Test OR Lynch-Syndrom* OR Lynch Syndrom* OR ((personalisiert* Medizin OR personalisiert* Gesundheit) AND genet*) OR ((((prädiktiv* OR präventiv*) AND (genet* OR genomi* OR genome)) OR genet* Veranlagung* OR Prädisposition) AND *test*)

English

((*Angelina* or *Jolie*) AND (genet* OR gene OR effect* OR BRCA* OR mastectomy or ovar*)) OR BRCA* OR gene* test* OR Lynch syndrom* OR ((personalised medicine OR personalised health) AND (genet* or genom*)) OR ((((predictive OR preventive) AND (genet* OR genom*)) OR genetic* predispos*) AND test*)

C-2 Codebook

Inclusion and exclusion criteria of articles

Inclusion criteria

The topic predictive genetic or genomic testing on born humans OR its consequences (i.e. preventive surgery) OR its social/economical/political implications is the main focus or at least one of the key points in the article (key point meaning that at least 1/5 of article is focusing on PGT).

- Including direct-to-consumer genetic testing when implicitly or explicitly about predictive genetic testing (assumed is mostly the case)
- Including gene patenting, if about predictive tests and their consequences
- Including genetic screening, when predictive (but not screening of embryos or unborn babies)

Exclusion criteria

Nothing to do with predictive genetic or genomic testing, especially:

1. Prenatal/preimplantation genetic testing
2. Genetic testing for diagnostic purposes (diagnose a certain disease, identify cancer sub-type etc.)
3. Genetic testing for treatment purposes (i.e. pharmacogenetic testing)
4. Genetic testing on animals, plants
5. Ancestry search/prove
6. Evolutionary genetics (yeti etc.)
7. Forensic purposes (gene tests for crime, identification of bodies...)
8. keywords used in a completely different context (e.g. Film tips)
9. Gene patenting
10. Very short articles (<100 words in main text). Cave: short announcements on front page are included, if the main article is included as well (merge them into one article)
11. Epigenetic tests

Bibliography

Gerhards, Jürgen; Schäfer, Mike S. (2007): Die Herstellung einer öffentlichen Hegemonie: Humangenomforschung in der deutschen und der US-amerikanischen Presse: Springer-Verlag.

Gerhards, Juergen; Schäfer, Mike S. (2009): Two normative models of science in the public sphere. human genome sequencing in German and US mass media. In Public Understanding of Science 18 (4), pp. 437–451. DOI: 10.1177/0963662507082891.

O'Mahony, Patrick; Schäfer, Mike Steffen (2005): The 'Book of Life' in the Press Comparing German and Irish Media Discourse on Human Genome Research. In Social Studies of Science 35 (1), pp. 99–130. DOI: 10.1177/0306312705046542.

Schäfer, Mike S. (2009): From public understanding to public engagement. An empirical assessment of changes in science coverage. In Science Communication 30 (4), pp. 475–505.

0 Copy-pasted data

Preliminarily collected during sampling process, excluded from pretest

0-1 Article title (title)

Copy-paste the title of the article

0-2 Keyword(s) (keyword)

Separate by ";" if several in the same article

0-3 No of words in the article (words)

Count the number of words by highlighting title, subtitles, article text, textboxes and subtext of pictures and click "Word Count" under the "Review" category in Microsoft Word. Please note: Even if a word count is already given by the database Factiva, use your own word count, since those are overestimating the real article length.

0-4 Publication date

DD MM YYYY

0-5 Name of medium where article was published

00	Unclear/unknown
11	Daily Telegraph
12	The Guardian
31	Tages-Anzeiger
32	Neue Zürcher Zeitung
99	Other

0-6 Author

State author of the article

0-7 Newspaper columns (column)

Zeitungsrubrik

0	not applicable
1	Title/front / editorial
2	Politics / Switzerland & the world / national
3	Feuilleton / Style / Literature / Media / Art / nonfiction / Education / Society / culture
4	Economics / Business / Finances / Investment / Money
5	Science / Technique
6	Opinion / reader's letters
7	Local news
8	Other (Sports / Cars / Travel / Others)
9	Magazine
10	News
11	Analyse (TA) / Hintergrund / Temps fort
12	International
13	People
14	Health
99	not specified

0-8 Additional front teaser

State if this article had an additional teaser on the front page of the newspaper

0	No
1	Yes

Article ID

State number assigned to the article (4-digit number)

0xxx-3xxx	Switzerland
4xxx	UK

Name of coder

1	Bettina	3	David
2	Fabian	4	Noah

Date of registration

DD MM YYYY

1 Topics covered by article

Table "topics", see below

Concerning genetics, what were the main topic(s) of the article?

The main topic is identified by: (1) mentioned in the title; (2) mentioned in subtitle/lead; (3) mentioned in the first paragraph; (4) which focus gets most space (wordcount) in the article.

Additional topics may be coded only if there are additional key aspects in the article that are also mentioned in title, subtitle, lead or first paragraph AND that take important space (at least 10 full sentences or more than half of the article body).

2 Topic evaluation throughout article

How were the previously identified topic(s) evaluated in the article? Look at all directive statements i.e., risks/benefit assessments regarding the topic throughout the article. Statements must be explicit. For certain topics, PGT in general is evaluated (see Topic list, last column). If the evaluation is uncertain, use evaluations in title, subtitle and lead to make a decision about the overall evaluation.

0	Unclear
1	**Clearly positive / benefits**: only or for the most part positive arguments / benefits
2	**Ambivalent / risks+benefits**: both positive (benefit) and negative (risk) arguments mentioned
3	**Clearly negative / risks**: only or for the most part negative arguments / risks
4	**Neutral**: no arguments stated
9	Not applicable: no topic identified

3 Sources/stakeholders

Can be sources (publications, reviews, studies, other articles, newspapers etc.) or stakeholders (individuals or collectives, i.e. persons, groups, committees, parties, organizations, institutions) that are **cited directly or indirectly** *in the article and that give* **information or opinions** *on the topic. An infinite number of sources/stakeholders can be coded. Journalists are only sources/stakeholders if they explicitly state their opinion (e.g. editorial, comment). If a source/stakeholder is only mentioned in the sense that it is info* ***about*** *it, it is not coded (it did not place its own message in the article).*

Experts that are made explicit later in the text are coded as the actual person/organization (e.g. "Expert" is mentioned in the title, and later in the text "Dr. XY" is quoted, and it is clear from the context that this Dr. is the expert from the title, then they are merged – Dr. XY consequently gets a "dominant" status because mentioned in title). **Please also code secondary sources (e.g. the New York Times, when mentioned that it published Angelina Jolie's statement).**

3.1 Source/stakeholder name

Please state here full name of the stakeholder as it appears in the text (including titles, i.e. Dr., Prof., etc). or name of institutions (full and abbreviation, if applicable); as well as the source's name/affiliation (not applicable if no source attributed).

3.2 Source/stakeholder category

If a person is cited, please code the person as a source (e.g. 21 → Prof. XY, author of research study Z, from University U). If e.g. a publication/study/report is cited code these separately (e.g. 24 → research paper). **If several attributes are connected to one stakeholder (e.g., "die Nationalrätin und Ärztin Yvonne Gilli") code the *first* attribute as category. The only exception is if the content *clearly* states otherwise (e.g., if the whole article is about Ärztin Yvonne Gilli, and the fact that she is a politician is merely an artefact mentioned only once).**

Table sources → see below!

3.3 Is this a secondary source?

A secondary source can be another newspaper (e.g. the New York Times, when mentioned that it published Angelina Jolie's statement); a journal's name where a study that is presented in the article has been published, etc.

For secondary sources, the evaluation is "not applicable"!

0 No

1 Yes

3.4 How predominant was this source/stakeholder in the article

If a stakeholder is coded several times (for several topics) in 1 article, the predominance remains the same!

0 Unclear/unknown

1 **Dominant**: this source/stakeholder had important space (title, lead, picture headlines/citations (blue text), first paragraph or majority of text) or was the only stakeholder mentioned in this article.

2 **Shared**: source/stakeholder took important, visible space, but others were also present

3 **Minor**: this source/stakeholder was merely mentioned on the side (1 sentence or less).

9 Not applicable

3.5 Evaluation(s) of topic(s) by source/stakeholder

How were the previously identified topic(s) evaluated by the source/stakeholder? Look at all directive statements of this source/stakeholder throughout the article. Statements must be explicit. (An evaluation is identified if the text passage answers the question "Is the topic good or bad?")

Example: *"Shortly after its launch, Time magazine named 23andMe's test the best invention of the year, describing it as one of the most crucial medical breakthroughs in decades." (Daily Telegraph, 11.4.2015) → answers the question "Is DTC good? Yes." by the Time magazine. For secondary sources, the evaluation is "not applicable"!*

0 Unclear

1 **Clearly positive / benefits**: only or for the most part positive arguments / benefits

2 **Ambivalent / risks+benefits**: both positive (benefit) and negative (risk) arguments mentioned

3 **Clearly negative / risks**: only or for the most part negative arguments / risks

4 **Neutral**: no arguments stated

9 Not applicable: for secondary sources

4 Occasion [Anlass] of article[3] (occasion)

Here, the occasion of the article is stated (has nothing to do with the topic), which is connected to the release of the article. The occasion can be in the past, the present or the future and could also be something that did not take place / was postponed. If several occasions apply, choose the higher ranked one (1=highest priority, 5=lowest priority). To identify the occasion, please screen the article for announcements which indicate what triggered the occasion of the article. In the case of press releases/conferences this can be quite implicit.
Examples*:*

"Gestern hat die Clubleitung die Resultate im Stade Olympique präsentiert" (TagesAnzeiger, 14.12.2013) → press conference

„Die kalifornische Life Technologies Corporation stellte gestern ein Analysegerät vor, mit dem eine vollständige DNA-Untersuchung für rund 1000 Dollar durchgeführt werden kann." (TagesAnzeiger, 11.1.2012) → press conference

„A four-year Europe-wide trial was announced on Monday" (The Guardian, 2.6.2015) → press release

"Experts said last night" (The Daily Telegraph, 27.6.2014) → press conference

"… research conducted by Bowel Cancer UK and the Royal College of Pathologists and released on Monday…" (The Guardian, 8.8.16) → press release

There is only one occasion per article.

a) If there are several mentioned, take the one that is mentioned in the title, subtitle or first in the article; b) If there are several actions by one stakeholder that led to the release of the article, code the earlier one; c) If a stakeholder A is referring to stakeholder B (e.g., a report is criticizing DTC companies), code the action of stakeholder A (the report: code 2 (if a released report) or 3 (if an internal report))

0 No occasion recognizable / not applicable

1 **Message placed into mass media**: editorials, comments, letter to the editor written by a stakeholder

2 **Mass media directly targeted**: a stakeholder aims to get his message into mass media (press release/conference)

3 **Media agenda setting**: the coverage of another medium (newspaper, TV, news agency, magazine, etc.) triggered the release of this article *(as initiator, please do not code the other medium here, but the stakeholder who originally initiated the coverage in this other medium)*

4 **Mass media NOT directly targeted**: there is an occasion, but it is not stated that media were directly targeted (e.g. paper publications, events, incidents) *(it is possible that no initiator is recognizable for such an occasion)*

5 **Case story / interview**: written by a journalist about a special case/person *(always code 11 as initiator)*

9 Other (please specify in comment section)

5 Initiator [Veranlasser] of occasion (initiator)

Who was the main initiator of the occasion that led to the article?

If there are several initiators, code the more dominant one; if same dominance, code the first that is mentioned.

If a stakeholder initiates the article via a secondary source (e.g. through another newspaper article that is referred to in this article), do NOT code the secondary source but the original stakeholder that placed his/her message in the media.

No occasion → no initiator

Table sources → See below

3 Adapted from Gerhards and Schäfer (2006).

6 Comments of coder (comments)

Please state here any comments, problems or thoughts

Topic list

No	Topic	Description	*Topic to evaluate*
0	Unclear/unknown		
19	Direct-to-consumer genetic testing	Genetic testing performed in a **do-it-yourself** manner -> direct connection between tested person and offering company (Gentests ohne Verschreibung durch Fachperson)	*See description*
20	Medical use of PGT	New PGTs, healthcare procedures, clinical eligibility criteria for PGT (i.e., eligibility for screening)	*See description*
21	Disclosure of disease risks	Doctor-patient relationship (counselling, information etc); right not to know; incidental findings; uncertainty	*See description*
22	Case story of affected person	Examples / cases / statements of people living with a genetic risk, their family → **code case stories of celebrities in Public debates (15)**	*PGT in general*
23	Preventive treatment	Only when in relation to a genetic risk -> preventive surgeries, preventive examination, preventive drugs	*See description*
14	Public health	implementation of genetic testing into healthcare (e.g. by NHS); general guidelines regarding PGT (i.e., screening, clinical use, preventive treatment, etc.), "genetic revolution", personalized medicine, genetics in society	*See description*
15	Public debates about PGT	Political discussions, public debates, going public (excl. Angelina Jolie), case story of a celebrity	*See description*
17	Legal and economic aspects	Actual law changes, patenting, public health economics, insurance coverage/prices of PGT	*See description*
16	Angelina Jolie case	Everything directly related to Angelina Jolie's statement, effects, etc.	*See description*
13	Research about predictive genetic testing	Any research study, new findings, research has shown → including clinical trials! → regarding genetic data for research, please code 12	*See description*
18	Ethical issues	Discrimination, stigmatization, privacy, data protection, data sharing, confidentiality issues	*See description*
11	Information (facts) about predictive genetic testing	Background information about predictive **genetic testing in general**	*PGT in general*
12	Genetic data / WGS	Big Data / looking at the whole genome instead of certain genes (Whole Genome analysis) / data sharing platforms	*See description*
99	Other (please specify)		
999	Article not included – stop coding!		

Source/stakeholder list

0	No source attributed	Facts without explicit information about source
10	**Media (rest)**	*Please state in the comment section*
11	Journalist / person writing the article	Stating clear personal opinion; including letter to the editor
12	Other journalist	
14	News agency	*e.g. SDA, DPA, PA, Reuters, AP: if news agency is stated in brackets below the article, its predominance is considered unclear (0)*
15	Other medium	e.g. TV, article in the same newspaper or in a different newspaper is cited
20	**Research/medicine (rest)**	*Please state in the comment section* *Code general term "experts" here!*
21	Natural / medical science	Person / research group / research organisation: Natural scientist, not treating patients directly but have a role as a researcher *Cave: could be an MD! But the way the person is presented in the text it's a research function and not a patient treating function*
22	Social science / art & humanities / law	Person, research group or research organisation - Bioethicist / philosopher / theologist …
23	Medical doctor / geneticist / nurse	Person: Treats patients, working as an MD is specifically mentioned Organisation: hospital, medical institutes
24	Research study / article / journal	Source: i.e. a paper cited
25	University	Source: On an institutional level
30	**Economics/company (rest)**	*Please state in the comment section*
31	DTC company	Source or person – if products are sold to consumers directly
32	Pharma company	Source or person – if not a DTC company
33	Biotech company	Source or person – if not a DTC company
40	**Society/individual (rest)**	*Please state in the comment section*
41	Affected patient	Person with a positive genetic risk for a certain disease (e.g. cancer) → *cave: can be healthy, but has an increased risk to get a serious illness!*
42	Healthy genetic test user	A healthy person using a predictive genetic test
43	Family of affected patient or healthy genetic test user	Family members of an affected person
44	VIP / celebrity	e.g. VIP/celebrity that makes genetic predisposition and/ or preventive treatment public (Angelina Jolie, Kelly Osborne, Betty Ford etc.)
50	**Public institutions / health (rest)**	*Please state in the comment section*
51	Political system / politicians / political commission national	Statements from political system in general, parliament, politicians etc. (can be persons or organizations!), courts, Health Department

52	Political system / politicians / political commission international	Statements from political system in general, parliament, politicians etc. (can be persons or organizations!), courts, Health Department
53	Governmental organisation	Any institution financed by government, e.g. Swissmedic,SNF, FDA, NIH, NHS, National Institute for Health and Care Excellence (Nice), counseling inst. (i.e. Medical Research Council) → check online 53 vs. 55
54	Health insurance	Coverage of predictive genetic test / policies of health insurances; citation of a health insurance's statement
55	Non-governmental organizations	Not financed by or relying on government, e.g. Patient organizations / NGOs / charity / civil movement / professional bodies / foundations …
90	**Other**	*Please state in the comment section*
91	Church / religious group	

C-3 Supplementary Table

Table C-1: Further characteristics of clusters in addition to Table 7-2.

	Cluster	Public debates		Economization		Medical use & treatment		Research		Personal stories	Total	
	Country	UK	CH	UK	CH	UK	CH	UK	CH	UK	UK	CH
Variable (var)		% of each variable										
Evaluation of topic by stakeholder	Positive / benefits	58.3	44.8	40.0	12.9	40.6	24.1	34.7	31.5	36.4	41.1	29.0
	Ambivalent	3.3	10.3	15.8	19.4	17.4	27.6	4.2	17.8	19.3	12.8	18.5
	Negative / risks	11.7	17.2	21.1	38.7	20.3	34.5	4.2	30.1	20.5	16.1	30.2
	Neutral / facts	26.7	27.6	23.2	29.0	21.7	13.8	56.9	20.5	23.9	29.9	22.2
Influence of stakeholders	No occasion recognizable / not applicable			17.8		5.1	3.4	2.2	18.1	8.0	7.1	8.5
	Message placed into mass media (i.e., comments, editorials)	37.2	15.9	14.9	6.1	29.5	3.4	2.2	1.2	29.5	21.9	5.8
	Mass media directly targeted (i.e., press release, conference)	9.0	15.9	19.8		2.6	27.6	12.4	9.6	2.3	9.7	12.2
	Media Agenda Setting (MAS)3	39.7	43.2	1.0	42.4	25.6	27.6		4.8	14.8	15.0	23.8
	Mass media NOT directly targeted	14.1	15.9	32.7	45.5	33.3	27.6	83.1	60.2	21.6	37.6	42.3
	Personal stories / interview		9.1	13.9	6.1	3.8	10.3		6.0	23.9	8.8	7.4

Table C-1 continued.

	Cluster	Public debates		Economization		Medical use & treatment		Research		Personal stories	Total	
	Country	**UK**	**CH**	**UK**	**CH**	**UK**	**CH**	**UK**	**CH**	**UK**	**UK**	**CH**
Variable (var)		**% of each variable**										
Article format	Notice (<200 words)	6.4	18.2	5.0		7.7		18.0	6.0	4.5	8.3	6.9
	News (200-800 words)	47.4	36.4	46.5	57.6	42.3	41.4	73.0	19.3	37.5	49.5	33.3
	Reports (>800 words)	46.2	45.5	48.5	42.4	50.0	58.6	9.0	74.7	58.0	42.2	59.8

Notes: All numbers are percentages except the column "No of cases". The sum of each column within one variable is 100%. Numbers in bold indicate a significantly higher occurrence compared to the other clusters of one country than expected by chance. Numbers in italic indicate lower occurrence than by chance. Significance was tested with the Chi-square residual method (Beasley and Schumacker, 1995) comparing cluster for each country separately with Pearson Chi-square, and corrected for multiple testing with the Benjamin Hochberg correction (false discovery rate set to 5%). In the case of evaluation, we also compared statistically significant differences between the two countries within each cluster. UK = United Kingdom, CH = German part of Switzerland.

C-4 Statement list[4]

Which statements is a stakeholder placing in the article? ONLY CODE STATEMENTS THAT ARE CONNECTED TO A STAKEHOLDER MENTIONED BEFORE!

Please note: Statements can occur in the form that is written in the list; or with the opposite meaning. Example: Statement "A preventive surgery has important side effects" would also be coded if a stakeholder says that a preventive surgery has no side effects (then the evaluation code below is negative, meaning no. 4 or 5).

Each statement gets an evaluation how the stakeholder is referring to it in the article.

Positive means that the stakeholder is formulating the statement in the way that is written in the codebook.

Negative means that the stakeholder is formulated in the opposite way.

0 Unclear or neutral (no arguments stated)

1 Normative positive (statement shall be as formulated in list)

2 Descriptive positive (statement is the case as formulated in list)

3 Ambivalent (both shall be/is and shall not be/is not mentioned)

4 Descriptive negative (statement is NOT the case as formulated in list)

5 Normative negative (statement shall NOT be as formulated in list)

1 SWOT (Risks & Benefits, Chances & Threats)

11 Predictive genetic testing - control

1121 A genetic test should not be used okay when there is no preventive treatment known for the disease

1122 A genetic test should not be used when it is not enough scientifically investigated.
Opposite: A genetic test shall also be used even when not completely scientifically investigated

12 Predictive genetic testing - outcome

1211 Genetic testing can calm / relieve / … the tested person
Opposite: A risk of a genetic test result is that it makes the affected person anxious / agitated / uncertain …

1212 Knowing about genetic risks motivates people to live healthier
Opposite: Genetic risks do not motivate people to live healthier

1213 Genetic testing helps people to plan their life better

1214 There are economic advantages of a predictive genetic test result
Opposite: Economic disadvantages (e.g. discrimination by health insurance, employer etc.)

4 Please note: This statement list was not included in the final version of the codebook due to low reliability (see chapter 2.1.2).

1221 There can/will be a societal pressure to perform genetic/genomic analyses
Opposite: There is no (risk of) social pressure

1222 There is a risk of social discrimination / stigmatization
Opposite: There is no social discrimination / stigmatization of persons with a genetic risk

13 Predictive genetic testing - usefulness

1311 Genetic testing can be used for personal prognosis

1312 Genetic testing can be used for early risk assessment / detection of a disease

1313 With genetic testing prevention of disease is possible

1314 With genetic testing therapy/treatment can be improved

1315 Genetic testing can improve performance (sport, diet...)
Opposite: Genetic testing does not improve performance / these tests are not serious

1321 A risk of genetic testing is that it can lead to incidental findings
Opposite: It is a benefit when genetic testing leads to incidental findings

1322 There is a risk to use predictive genetic testing too much / too often
Opposite: Genetic testing should be used more often/widespread - It is beneficial when predictive genetic testing is used often / frequently

14 Direct-to-consumer genetic testing

1411 With DTC users can calm their curiosity; DNA can be fun

1412 With DTC users support research
Opposite: With DTC research is not supported

1413 DTC genetic testing does not lead to more unnecessary preventive treatments
Opposite: DTC genetic testing leads to unnecessary preventive treatments

1414 DTC customers have no problem interpreting their test results
Opposite: DTC genetic test results are not well interpretable by customers and counseling support is bad

1415 With DTC, individuals can take their own responsibility on their health

1416 DTC tests are easy to do

1421 Tests for genes that play a role in diseases should not be sold on the internet

15 Preventive treatment

1511 Due to a preventive treatment, disease risk is lower
Opposite: Preventive treatment does not reduce risk for the genetic disease

1512 Preventive surgery leads to positive feelings (relief / happy / still female / reduced feeling of threat or fear, ...)
Opposite: Preventive surgery leads to negative feelings (regret / sadness / self-mutilation, …)

1513 Preventive treatment prolonges life
Opposite: preventive treatment has no effect on life expectancy

1514 Preventive surgery because of high genetic risk is medically recommended to every affected person
Opposite: Preventive surgery is not the right solution for every person with genetic cancer risk

1521 A preventive treatment has important side effects
Opposite: a preventive treatment does not have important side effects

1522 There is a risk of economic pressure to do a preventive treatment (because it is the cheaper option)
Opposite: there is no economic pressure on patients to do a preventive treatment

16 Big data

1611 Due to big data combinations data protection is in danger
Opposite: Big data does not lead to worse data protection

2 Ethical, legal and social implications (ELSI)

21 Societal aspects - "Genetic revolution"

2111 (In the future,) public health (will be / is) based on big data and personalized medicine, which (will) change medicine deeply

2112 The "genetic revolution" will change how society decides over costs, prevention, data protection, etc.

2113 The "genetic revolution" will change how one specific person decides over his/her options, preventive treatments, tests, etc.

22 Ethical principles

2211 How to deal with genetic risks is a person's own individual decision

2212 What a person wants to know or not about the own genetic risk is a personal decision

2221 Under certain circumstances, patients should be informed about incidental findings even though they did not give permission beforehand

2231 It is important to respect privacy and data protection with regards to genetic test results

2232 Disclosure of test results to family members can be an ethical issue

2233 Data sharing is beneficial

23 Legal aspects

2311 We need / there are stricter regulations for DTC companies

2312 Data protection towards health insurance, employer etc. should be / is secured by law

2313 Genetic analyses and counselling should be / is strictly regulated by law

2314 Law about genetic testing needs actualisation

24 Public health

2411 Healthcare procedures before/during/after genetic testing

2412 Implementation of genetic testing in the clinical practice is challenging

2413 There are not enough facilities for genetic testing

2421 Political discussions about genetics in public health

3 Decision making

31 Decision making genetic testing

3111 Family member Y from person X has / had the disease / the mutation

3112 Family history can help reveal health risks

3121 A genetic test should be performed when a disease is common in a family, and should first be done with an affected person before testing family members

3122 Genetic screening / cascade testing is used on family members once a person is identified with a genetic mutation

32 Decision making preventive treatment

3211 A preventive treatment was performed on person(s) X due to a genetic risk

3212 After a positive predictive genetic test result, a stakeholder recommends to take a certain preventive treatment

4 Preventive treatment

41 preventive treatment due to genetic risk

4112 Routine control examinations are a preventive treatment option for at-risk people

4113 Medication as preventive treatment

4121 In Switzerland, preventive surgeries due to genetic risks are not as common as in other countries (i.e. USA)

4122 A detailed description of preventive surgery procedure

5 Knowledge

51 Explanations / background info

5111 A mutated gene X predispose to disease Y

5112 There is a difference between sporadic and inherited cancer

5113 When a parent has a genetic disease, the children can inherit the mutation

5121 The DTC testing procedure is simple; (you simply send in a saliva sample)

5122 DTC genetic testing companies do not provide medical interpretation

5123 Results/outcomes of DTC genetic testing

5131 Genomic analysis means X / is used for Y / etc.

5141 Person X has an increased genetic risk for disease Y (because of reason Z)

5142 A tested person has to deal with uncertainties
Risk percentage; not sure if one gets the disease really or not

5161 New genetic test identified

52 Knowledge transfer

5211 People should know when a predictive genetic test is useful, (thus public debates are important)

5212 DTC costumers are not enough informed about the test / results / implications

5221 Doctors have to / should / do inform affected people about treatment options and alternatives

54 Public awareness

5411 Due to the Angelina Jolie effect there were more people interested in genetic testing

5412 The Angelina Jolie effect lead to more genetic counselling

5413 The Angelina Jolie effect lead to more preventive treatments

5414 The Angelina Jolie effect lead to more genetic tests

5421 A person X goes public with genetic history to lead by example / raise awareness / encourage affected people

5422 To go public with a preventive surgery due to genetic risk is positive (brave, admirable, exemplary, etc.)

5423 A person makes DNA profile and/or its consequences public
Rather descriptive

6 Research

6111 Genomic/genetic research is complex / challenging, many things are still unknown

6112 For research you need genetic data from a lot of persons, which you can get e.g. by registries, DTC databases, etc.

6113 Researchers are trying to bring together genetic variants with diseases, e.g. by comparing healthy and sick individuals

6114 A research study searched for (a) new genetic variant(s) involved in (a) certain disease(s)

6115 Implementation of research finding into clinical practise

7 Economics

71 Economic aspects of genetic testing

7111 DTC genetic tests do not cost much

7112 Economical aspects of DTC companies

7121 Genetic testing / a genetic/genomic test is too expensive

7122 Costs for genetic/genomic testing are decreasing

7131 The health insurance is covering the genetic/genomic test

7132 The individual is covering costs for genetic/genomic testing himself

7141 Health insurances and genetic test providers should / do collaborate

72 Economic aspects of preventive treatment

7211 Non-invasive preventive treatment is cost-effective

7221 Preventive surgeries are cost-effective

Appendix D: Media Coverage of Ethical Issues in Predictive Genetic Testing

Table D-1: Articles included for analysis.

Title	Newspaper	Author	Date of publication	Length (words)
Family tests for fatal cholesterol disease 'are not being offered'	*Daily Telegraph*	Rebecca Smith	25.01.2011	491
Doctors' failures led to Rianna's early death; A schoolgirl was not given a simple blood test despite warning signs that she had dangerous cholesterol levels – and thousands more adults and children	*Daily Telegraph*	Jane Feinmann	31.01.2011	1277
Genetics tests flawed, say Dutch scientists	*The Guardian*	Ian Sample	31.05.2011	250
NHS braced for DNA patents fight	*Daily Telegraph*	Stephen Adams	06.08.2011	320
Older women at risk of cancer 'should have their ovaries removed'	*Daily Telegraph*	Martin Beckford	06.09.2011	511
Patients' genomes to be stored with medical records: US pilot study hopes to improve drug choices Cost of sequencing now similar to single gene test	*The Guardian*	Ian Sample	29.12.2011	898
Genetic test may predict lifespan, say researchers	*Daily Telegraph*	Auslan Cramb	10.01.2012	274
We can tell if you will live past 100, say scientists; Genetic test could lead to anti–ageing treatments	*Daily Telegraph*	Nick Collins	19.01.2012	522
Sisters have surgery to beat cancer gene that killed mother; Life savers	*Daily Telegraph*		01.02.2012	150
Faulty gene makes prostate cancer four times more likely	*Daily Telegraph*	Rebecca Smith	21.04.2012	378
The charity helping to keep mum; Advances in genetic screening mean that Clare Delaney can enjoy being a parent	*Daily Telegraph*		15.09.2012	697
Family: A cruel inheritance: The Smiths have a family history of Huntington's Disease - an incurable neuro-degenerative disorder that has changed life for them all.	*The Guardian*	Joanna Moorhead	22.09.2012	2018
Sharon Osbourne tells of double mastectomy: Genetic factors raised risk of second bout of cancer 'I want to be around to be a grandmother to Pearl'	*The Guardian*	Lizzy Davies	05.11.2012	523

Table D-1 continued.

Title	Newspaper	Author	Date of publication	Length (words)
Genomics could alter this baby's life, but will the NHS be quick enough to feel the benefit?: DNA maps for all? Not so far off, or as costly, as you thought	*The Guardian*	Sarah Boseley	05.12.2012	2085
Danger gene could help fight disease	*Daily Telegraph*		17.12.2012	181
500,000 to be offered breast cancer drugs: At-risk women should be eligible for preventative treatment, NHS told	*The Guardian*	Denis Campbell	15.01.2013	735
Cancer lifeline for at-risk women; Annual breast screenings and tamoxifen for those with family history of disease, guidance states for first time	*Daily Telegraph*	Rebecca Smith	15.01.2013	467
Blood screening that is preventing heart attacks - but not in England: NHS elsewhere in Britain is tracking down and treating people with inherited condition	*The Guardian*	Denis Campbell	23.01.2013	1371
Breast cancer is still a risky business	*Daily Telegraph*	Max Pemberton	28.01.2013	705
Genetic test could help fight cancer	*Daily Telegraph*		20.03.2013	189
Scientists hail 'leap forward' on cancer tests : Landmark study into genetic causes could herald national screening plan: Saliva test could predict risk of prostate cancer	*The Guardian*	Ian Sample	28.03.2013	1012
Ros Eeles: Analysis 'We can focus on those most in danger'	*The Guardian*	Ros Eeles	28.03.2013	407
Could new cancer tests make you uninsurable?	*Daily Telegraph*	Rosie Murray–West	06.04.2013	1669
Gaby Koppel 'By the time I found out about my risk, I probably had cancer already': Rosie Lord 'My surgeon said I was the youngest patient he'd performed the operation on'	*The Guardian*	Gaby Koppel	15.05.2013	541
'She has challenged not just her own public image but also the wearisome cliche of how a woman considered sexy talks about her body'	*The Guardian*	Hadley Freeman	15.05.2013	1512
Experts hail Jolie for going public on the private affair of her life-saving treatment: Actor raises awareness of test for defective gene Screening detects risk of developing breast cancer	*The Guardian*	Ed Pilkington; James Meikle and Sarah Ridley	15.05.2013	1399
Jolie hailed as inspiration to women, and a heroine to Brad; Actress had double mastectomy after being told faulty gene gave her 87pc risk of cancer	*Daily Telegraph*	Jon Swaine / Tim Walker	15.05.2013	962

Table D-1 continued.

Title	Newspaper	Author	Date of publication	Length (words)
A GIANT LEAP FOR WOMANKIND; By writing about her double mastectomy Angelina Jolie has normalised the idea of lifesaving surgery and flagged up the need to make genetic testing affordable, says Anj	*Daily Telegraph*	Anjana Ahuja	15.05.2013	1485
Jolie 's doctor reveals intimate details of double mastectomy	*The Guardian*	Denis Campbell	16.05.2013	528
Jolie mastectomy was a surprise to me as well, says her actor father	*Daily Telegraph*	Nick Allen	16.05.2013	614
They're gone, but she's staying	*Daily Telegraph*	Allison Pearson	16.05.2013	264
Genetic cancer risk; Letters to the Editor	*Daily Telegraph*	Marcus Woodhouse Wolverhampton	16.05.2013	276
Jolie deserves praise, but why so much surprise?	*The Guardian*		18.05.2013	280
Angelina Jolie: Spare me my mother's fate	*Daily Telegraph*	Judith Woods	18.05.2013	1363
Cancer–risk man removes prostate	*Daily Telegraph*	Victoria Ward	20.05.2013	320
Many common maladies, such [...]; Doctor's Diary	*Daily Telegraph*	James Le Fanu	20.05.2013	236
More women to be offered screening; Gene tests	*Daily Telegraph*		21.05.2013	192
Jolie 's aunt dies of breast cancer aged 61	*The Guardian*	Sam Jones and agency	28.05.2013	376
Angelina Jolie 's aunt dies of breast cancer; World Bulletin	*Daily Telegraph*		28.05.2013	107
I'm so happy to have got everyone talking; Angelina Jolie tells of her 'gratitude' that her double mastectomy has opened up a worldwide debate on women's health	*Daily Telegraph*	Melanie Hall	03.06.2013	615
Breakthrough may ease threat of breast cancer	*Daily Telegraph*	Laura Donnelly	14.06.2013	134
Women at risk from breast cancer to be offered daily pill: New NHS guidelines aim to prevent disease Drug course provides alternative to surgery	*The Guardian*	Sarah Boseley	25.06.2013	724
Breast cancer drugs offered to women at high risk	*Daily Telegraph*	Nick Collins	25.06.2013	450
A £10 test to help prevent baby blues	*Daily Telegraph*	Claire Carter	02.07.2013	627
Catch ovarian cancer before the disease catches you	*Daily Telegraph*	Max Pemberton	08.07.2013	370

Table D-1 continued.

Title	Newspaper	Author	Date of publication	Length (words)
Hiking in memory of a life cut short; Steve Boryszczuk lost his wife to Alzheimer's when she was just 43. Now, he is a man with a mission, says Peter Stanford	*Daily Telegraph*	Peter Stanford	13.07.2013	1407
' Jolie effect ' leads to women asking for mastectomies they don't need	*Daily Telegraph*	Sam Marsden	03.10.2013	598
Clinic offers breast cancer test on Skype; News Bulletin	*Daily Telegraph*		31.10.2013	122
NHS to offer genetic test for heart defects	*Daily Telegraph*		06.12.2013	202
Family: Problem solved: Annalisa Barbieri	*The Guardian*	Annalisa Barbieri	14.12.2013	850
Genetic screening from birth to identify disease risks 'within 40 years'	*Daily Telegraph*	Sarah Knapton	10.01.2014	443
Routine tests for ' Jolie ' gene in ovarian cancer	*Daily Telegraph*		23.01.2014	139
Double mastectomies may save more lives	*Daily Telegraph*		12.02.2014	182
Gene tests for aggressive prostate cancer come a step closer: Screening would identify those at real risk of dying Just 14 mutations could signal need for treatment	*The Guardian*	Sarah Boseley	21.02.2014	498
Gene tests may hold the key to halting deadly Prostate Cancer	*Daily Telegraph*	Sarah Knapton	21.02.2014	558
Genetic testing that may help to increase the nation's IQ	*Daily Telegraph*		24.03.2014	199
Family: How much do you want to know?: If you could manipulate your child's future - would you? Stuart Jeffries meets geneticist Dr Sharon Moalem	*The Guardian*	Stuart Jeffries	05.04.2014	1795
Half a million 'healthy' adults at risk from heart disorders	*Daily Telegraph*		12.05.2014	210
Women offered hope of a blood test that can predict the risk of breast cancer years ahead	*Daily Telegraph*	Laura Donnelly	27.06.2014	486
Gene tests could help target the men at risk of prostate cancer	*Daily Telegraph*	Rebecca Smith	15.09.2014	218
Should it be Angelina 's job to educate us about health?	*Daily Telegraph*	Max Pemberton	22.09.2014	736
'The trouble is, breast cancer treatment is aimed at women'	*Daily Telegraph*	Cherrill Hicks	10.11.2014	1490
Bowel cancer risk	*Daily Telegraph*		13.11.2014	254

Table D-1 continued.

Title	Newspaper	Author	Date of publication	Length (words)
The revolution that's transforming medicine	*Daily Telegraph*	TOM CHIVERS	23.12.2014	956
Why genome project is well worth pursuing	*The Guardian*	Dr Marc Tischkowitz	06.01.2015	303
Should you worry about your genes?	*Daily Telegraph*	Tanith Carey	07.03.2015	1367
Angelina Jolie and ovarian cancer: the facts about screening and surgery	*The Guardian*	Hannah Devlin	24.03.2015	577
Angelina Jolie says the decision to deal with her cancer was simple. Mine is not	*The Guardian*	Fay Schopen	24.03.2015	843
Angelina Jolie reveals she had ovaries removed after cancer scare	*The Guardian*	Melissa Davey	24.03.2015	817
My children won't have to say 'Mom died of ovarian cancer'	*Daily Telegraph*	Angelina Jolie Pitt	25.03.2015	1097
'I had a ticking time bomb inside me': four women who faced Angelina Jolie 's choice	*The Guardian*	Paula Cocozza	26.03.2015	2255
Dangers of the ' Angelina effect '	*Daily Telegraph*	Max Pemberton	30.03.2015	283
NHS in Wales won't perform life-saving tests	*Daily Telegraph*		04.04.2015	330
'After watching my father lose his memory, I had to know my fate'	*Daily Telegraph*	Josie Ensor	11.04.2015	1201
Breast cancer gene testing set to become cheaper and easier	*The Guardian*	Jessica Glenza	22.04.2015	553
Doctors aim for single test to detect risk of four cancers in women	*The Guardian*	Joanna Moorhead	02.06.2015	602
' Jolie gene ' test could save 2,000 a year from cancer	*Daily Telegraph*		03.06.2015	181
Genetic tests at four could spot trouble ahead	*Daily Telegraph*	Javier Espinoza	18.06.2015	305
Why a double mastectomy before the age of 30 can be a laughing matter	*The Guardian*	Olivia Hirst	13.08.2015	1009
Goodstock at Edinburgh festival review – touching look at the genetic lottery of cancer	*The Guardian*	Lyn Gardner	27.08.2015	329
Serious flu risk could be identified with genetic test	*The Guardian*	Ian Sample	08.09.2015	666
DNA-screening test 23andMe launches in UK after US ban	*The Guardian*	Samuel Gibbs	02.10.2015	930
Can knowing you and your family may get Alzheimer's ever be positive?	*The Guardian*	Giulia Rhodes	02.11.2015	1016
DNA- testing kit 23andme: patient-powered healthcare or just confusing?	*The Guardian*	Ann Robinson	12.01.2016	1022
Faulty gene can triple risk of developing ovarian cancer	*Daily Telegraph*	Sarah Knapton	19.01.2016	317

Table D-1 continued.

Title	Newspaper	Author	Date of publication	Length (words)
Cancer prevention	*Daily Telegraph*		27.01.2016	190
New test 'detects genes for every known inherited heart condition'	*The Guardian*	Haroon Siddique	19.02.2016	580
Why I gave up my job to go travelling after a double mastectomy	*The Guardian*	Claira Hermet	17.03.2016	852
I should come with a letter of warning ...'	*Daily Telegraph*	Siddhartha Mukherjee	21.05.2016	2704
Are DIY gene -testing kits a good idea??	*The Guardian*	Sharon Brennan	13.06.2016	1133
Breast cancer cell growth halted by osteoporosis drug, study shows	*The Guardian*	Ian Sample	20.06.2016	494
Cancer breakthrough for women with the ' Jolie gene '	*Daily Telegraph*	Sarah Knapton	21.06.2016	124
Signs of dementia 'can start at age of three'	*Daily Telegraph*	Sarah Knapton	14.07.2016	505
Bowel cancer: 29% of UK hospitals not following guidance on testing younger patients	*The Guardian*	Kate Lyons	08.08.2016	487
Test identifies heart risk in young people	*Daily Telegraph*		22.09.2016	118
Cholesterol tests for children could prevent 600 heart attacks a year	*Daily Telegraph*		27.10.2016	217
' Angelina Jolie effect ' boosted genetic testing rates, study suggests	*The Guardian*	Nicola Davis	15.12.2016	690
Jolie effect saw sharp rise in cancer gene tests	*Daily Telegraph*		15.12.2016	113
' Jolie effect ' leading to needless mastectomies	*Daily Telegraph*		30.12.2016	523

Table D-2: Deductive coding list with references.

Underlying principle	Ethical issue	Comments, keywords	References
Autonomy	Right to know	Access to predictive genetic information (links to beneficence)	Lolkema et al. 2013
Autonomy	Right not to know	(links to nonmaleficence); Sharing predictive genetic information with others ; confidentiality; privacy	Andorno 2004
Autonomy	Competence: Understanding predictive genetic information	Interpretation of test results; informed consent; genetic counselling; decision making	Marteau and Croyle 1998; Harris, Winship, and Spriggs 2005; Lolkema et al. 2013
Autonomy	Determination and authenticity	Genetic information can lead to reduced autonomy	Huibers and van `t Spijker 1998

Table D-2 continued.

Underlying principle	Ethical issue	Comments, keywords	References
Autonomy	(Perceived) lack of alternativity	Wish to avoid cognitive dissonance, feeling of having no choice	Huibers and van `t Spijker 1998
Autonomy	Voluntariness of decision making	Cut short by medical staff or relatives	Huibers and van `t Spijker 1998
Beneficence	Psychological benefit	Relief, taking action/ control	Lolkema et al. 2013
Beneficence	Medical benefit	Prevention, treatment	Lolkema et al. 2013
Beneficence	Duty to warn patients		Fulda and Lykens 2006; Rothstein 2018; Elger, Bernice, Michaud, and Mangin 2010
Beneficence	Reproductive benefit		Wertz, Fletcher, and Berg, K. 2003; Yarborough, Scott, and Dixon 1989; Fulda and Lykens 2006
Beneficence	Utility	Balancing risks, benefits and costs	Evans, Skrzynia, and Burke 2001
Beneficence	Genetic research progress		Mathaiyan, Chandrasekaran, and Davis 2013
Nonmaleficence	Uncertainty of disease development		Evans, Skrzynia, and Burke 2001
Nonmaleficence	Psychological harm	Anxiety, stigma; information about non-preventable/non-treatable diseases; suicide	Wertz and Fletcher 1991; Markel 1992; Fulda and Lykens 2006; Hamilton, Lobel, and Moyer 2009; Wiggins et al. 1992
Nonmaleficence	Medical harm	Unnecessary medical interventions	Evans, Skrzynia, and Burke 2001
Nonmaleficence	False reassurance of negative test result	Also autonomy (only occurs if people misunderstood information)	Jackson, Goldsmith, and Skirton 2014
Justice	Discrimination / Stigma	Employers, health/life insurance, mortgages	Harris, Winship, and Spriggs 2005
Justice	Costs and funding	Distributive justice	American Society of Clinical Oncology 2003
Justice	Access to predictive genetic information	Equality	Ormond 2008
Justice	Gene patenting	Inequity of access	Harris, Winship, and Spriggs 2005

Table D-2 continued.

Underlying principle	Ethical issue	Comments, keywords	References
Cross-cutting	Information to family members about genetic risks	Cascade testing	Fulda and Lykens 2006; Lucassen and Parker 2010; Rothstein 2018; Elger, Bernice, Michaud, and Mangin 2010; Haupt 2018; Brassington 2011
Cross-cutting	Predictive genetic testing of children		Harris, Winship, and Spriggs 2005; Mand et al. 2012; Caulfield et al. 2015; Millum 2014; Elger, Bernice S. 2010
Cross-cutting	Predictive genetic testing without medical indication/relevance	curiosity, DTC	Vayena, E. et al. 2012; Su, Howard, and Borry 2011; Hofmann 2016; Vayena, Effy 2015
Cross-cutting	Incidental findings	Clinical or research	Roche and Berg, J. 2015; Lockhart et al. 2018; Hofmann 2016

References

American Society of Clinical Oncology. 2003. "American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility." Journal of clinical oncology : official journal of the American Society of Clinical Oncology 21 (12): 2397–2406. doi:10.1200/JCO.2003.03.189.

Andorno, Roberto. 2004. "The Right Not to Know: An Autonomy Based Approach." J Med Ethics 30 (5): 435-9; discussion 439-40. doi:10.1136/jme.2002.001578.

Brassington, Iain. 2011. "Is There a Duty to Remain in Ignorance?" Theoretical medicine and bioethics 32 (2): 101–15. doi:10.1007/s11017-010-9145-x.

Caulfield, Timothy, Pascal Borry, Maeghan Toews, Bernice S. Elger, Henry T. Greely, and Amy McGuire. 2015. "Marginally Scientific? Genetic Testing of Children and Adolescents for Lifestyle and Health Promotion." Journal of Law and the Biosciences 2 (3): 627–44. doi:10.1093/jlb/lsv038.

Elger, Bernice, Katarzyna Michaud, and Patrice Mangin. 2010. "When Information Can Save Lives: The Duty to Warn Relatives About Sudden Cardiac Death and Environmental Risks." Hastings Center Report 40 (3): 39–45. doi:10.1353/hcr.0.0254.

Elger, Bernice S. 2010. "Ethical, Legal and Social Issues in the Genetic Testing of Minors." In Handbook of Genomics and the Family: Psychosocial Context for Children and Adolescents, edited by Kenneth P. Tercyak, 485–521. Boston, MA: Springer US. https://doi.org/10.1007/978-1-4419-5800-6_20.

Evans, James P., Cécile Skrzynia, and Wylie Burke. 2001. "The Complexities of Predictive Genetic Testing." BMJ 322 (7293): 1052–56.

Fulda, Kimberly G., and Kristine Lykens. 2006. "Ethical Issues in Predictive Genetic Testing: A Public Health Perspective." Journal of Medical Ethics 32 (3): 143–47. doi:10.1136/jme.2004.010272.

Hamilton, Jada G., Marci Lobel, and Anne Moyer. 2009. "Emotional Distress Following Genetic Testing for Hereditary Breast and Ovarian Cancer: A Meta-Analytic Review." Health psychology : official journal of the Division of Health Psychology, American Psychological Association 28 (4): 510–18. doi:10.1037/a0014778.

Harris, Marion, Ingrid Winship, and Merle Spriggs. 2005. "Controversies and Ethical Issues in Cancer-Genetics Clinics." The Lancet Oncology 6 (5): 301–10. doi:10.1016/S1470-2045(05)70166-2.

Haupt, Laura. 2018. "Rescue via Genetic Findings." Hastings Cent Rep 48 (3): 2. doi:10.1002/hast.844.

Hofmann, Bjørn. 2016. "Incidental Findings of Uncertain Significance: To Know or Not to Know—That Is Not the Question." BMC Medical Ethics 17:13. doi:10.1186/s12910-016-0096-2.

Huibers, Alex K., and Adriaan van 't Spijker. 1998. "The Autonomy Paradox: Predictive Genetic Testing and Autonomy: Three Essential Problems." Patient education and counseling 35 (1): 53–62. doi:10.1016/S0738-3991(98)00083-4.

Jackson, Leigh, Lesley Goldsmith, and Heather Skirton. 2014. "Guidance for Patients Considering Direct-to-Consumer Genetic Testing and Health Professionals Involved in Their Care: Development of a Practical Decision Tool." Family practice 31 (3): 341–48. doi:10.1093/fampra/cmt087.

Lockhart, Nicole C., Carol J. Weil, Latarsha J. Carithers, Susan E. Koester, A. R. Little, Simona Volpi, Helen M. Moore, and Benjamin E. Berkman. 2018. "Development of a Consensus Approach for Return of Pathology Incidental Findings in the Genotype-Tissue Expression (GTEx) Project." J Med Ethics 44 (9): 643–45. doi:10.1136/medethics-2017-104691.

Lolkema, Martijn P., Christa G. Gadellaa-van Hooijdonk, Annelien L. Bredenoord, Peter Kapitein, Nancy Roach, Edwin Cuppen, Nine V. Knoers, and Emile E. Voest. 2013. "Ethical, Legal, and Counseling Challenges Surrounding the Return of Genetic Results in Oncology." Journal of clinical oncology : official journal of the American Society of Clinical Oncology 31 (15): 1842–48. doi:10.1200/JCO.2012.45.2789.

Lucassen, Anneke, and Michael Parker. 2010. "Confidentiality and Sharing Genetic Information with Relatives." The Lancet 375 (9725): 1507–9. doi:10.1016/S0140-6736(10)60173-0.

Mand, Cara, Lynn Gillam, Martin B. Delatycki, and Rony E. Duncan. 2012. "Predictive Genetic Testing in Minors for Late-Onset Conditions: A Chronological and Analytical Review of the Ethical Arguments." Journal of Medical Ethics 38 (9): 519–24. doi:10.1136/medethics-2011-100055.

Markel, H. 1992. "The Stigma of Disease: Implications of Genetic Screening." The American journal of medicine 93 (2): 209–15.

Marteau, Theresa M., and Robert T. Croyle. 1998. "Psychological Responses to Genetic Testing." BMJ 316 (7132): 693–96.

Mathaiyan, Jayanthi, Adithan Chandrasekaran, and Sanish Davis. 2013. "Ethics of Genomic Research." Perspectives in clinical research 4 (1): 100–104. doi:10.4103/2229-3485.106405.

Millum, Joseph. 2014. "The Foundation of the Child's Right to an Open Future." Journal of social philosophy 45 (4): 522–38. doi:10.1111/josp.12076.

Ormond, Kelly E. 2008. "Medical Ethics for the Genome World: A Paper from the 2007 William Beaumont Hospital Symposium on Molecular Pathology." J Mol Diagn 10 (5): 377–82. doi:10.2353/jmoldx.2008.070162.

Roche, Myra I., and Jonathan S. Berg. 2015. "Incidental Findings with Genomic Testing: Implications for Genetic Counseling Practice." Current genetic medicine reports 3 (4): 166–76. doi:10.1007/s40142-015-0075-9.

Rothstein, Mark A. 2018. "Reconsidering the Duty to Warn Genetically at-Risk Relatives." Genet Med 20:285 EP -. doi:10.1038/gim.2017.257.

Su, Yeyang, Heidi C. Howard, and Pascal Borry. 2011. "Users' Motivations to Purchase Direct-to-Consumer Genome-Wide Testing: An Exploratory Study of Personal Stories." Journal of Community Genetics 2 (3): 135–46. doi:10.1007/s12687-011-0048-y.

Vayena, E., E. Gourna, J. Streuli, E. Hafen, and B. Prainsack. 2012. "Experiences of Early Users of Direct-to-Consumer Genomics in Switzerland: An Exploratory Study." Public Health Genomics 15 (6): 352–62. doi:10.1159/000343792.

Vayena, Effy. 2015. "Direct-to-Consumer Genomics on the Scales of Autonomy." J Med Ethics 41 (4): 310–14. doi:10.1136/medethics-2014-102026.

Wertz, D. C., and J. C. Fletcher. 1991. "Privacy and Disclosure in Medical Genetics Examined in an Ethics of Care." Bioethics 5 (3): 212–32. doi:10.1111/j.1467-8519.1991.tb00161.x.

Wertz, D. C., J. C. Fletcher, and K. Berg. 2003. "Review of Ethical Issues in Medical Genetics: Report of Consultants to WHO." http://www.who.int/genomics/publications/en/ethical_issuesin_medgenetics%20report.pdf.

Wiggins, S., P. Whyte, M. Huggins, S. Adam, J. Theilmann, M. Bloch, S. B. Sheps, M. T. Schechter, and M. R. Hayden. 1992. "The Psychological Consequences of Predictive Testing for Huntington's Disease. Canadian Collaborative Study of Predictive Testing." New England Journal of Medicine 327 (20): 1401–5. doi:10.1056/NEJM199211123272001.

Yarborough, Mark, Joan A. Scott, and Linda K. Dixon. 1989. "The Role of Beneficence in Clinical Genetics: Non-Directive Counseling Reconsidered." Theor Med Bioeth 10 (2): 139–49. doi:10.1007/BF00539879.

Curriculum Vitae Bettina Zimmermann

Personal information	
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Academic positions	
Since 04.2021	Postdoctoral fellow at the Institute for Biomedical Ethics (IBMB), University of Basel (Switzerland). Project: "Public debates and citizens' motives facing restrictions during the COVID-19 pandemic: a mixed-methods study".
Since 05.2020	Postdoctoral fellow at the Institute of History and Ethics in Medicine, Technische Universität München, Germany. Leader: Prof. Alena Buyx. Research projects: (1) "Ethical issues in social media-based recruitment for the TherVacB clinical trial"; (2) "Solidarity in times of a pandemic (SolPan)".
04.2020 - 03.2021	Postdoctoral fellow at the Institute for Biomedical Ethics (IBMB), University of Basel (Switzerland). Project: "A Fair Reimbursement System for Orphan Drugs (FROND)", funded by a Käthe Zingg-Schwichtenberg seed grant.
Education	
07.2016 – 03.2020	**PhD in Bioethics** (summa cum laude), University of Basel, Institute for Biomedical Ethics, Faculty of Science Title of PhD: "Informed Decision-Making for Genetic Testing: Public Discourse and Individual Choice." (self-designed research project) Supervisors: Prof. Bernice Elger, Dr David Shaw, Prof. Henriette Meyer zu Schwabedissen, Prof. Steffen Kolb (HTW Berlin), Prof. Barbara Prainsack (University of Vienna, external expert)
09.2014 – 06.2016	**MSc Molecular Medicine** (English, 120 ECTS), Uppsala University, Uppsala, Sweden. Master thesis at Basel University, Department of Biomedicine, Prof. Primo Schär. "Triggering DNA methylation changes in colorectal cancer cell line upon *BRAF* mutation induction."
09.2011 – 06.2014	**BSc Biomedical Sciences** (German and French, 180 ECTS), University of Fribourg, Switzerland.
09.2008 – 06.2011	**BA Media & Communication Sciences** (German, 90 ECTS), Contemporary History (60 ECTS), Business Economics (30 ECTS). University of Fribourg, Switzerland.
Research publications (peer reviewed journals)	
2021	**Zimmermann BM**, Fiske A, McLennan S, Sierawska A, Hangel N, Buyx A (2021): Motivations and Limits for COVID-19 Policy Compliance in Germany and Switzerland. *International Journal of Health Policy and Management*, in press.
2021	**Zimmermann BM**, Fiske A, Prainsack B, Hangel N, McLennan S, Buyx A. (2021): Early Perceptions of COVID-19 Contact Tracing Apps in German-Speaking Countries: Comparative Mixed Methods Study. *Journal of Medical Internet Research* 23(2):e25525. doi: 10.2196/25525

2021	**Zimmermann BM**, Fanderl J, Koné I, Rabaglio M, Bürki N, Shaw D, Elger B (2021): Examining information-seeking behavior in genetic testing for cancer predisposition: A qualitative interview study. *Patient Education and Counseling* 104(2):257-264. doi: 10.1016/j.pec.2020.09.019
2021	**Zimmermann BM**, Koné I, Shaw D, Elger B (2021): Autonomy and social influence in predictive genetic testing decision-making: A qualitative interview study. *Bioethics* 35(2):199-206. doi: 10.1111/bioe.12816
2020	**Zimmermann BM**, Starke G, Shaw D, Elger B, Koné I (2020): Actionability and scope should determine the extent of counselling for presymptomatic genetic testing. *Swiss Medical Weekly* 150:w20274. doi: 10.4414/smw.2020.20274
2020	**Zimmermann BM**, Shaw D, Heinimann K, Knabben L, Elger B, Koné I (2020): How the "control-fate continuum" helps explain the genetic testing decision-making process: a grounded theory study. *European Journal of Human Genetics* 28(8):1010-1019. doi: 10.1038/s41431-020-0602-3
2020	Kollbrunner L, Rost M, Koné I, **Zimmermann BM,** Padrutt Y, Wangmo T, Elger B (2020): Acute and transitional care or rehabilitation? Retrospective analysis of discharge planning from a municipal hospital in Switzerland. *BMC Health Serv Res* 20(1). doi: 10.1186/s12913-020-05547-1.
2019	**Zimmermann BM**, Kolb S, Zimmermann F, Elger B, Shaw D (2019): Influence of content, events and culture on the public discourse about medical genetics in Switzerland – A quantitative media content analysis. *Communication & Medicine* 16(1): 92-106. doi: 10.1558/cam.34832
2019	**Zimmermann BM**, Elger B and Shaw D (2019): Media coverage of ethical issues in predictive genetic testing: A qualitative analysis. AJOB Empirical Bioethics 10(4):250-264. doi: 10.1080/23294515.2019.1670275
2019	**Zimmermann BM**, Aebi N, Kolb S, et al. (2019): Content, evaluations and influences in newspaper coverage of predictive genetic testing: A comparative media content analysis from the United Kingdom and Switzerland. Public understanding of science 28(3): 256–274. doi: 10.1177/0963662518816014
2019	**Zimmermann BM**, Koné I, Rost M, et al. (2019): Factors associated with post-acute discharge location after hospital stay: a cross-sectional study from a Swiss hospital. BMC health services research 19(1): 289. doi: 10.1186/s12913-019-4101-6
2018	Koné I, **Zimmermann BM**, Nordström K, Elger BS and Wangmo T (2018): A scoping review of empirical evidence on the impacts of the DRG introduction in Germany and Switzerland. The International journal of health planning and management. doi: 10.1002/hpm.2669
2018	Koné I, **Zimmermann BM**, Wangmo T, Richner S, Weber M and Elger B (2018): Hospital discharge of patients with ongoing care needs: The current situation using data from a city hospital under SwissDRG. Swiss Medical Weekly, 2018 Jan 29;148:w14575. doi: 10.4414/smw.2018.14575.
2017	Farnsworth B, Radomska KJ, **Zimmermann BM**, Kettunen P, Jazin E and Emilsson LS (2017): mRNA levels are upregulated in schizophrenia and predict expression. Brain Research, 1669, 63-68. doi: 10.1016/j.brainres.2017.05.027
2016	Farnsworth B, Peuckert C, **Zimmermann BM**, Jazin E, Kettunen P and Emilsson LS (2016): Gene Expression of Quaking in Sporadic Alzheimer's Disease Patients is Both Upregulated and Related to Expression Levels of Genes Involved in Amyloid Plaque and Neurofibrillary Tangle Formation. Journal of Alzheimer's Disease, 53(1), 209–219. doi: 10.3233/JAD-160160

Research conferences with contributions	
06.2020	**Zimmermann, B.**, Shaw D, Heinimann K, Knabben L, Elger B, Koné I (2020) Poster: How the "control-fate continuum" helps explain the genetic testing deci-sion-making process: a grounded theory study. European Society for Human Genetics (ESHG) conference 2.0, life from your living room (online conference).
09.2019	**Zimmermann, B.**, Elger, B., Shaw, D. (presented by Bettina Zimmermann). Talk: "To what extent should the public be informed about genetic and genomic testing possibilities?" European Association of Centres of Medical Ethics (EACME) conference, Oxford, UK.
06.2019	**Zimmermann, B.,** Elger, B., Shaw, D. (presented by Bettina Zimmermann). Poster: "Most issues covered but not in their full complexity: A newspaper content analysis of ethical issues in predictive genetic testing." European Society for Human Genetics (ESHG) conferences, Gothenburg, Sweden.
06.2018	**Zimmermann, B.**, Kolb, S., Aebi, N., Elger, B., Shaw, D. (presented by Bettina Zimmermann). Poster: "Who has a say in mass media coverage of predictive genetic testing? A comparative media content analysis in the UK and Switzerland." European Society for Human Genetics (ESHG) conferences, Milan, Italy.
09.2017	**Zimmermann, B.** Talk: "Genetik in den Schweizer Printmedien". ScienceComm (Swiss National Science Communication Conference), Solothurn, Switzerland.
06.2017	**Zimmermann, B.**, Kolb, S., Shaw, D., Elger, B. Talk: "Perception of genetics among the public: a content analysis of Swiss media". Communication, Medicine and Ethics (COMET) conference, Indianapolis, USA.
05.2017	**Zimmermann, B.,** Kolb, S., Shaw, D., Elger, B. Talk: "Genetic Testing in Public Discourse". SCOPES (Scientific co-operation between Eastern Europe and Switzerland) conference, Chisinau, Moldowa.
Grants	
12.2020	CHF 79'677.00 Novartis Universität Basel Excellence Scholarships for Life Sciences. Universität Basel Forschungsfonds, Switzerland. Project title: 'Public debates and citizens' motives facing restrictions during the Covid-19 pandemic: a mixed-methods study', main applicant. 2021-2022.
06.2020	€152'645.00 Global Health Research in the Wake of the Sars-CoV-2 Outbreak Grant. Federal Ministry of Education and Research, Germany. Project title: 'SolPan – Solidarity in the time of a Pandemic? A longitudinal, international, comparative study to assess and evaluate behavior', co-applicant. 2020-2022.
11.2019	CHF 48'690.00 Swiss Academy of Medical Sciences, Käthe Zingg-Schwichtenberg Seed Grant for Junior Researchers. Project title: 'Fair Reimbursement System for Orphan Drugs (FROND)', main applicant. 2020-2021.
09.2019	CHF 7'235.00 Personal grant from Freie Akademische Gesellschaft (FAG) Basel to finalize the dissertation, main applicant. 2020.
07.2017-06.2019	CHF 2'867.00 Travel grants for research conferences, University of Basel

Awards and scholarships	
06.2019	European Conference for Human Genetics, Gothenburg 2019: Best Poster Award.
09.2012-today	Scholar of the Swiss Study Foundation, since 2021 alumna. CHF 7'235.00
07.2007	Kantonsschule Wil (SG): Award for the best graduation grade (5.4*)
Teaching	
2021	Digital Literacy online class for medical students (TU Munich).
2019	Julia Fanderl, internship (MSc Epidemiology): individual teaching of methodological skills: thematic analysis of interviews (University of Basel).
2018	Jana Möller, internship (BSc Biology): individual teaching of methodological skills: qualitative media content analysis (University of Basel).
2017-2021	Yearly tutorial for pharmacy and biology students: "Ethical issues in genome editing". University of Basel. Bachelor level (University of Basel).
2017-2021	Yearly tutorial for medical students: "Ethical decision-making concerning assisted suicide". University of Basel. Bachelor level (University of Basel).
01.2018-06.2018	Noah Aebi, Co-supervision of master thesis in Pharmaceutical Sciences: "How are gene therapies portrayed to the public? A qualitative content analysis of newspaper coverage on somatic gene therapies in the United States, Germany and Switzerland" with Prof. H. Meyer zu Schwabedissen, University of Basel.
02.2018-06.2018	Organization of the seminar "Contemporary Debates in Bioethics: Ethical Issues in Genetics" (Master and doctoral level), Institute for Biomedical Ethics, University of Basel.
Outreach activities	
07.09.19	Invited as speaker at the public event "rendez-vous science", organized by reatch and The Swiss academies of arts and sciences. https://www.rendez-vous-science.ch/
03.05.18	Podium: **Zimmermann, B.,** Ienca, M., Kraemer, P. nanoTalks Basel: Reading brains, reading choices? Basel, Switzerland. https://www.reatch.ch/de/content/nanotalks-basel-reading-brains-reading-choices
05.04.18	Workshop: **Zimmermann, B.,** Jegmirat, J. The Genetic Oracle: Prevention at all Costs? Basel, Switzerland. https://www.reatch.ch/de/content/rrh-genetic-oracle-prevention-all-costs
14.03.18	Workshop: **Zimmermann, B**., Wittwer, J., Gruppendiskussion: Humangenetische Beratung. Bern, Switzerland. https://www.reatch.ch/de/content/gruppendiskussion-humangenetische-beratung
24.10.17	Workshop: **Zimmermann, B.,** Tschäni, M., Weber, P., Schuler, A., Köhler, C. Genetik im Alltag: Top oder Flop? Basel, Switzerland. https://www.reatch.ch/de/content/rueckblick-genetik-im-alltag-top-oder-flop
25.09.17	Blog article: **Zimmermann, B.** Moral enhancement: illegal brain doping or ethical duty? reatch blog. https://www.reatch.ch/de/content/moral-enhancement-illegal-brain-doping-or-ethical-duty

Other skills and qualifications	
Languages	German: Mother Tongue English: Proficient User (C2), local language study in 10.2006 (Bournemouth, UK) French: Advanced User (C1), local language study in 08.2010 (Antibes, FR) Swedish: Basic knowledge (B1), local language study 9.2014-10.2015 (Uppsala, SE) Italian: Basic knowledge (A2)
Research methods	Quantitative content analysis Qualitative content analysis (thematic analysis, Mayring's summarizing content analysis) Experience with personal interviews (patients and experts) Grounded theory Scoping review of scientific literature Statistics: descriptive statistics, regression analysis, hypothesis testing
EDP	Excellent knowledge in MS office programs (Word, Excel, Powerpoint) Profound knowledge of analytical software for quantitative and qualitative social science research (SPSS and MaxQDA)
Other	Policy and science communication: Organization of public science events and policy paper writing in collaboration with the think tank reatch (www.reatch.ch) Leadership skills: Since Mai 2017 responsible of reatch Basel 2011-2014 Trainer and coach of two junior volleyball teams 2003-2014: Organization of summer camps and other events for children and adolescents