Background Health Literacy has become an important issue area during the past decades, both in the academic and public dialogue. At the same time, new possibilities in the emerging field of personal genomics and the interpretation of health-related genetic information create seemingly extraordinary benefits and - for some - underestimated risks and disadvantages of epic proportions. The main purpose of this exploratory research is to systematically summarise and discuss the most fundamental scientific literature on health literacy and personal genomics, to illustrate the most important frameworks, academic mindsets and driving developments in both clinical and commercial direct-to-consumer genetic testing and finally, to conduct an empirical study associated with the constructed hypothesis, asking for the attitude towards genetic testing and the willingness to undergo such a procedure of individuals with either well-marked health literacy or a lack of such skills. In order to accomplish two things at once this thesis is dedicated to serve as an educational overview for all interested parties, both academic and non-professional.

Results Empirically aggregated qualitative data suggests a wide array of varying attitudes and opinions towards health-related, preventive genetic testing, ranging from highly enthusiastic and curiosity-driven affirmation to a more critical and cautious mindset. A first quantitative cluster analysis of the aggregated data shows a quite affirmative outlook in relation to health-related application of personal genomics. However, methodological shortcomings in the study design and possibly biased survey results suggest that further research is needed.

Please note that this thesis should be thought of and read as a primer, presenting the main concepts and developments behind health literacy and personal genomics, discussing the most important issues related to both topics and analysing the empirical data aggregated during the scientific process. At the same time it however does raise no claim to completeness on a topic as controversial and complex as the existing

“To learn to read is to light a fire; every syllable that is spelled out is a spark.”

Victor Hugo

(Les Misérables)

INTRODUCTION

The beginning of the new millennium has been marked by a pioneering scientific breakthrough. While standing on the shoulders of giants like Gregor Johann Mendel, William Bateson,
Rosalind Elsie Franklin, James Watson and Francis Crick[1], Frederick Sanger, Mary-Claire King and many others, a collaborative group of international researchers brought "the largest single undertaking in the history of biological science"[2] to an end: the Human Genome Project. With the introduction of the Harvard Personal Genome Project[3] in 2005 and the launch of the personal genomics and biotechnology company 23andMe in 2006 the concept of genetic testing became known to a wider audience. Since then, the medical world is confronted with a wide range of technological and societal developments ushering an often-quoted revolution in healthcare.

The emerging field of personal genomics, described as "the branch of genomics concerned with the sequencing and analysis of the genome of an individual"[4], yields new approaches to preventive healthcare and patient participation. It aims to provide individuals with information about their probability ("risk") of developing certain diseases in the future by analysing their DNA. In addition to that, personal genomics and the associated research efforts attempt to discover potential relationships between the genotype and the phenotype in different individuals.[5] Direct-to-Consumer genetic testing has become a lifestyle product for many, interested in their ancestry and the genetic basis of their own physical traits.[6] Society is faced with enormous challenges in order to fully anticipate future developments and understand the possible aftermaths. In the light of current events this research examines the role of the individual, its understanding of the topic and the imperative necessity of such an understanding.

Everything starts with literacy.

The United Nations Educational, Scientific and Cultural Organization (UNESCO) defined Literacy as an implicit part of the right to education in accordance with the 1948 Universal Declaration of Human Rights.[7] The UNESCO Education for All Global Monitoring Report[8] "Literacy for life" from 2006 presents four understandings of literacy: literacy as reading, writing and oral skills, literacy as a learning process, literacy as applied, practised and situated and literacy as text. Numeracy[9] is described as "the ability to process, interpret and communicate numerical, quantitative, spatial, statistical and even mathematical information in ways that are appropriate for a variety of contexts"[10] and a broader definition of literacy is introduced with the discussion about skills that are enabling access to knowledge and information.

"Literacy is the ability to identify, understand, interpret, create, communicate and compute, using printed and written materials associated with varying contexts. Literacy involves a continuum of learning in enabling individuals to achieve their goals, to develop their knowledge and potential, and to participate fully in their community and wider society."[11]

Although the ability to read a text or a symbol, to write and calculate are the most known and used skills associated with the term literacy, the concept itself is composed of a plethora of sub-categories, such as scientific literacy, media literacy or political literacy to name but a few. The work in hand focusses on the concept of health literacy as "the ability to obtain, read, understand and use healthcare information to make appropriate health decisions and follow instructions for treatment".[12] It is therefore limited to the field of health-related forms of literacy.

This thesis should be thought of and read as a primer, presenting the main concepts and developments behind health literacy and personal genomics, discussing the most important issues related to both topics and analysing the empirical data aggregated during the scientific process. At the same time it however does raise no claim to completeness on a topic as controversial as the existing.
“Education does not result from reading but from reflecting on what we read.”

Carl Hilty

RESEARCH AIMS AND OBJECTIVES

The main purpose of this exploratory research is to systematically summarise and discuss the most fundamental scientific literature on health literacy and personal genomics and to conduct an empirical study associated with the constructed hypothesis.[13] In order to accomplish two things at once the research is dedicated to serve as an educational overview for all interested parties, both academic and non-professional.

Generally speaking the work in hand consists of two different parts. The first one is the theoretical part in the form of a literature review on the most important issues. The second part is mainly empirical and looks for a way to answer the main question derived from the hypothesis.

Right at the beginning the basic concepts of health literacy and personal genomics are introduced and explained in detail. In so doing the gentle reader becomes familiar with the topic and aware of the possible legal, social and economic implications that will emerge during the further procedure. In a second step the most important international[14] and national legal frameworks and regulations regarding genetic testing will be presented, while keeping a focus on a comprehensible description and a deepening understanding of the mentioned implications. In a final step the paper elaborates on the clinical utility of genetic testing and discusses the emergence and impact of projects and commercial services in the field of personal genomics. Ultimately the empirical study and the main results will be presented and discussed. Although this empirical data is being introduced at a later point of the thesis, the gentle reader will be able to grasp carefully selected excerpts of it as she reads through it: following almost every chapter a concluding comment by a survey participant will be quoted. In most cases the quotes have been selected based on the topic of the preceding chapter and they serve the specific purpose of a pause of reflection.

Even though the gentle reader might not be familiar with the concepts that are going to be introduced later on, we will try to anticipate the main thoughts behind the empirical study conducted for this thesis without anticipating the results[15] and key findings so far.

The empirical study was conducted by employing an online survey. During the time period of three months a total of 559 study participants were recruited to answer the questions of the survey. The main task of the survey was to find a way to answer the question whether a health-literate person – where “health-literate” was put on almost the same level as health-conscious – would be more interested in using a genetic test in order to find out more about his or her current and future health state compared to an individual who lack a specific state of health literacy.

HYPOTHESIS

“Individuals with well-marked health literacy would consider genetic testing more often than individuals with lacking a certain state of health literacy.”

The individual state of health literacy of the study participants was scrutinized by asking questions that were considered to be crucial factors when living a healthy and health-conscious
life.[16] In a second step study participants were asked for their personal attitude towards genetic testing and their own willingness to undergo such a procedure.

Survey participants were recruited by using Social Media, especially through the social media network Twitter. Due to the fact that potential candidates were invited to take the survey based on specific catchwords (hashtags) they would use on their social media profiles, the results of the study might be prone to a specific bias. This issue – and others like the adverse events of convenience sampling – will be addressed in the chapter “limitations of the study”. For reasons of better comprehension the exact methodology used when conducting the empirical study will be described in a separate chapter later in this paper. Due to the complexity of the issue the possible limitations of the the study will be discussed at a later time.

"The more you know [.] the better you can improve yourself"
Survey Participant - ID 407

KEY CONCEPTS

Whether in everyday life or in the academic world, terminology turns out to be key. In order to be able to talk, read, write and - more generally - learn about the world and our society we use words. Without the word for apple humans could not possibly talk or write about a very commonplace fruit we all recognise. In the same way the word god enables us to create a whole variety of possible concepts about something that seems to be so far beyond our cognitive abilities. Even though an apple is apparently something very concrete and god, or rather the conceptions of god, arguably something very abstract, most people use these words without thinking too much. It is therefore vital for the understanding of this research to write about key concepts first.

3.1 THE CONCEPT OF HEALTH LITERACY

An apple a day keeps the doctor away. A mnemonic this simple and commonly used when referring to maintaining a healthy lifestyle contains almost everything a person needs to know about the way health literacy works. It provides us with simple guidance on what to do – in this case to eat healthy – and informs us about the positive effects of such a measure, namely to stay healthy and avoid illness. As a matter of course the concept of health literacy itself consists of far more than a simple dietary advice.

One of the most elaborated definitions of health literacy has been established by Don Nutbeam in 2000. Based on the fact, that health literacy is ought to “represent the cognitive and social skills which determine the motivation and ability of individuals to gain access to, understand and use information in ways which promote and maintain good health”[17], Nutbeam proposes an advanced classification. For that reason he differentiates the concept of health literacy into three subsumable concepts. The first one, basic or functional literacy, is “characterized by sufficient basic skills in reading and writing to be able to function effectively in everyday situations.”[18]

Communicative or interactive literacy consists of “more advanced cognitive and literacy skills which, together with social skills, can be used to actively participate in everyday activities, to extract information and derive meaning from different forms of communication, and to apply..."
information to changing circumstances.”[19] The third term Nutbeam introduces is called critical literacy, which involves the idea of “critically analys[ing] information, and [to] use[ing] this information to exert greater control over life events and situations.”[20]

Evidently the definition and meaning of health literacy can be expanded to a wide sphere of action. It is of crucial importance whether these concepts are applied on a basic level, for instance in the course of education of an under-age person, or on a higher level, where the person or group concerned is thought of having an advanced ability to reason. To instruct a child on how to promote his or her own health and to accomplish the same with a grown person depicts two quite opposite ends on how to approach this task. In the same way it is arguably impossible to use the same strategy when trying to implement a desired behaviour - for example patient compliance - on an ill person compared to dealing with a consumer who is in good health and tries to be well in the future. In order to illustrate these distinctions a broader definition of the concept of health literacy becomes necessary. With this in mind we take a closer look at the social determinants of health in what follows.

The social determinants of health constitute a comprehensive description of the possible conditions “in which people are born, grow, work, live, and age, and the wider set of forces and systems shaping the conditions of daily life”. [22] As shown in the picture on the preceding page, these conditions are divided in individual lifestyle factors, social and community networks, living and working conditions and the general socio-economic, cultural and environmental conditions. At the heart we see the core determinants, which are age, sex and constitutional factors. The social determinants of health are often used as a universal model, especially when topics like health promotion and health inequality are brought up for discussion.

The consideration of the social determinants of health in this chapter can be seen as an essential part of our understanding of the concept of health literacy. It is of major importance whether a person and/or patient lives in a highly developed country, with sound socio-economic and environmental conditions and a reliable social network or whether a person is faced with changing circumstances in a hostile geopolitical environment. The question, whether a person has been provided with a good (basic) education is just one of many factors that may have influence on her ability to control her own state of health. Work environment, (un-)employment, (non-)existing health care services, housing, a good sanitation and other factors take on an important role.
Therefore the concept of health literacy emerges from a multilayered interplay of various skills and abilities on an individual level and the demands and the complexity of modern societies. A more detailed consideration of the influencing factors will be discussed subsequently.

One of the most sophisticated conceptual models of health literacy has been established during the implementation of the European Health Literacy Survey (HLS-EU). This model introduced a broader definition of health literacy, where the field of health care, disease prevention and health promotion were considered the three main fields of application (see figure below). Every individual makes contact with one of the three main domains during his or her life course and hence faces four different modes of dealing with health relevant information. These four modes are described as the ability to access/obtain information relevant to health, to understand information relevant to health, to appraise/judge/evaluate information relevant to health and finally to apply/use this information.

On the left side of the conceptual model it can be seen that the social determinants (of health), both personal and situational, were included in order to present a comprehensive picture of the far reaching concept of health literacy. Since there would be no practical benefit in delving into the results of the European Health Literacy Survey for the purpose of this thesis, we will – as was intended – focus on health literacy in association with the field of genetic testing. With this in mind we turn over a new leaf and have a closer look on the genesis of personal genomics.

"I make choices to live as healthy and responsibly as I can, and am very aware and listen to signs my body gives me, a genetic profile could help me tailor this more specifically to me. However, it makes me think of breast cancer, and people removing their breasts to avoid getting it. There is potential to try to fix it rather than truly care for ourselves."

Survey Participant - ID 394
3.2 THE GENESIS OF PERSONAL GENOMICS

Gregor Mendel, who conducted his research on pea plants in the 19th century, has become the father of modern genetics. The results from his research is common knowledge today and essential part of the national curriculum worldwide. In the mid-twentieth century two now world-famous scientist, namely James Watson and Francis Crick, pushed the envelope of modern genetics even further. By establishing an scientifically accurate model of DNA and its very structure they opened a new chapter in the history of science. The "double-helix", composed of the four primary bases Adenine, Cytosine, Thymine and Guanine was considered to represent the blueprint of every lifeform on earth, including the human body. It took another 50 years and the effort of an international scientific research project - the Human Genome Project - to identify the sequence of the basepairs and map the genes of the human genome for the first time. Ever since then the health-related analysis of the genetic profile of individuals was considered within reach.

The Human Genome Project was estimated to cost a total of 2.7 billion dollar over a period of almost 15 years (1990 – 2005) and was publicly funded. Soon after the announcement of the successful completion of the project in 2003 the costs for sequencing a single human genome were still above the 10 million dollar mark.
During the next ten years these costs would fall dramatically due to the groundbreaking developments in the field of genomic sequencing technology. In the year 2013 the same task would cost less than 10,000 dollar (see figure below).

DNA Sequencing Costs [27]

Sequencing the genomes of a large number of individuals and even whole populations seemed achievable for the first time. It is argued that the costs for sequencing a human genome would fall under the mark of 1.000 dollar[28] in 2015. Today, both commercial services and public health efforts in several states, explore the possibilities of genetic sequencing. To fully understand the possible consequent benefits of this intention we now take a closer look at the concept of genetic testing.

“I think that any healthcare advances made, that can aid people in living more healthy lives, should be properly communicated to bring about a greater awareness to the public about any health matters.”

Survey Participant – ID 454

3.3 THE CONCEPT OF GENETIC TESTING

Every human genome consists of about 20,000 protein-coding genes, which are considered relevant to the development of the human body and certain conditions. Genetic testing is a means of “reading” the human genome in order learn about the genetic profile of an individual and is described as the "the analysis of human DNA, RNA, chromosomes, proteins and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes or karyotypes for clinical purposes".[29] It is important to differentiate between a variety of different types of genetic testing, as there are newborn screening, diagnostic testing, carrier testing, preimplantation genetic diagnosis, prenatal diagnosis and predictive and presymptomatic testing. The work in hand focusses mainly on the ramifications of carrier testing and especially predictive and presymptomatic testing.
In order to be able to read the genetic information of an individual a blood sample - in some cases a saliva sample - is taken. With the help of modern genome sequencing technology this sample is examined in order to find certain genetic variations that could provide an indication to a genetically caused condition, such as cystic fibrosis, Huntington's or sickle-cell disease to name but a few. Some of these conditions are monogenetic, which means that a single (mutated) gene is the cause of the conditions.

Others are caused through a complex interplay between more than one gene. The multiple factors in this case are called polygenic.

It is self-evident that the enormous amount of data - and its meaningfulness - that is produced during a sequencing process is put under scrutiny by researchers and genetic counsellors. At the same time there is a discussion about whether a certain condition is caused by genes and genes only or whether the environment plays an additional role in developing the disease (which happens to be a frequent occurrence with health-related diagnosis). Very often genetic testing provides the patient with his or her personal genetic risk of developing a condition and this is where things can become inscrutable in. In order to provide the gentle reader with more information about this issue we subsequently discuss the problem of absolute and relative risks and the (potentially) resultant fallacy.

Risk estimation and the base rate fallacy

Epidemiology - often referred to as the science of public health - looks for the occurrence and existence (incidence and prevalence) of diseases in individuals, various demographic groups and whole populations, in order to generate information about the risk estimates for almost every known disease. These risk estimates need to be understood in different ways, depending on whether they have been related to a larger number of individuals, whole populations or to only one specific individual. The risk estimate for the letter is described as the relative risk, meaning that the calculated risk only applies to this specific individual. In contrast to this, the absolute risk refers to the risk estimates of the specific demographic group or population the mentioned individual lives in or is part of. As a matter of fact the risk estimates for the relative risk and the absolute risk of developing a certain disease might vary in a great measure and sometimes the head-to-head comparison between relative and absolute risks can lead to wrong conclusions in terms of estimating the individual risk of developing a certain disease or condition. This might be the particular case where a generally low absolute risk is confronted with an above-average relative risk for developing a disease or condition: with the (general) absolute risk being low in the first place even a slightly increased relative risk could lead to error in reasoning in some circumstances. Individuals with a lack of much needed scientific literacy are considered to be prone to this specific fallacy, also referred to as “base rate fallacy”. This
phenomenon specially applies to risk estimates derived from preventive genetic testing results.

“We should embrace the information flows that create knowledge. Diagnosis produced based on biometrics, standard labs, symptoms and patient history has never been 100 percent perfect. But, denying access to genetic and genomic data because it has the potential to be inconclusive or even wrong is as bad as denying access to primary care. Let’s foster curiosity instead of trying to stifle it.”

Survey Participant - ID 63

3.3 METAPHOR AND ANALOGY

The main purpose of this chapter is to communicate the importance of metaphors and analogies in modern science and scientific communication. The title of this thesis is a question on whether a person would want to “read” his or her own genome. Apparently the process of reading a human genome and – for instance – reading a book are two profoundly different domains. When reading a book, we use our basic skills of literacy, where letters turn from words to whole sentences. By reading one sentence after another we experience a coherent chronology of what is being told by the author of the book. Sometimes we might even be able to “read between the lines” of a book, a phrase which is often used when referring to a course of action that has somehow been communicated to the reader in an implicit way.

However, reading a human genome is a far more complex process. First of all, the human genome has its very own “alphabet”, consisting of the already mentioned four bases Adenine, Cytosine, Thymine and Guanine. Genes are a product of a certain sequence of these four bases. The genes themselves might – for instance – give a hint on whether a person might have green, blue or hazel eyes. Nevertheless, even such seemingly trivial information on a specific trait of an individual has very often a polygenic character, where more than one gene can play a role. In other words, when a gene is identified - or “read” so to say - the very meaning of the gene in terms of the manifestation in the human body is sometimes still uncertain.

“Imagine that the genome is a book. […] There are one billion words in the book, which makes it […] as long as 800 Bibles. If I read the genome out to you at the rate of one word per second for eight hours a day, it would take me a century. If I wrote out the human genome, one letter per millimeter, my text would be as long as the River Danube. This is a gigantic document, an immense book, a recipe of extravagant length, and it all fits inside the microscopic nucleus of a tiny cell that fits easily upon the head of a pin.”[33]
Despite this arguably very metaphorical description of a genome the analysis of the human genome is a multilayered process with various involved parties, such as patients, consumers, family members, physicians, researchers, clinicians and genetic counsellors, biotech companies and sometimes even insurance companies.

Even though this process is metaphorically described as an apparently simple task, in which a certain data carrier - in this case the human genome - is just read out of, the actual task itself consists of far more and the possible implications are manifold. As in the case of reading a book there is a distinctive difference between reading and understanding or interpreting the data in a meaningful way. In an analogous manner and as a matter of course the technique behind the analysis of a genome is far more complicated than just reading a text such as the one in hand. Almost everyone in the western world is able to read a book according to their own taste but not everyone is able to read their own genome at their convenience, at least not yet.[34]

The ability to read is considered a crucial part of a good education. Reading has become a means to open up one's horizons and to learn about the world, about our society and about ourselves. Humankind developed the braille alphabet in order to ensure that even those individuals who lost their sight might be able to read and therefore learn. In contrast to this the analysis of the human genome is – at least today – reserved to scientists, researchers, clinicians and a few opinion leaders and that happens for a good reason. It is the key task of such demographic groups to gain new and meaningful insights in the field of personal genomics long before the technology has been democratized through biotech companies and commercial services and made available to the wider public.

Reading the human genome has often been referred[35] to as a process similar to “opening Pandora’s box”, recalling the ancient greek myth in which unknown repercussions and a possible disaster would loom behind an unwitting action driven purely from curiosity. This sort of analogy is used in order to awaken the interest of various involved parties and to convince them, that they should exercise caution in whatever they do.

Hence, all of the above mentioned issues depict a metaphorical and very rough approximation towards the issue of genetic analysis as part of an ongoing public dialog. Whenever there is talk of a mysterious code of life, a blueprint with unknown contents or an alleged book of life the gentle reader should keep in mind that a metaphor and analogy has been used in order to be able to communicate a complex topic to a wider audience.

"A genetic predisposition is merely like a gun. I believe that environmental/lifestyle choices are the triggers which would shoot the gun. A genetic predisposition, or gun, is harmless in and of itself."

Survey Participant - ID 663
Human coexistence in social systems has been based on social norms for a long time. Social norms and the adherence to such norms are considered the foundation of peaceful coexistence within a modern society. When social norms are written down and enacted in the space of social institutions they become a multiplex system of rules which are commonly known as the law. The law defines if a questionable action that is made by a member of the society is rated as wrongdoing and, if so, it allows for appropriate compensation. It comes as no revelation that personal health care and the associated personal information is liable to certain legal requirements. The predominant cause of the existence of these requirements is undoubtedly the founded concern about the patients’ well-being and safety.

“Legal regulations have to be drawn up to ensure that the individuals affected are protected against discriminatory practices. The use and assessment of genetic information by organizational actors has likewise to be regulated, and legitimate uses distinguished from illegitimate ones.”[36]

The current chapter examines the most important standards and statutory regulations that were set up in association with the field of the analysis of the human genome. Interestingly enough, many states (from the western world) have inherited a large part of the regulations established within the inter- and transnational frameworks, such as the Universal Declaration on the Human Genome and Human Rights (UNESCO) or the Convention on Human Rights and Biomedicine (European Union). Furthermore, the point of time of entry into force shows how emergent the issue on the human genome, genetic testing and genetic information has become during the past decades. How the execution of these regulations will play out in the future years, where (personal) genomic data might become a ubiquitous commodity, will surely become a leading question for all involved.

4.1 INTERNATIONAL FRAMEWORKS

The United Nations Educational, Scientific and Cultural Organization (UNESCO) adopted the "Universal Declaration on the Human Genome and Human Rights”[37] in 1997. The document illustrates the most fundamental regulations concerning the proper handling in terms of the human genome. Altogether a whole of 77 national representatives agreed on these regulations. It is composed of seven sections with a total of 25 articles. The seven chapters are given the titles (A) Human Dignity and the Human Genome, (B) Rights of the Persons Concerned, (C) Research on the Human Genome, (D) Conditions for the Exercise of Scientific Activity, (E) Solidarity and International Co-Operation, (F) Promotion of the Principles Set Out in the Declaration and (G) Implementation of the Declaration.

"States should take appropriate measures to encourage other forms of research, training and information dissemination conducive to raising the awareness of society and all of its members of their responsibilities regarding the fundamental issues relating to the defence of human dignity which may be raised by research in biology, in genetics and medicine, and its applications. They should also undertake to facilitate on this subject an open international discussion, ensuring the free expression of various sociocultural, religious and philosophical opinions.”[38]

Additionally to the Declaration on the Human Genome the UNESCO adopted the "International Declaration on Human Genetic Data" in 2003 and the "Universal Declaration on Bioethics and Human Rights" two years later. While the former framework - among other things - aimed to
"ensure the respect of human dignity and protection of human rights and fundamental freedoms in the collection, processing, use and storage of human genetic data [...]"[39], the latter included without limitation the goal to "provide a universal framework of principles and procedures to guide States in the formulation of their legislation, policies or other instruments in the field of bioethics".[40] Apart from these declarations, which as such have no legal character by itself, other transnational regulations and norm-setting instruments were introduced by the European Union.

In 1997 the European Union - at that time being referred to as the European Community - and its member states agreed upon the "Convention of Human Rights and Biomedicine"[41], consisting of a total of 14 chapters, including - but not limited to - issues like non-discrimination, scientific research or the public debate.

"Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling."

In an additional protocol[43], adopted in 2008 and devoted primarily to the field of genetic testing for health purposes, the EU addressed the vital role of adequate quality[44] of genetic services - particularly the scientific and clinical validity and clinical utility - genetic counselling[45] and the right to remain ignorant.[46] Article 7 of the protocol, stating that "A genetic test for health purposes may only be performed under individualised medical supervision" can be considered to be of main importance in relation to the emergence of direct-to-consumer genetic testing, commonly not known for providing such a service in a satisfactory way.

All of the above mentioned frameworks can be seen as universal guidelines for all signing parties on how to conduct on a national level when concerned with genetic information and the implications of genetic research at all levels. As a matter of fact a large part of these basic regulations have been inherited and legally incorporated by many states in Western Europe, such as France, Germany, Sweden, Switzerland or Austria.[47] Subsequently the work in hand discusses the genesis of national legislations in excerpts and the proper handling in terms of genetic testing.

"I would worry that, regardless of current health system mandates, information could be used against someone, either by patenting genes, or by prejudicial selection."

Survey Participant - ID 753

4.2 NATIONAL FRAMEWORKS

The first European country enacting an explicit law on the approach towards human genetic information was considered to be France in 1994 (revised in 2004).[48] The Austrian "Gene Technology Act" came in force one year later (1995) and was revised in 2005, classifying genetic tests into 4 different types. The first two types remain reserved to the purpose of
determining a present disease, which makes them not applicable in terms of preventive genetic testing. However, the remaining two types do serve the particular purpose of genetic predisposition testing, focusing on treatable (type 3) diseases and diseases that are considered to be not treatable (type 4).\[49] The execution of type 3 and type 4 genetic testing is strictly reserved to licensed facilities and is subject to the condition that a qualified health care professional is involved.\[50]

<table>
<thead>
<tr>
<th>Country</th>
<th>Name of the Act</th>
<th>In force as of</th>
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<tbody>
<tr>
<td>Germany</td>
<td>Human Genetic Examination Act</td>
<td>2010</td>
</tr>
<tr>
<td>Norway</td>
<td>Act on human medical use of biotechnology etc.</td>
<td>2004</td>
</tr>
<tr>
<td>Portugal</td>
<td>Act on Personal Genetic Information and health information</td>
<td>2005</td>
</tr>
<tr>
<td>Spain</td>
<td>Act on biomedical investigations</td>
<td>2007</td>
</tr>
<tr>
<td>Sweden</td>
<td>Act on genetic integrity</td>
<td>2006</td>
</tr>
<tr>
<td>Switzerland</td>
<td>Federal Act on Human Genetic Analysis</td>
<td>2007</td>
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</tbody>
</table>

"Legal acts governing the use of genetic testing in Western Europe" [51]

The United States of America enacted the "Genetic Information Nondiscrimination Act" in 1998, addressing the potential "employment discrimination on the basis of genetic information." [52] The US Act aims for the protection of the individual and the genetic data in a similar way as the already mentioned national and international frameworks.

"The potential for gene based discrimination concerns me.'

Survey Participant - ID 367

CLINICAL GENOMICS

The analysis and interpretation of genomic information of an individual has become increasingly sophisticated. Clinical application of genomics and genomics-based technology itself has been standard practice long way before personal genetic testing – for example in the form of direct-to-consumer genetic testing - was at disposal for the wider public. During the last decades a variety of clinical genetic testing methodologies[53] have emerged, each of them with different levels of analytical and clinical sensitivity, analytical and clinical specificity, clinical and personal utility and cost.

The main purpose of this chapter is to shed light on some of the most important standards, research methods and processes that are applied when genetic information is identified and used as a health indicator in clinical care. For that to happen, this chapter contains elementary information about false positives and false negatives in medical testing, fundamentals about the validity of genetic testing, the fundamental principles of genome-wide association studies (GWAS) and the basics on genetic counseling.

"Keeping pace with emerging clinical genetic technologies requires specialized genetic training as well as broad genetic literacy for patients and clinicians ordering and receiving genetic test results."[54]

Medical testing of any sort has always involved a fraction of uncertainty when a test result was found to hint at a possible impairment to health. A diagnosis that was produced based on
biometrics and standard labs often needs to be verified by repeating the test in order to exclude misleading or even wrong results, referred to as so called false positives or false negatives. Both false positive and false negative results are considered to pose a great danger to patients ordering genetic testing and clinicians or researchers when interpreting the results in their daily work.

**False Positives and false negatives**

Every medical test aims to provide patients and clinicians with some indication of an individuals' physical state, be it a test for the purpose of detecting whether a woman might be pregnant, a test that would indicate an infectious disease or a genetic test that would suggest a genetic risk for a certain condition. Commonly such a test has two possible outcomes, the first one being positive and the second being negative. A true positive test result (A) usually (see chart below) reveals an existing condition or disease, whereas a true negative test (D) result predicates no existing disease or condition. However, almost every medical test is sometimes considered to be prone to delivering misleading results. In this case, the false positive test result (B) incorrectly indicates an existing disease or condition when - in actual fact - the individual under test has not acquired the particular disease or condition. To the same effect, a false negative test result (C) erroneously provides no evidence for an existing disease or condition, when in reality such a disease or condition does exist.

<table>
<thead>
<tr>
<th>Test Positive</th>
<th>Test Negative</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Existing Disease or Condition</strong></td>
<td><strong>No Disease or Condition</strong></td>
</tr>
<tr>
<td>(A) True Positive</td>
<td>(B) False Positive</td>
</tr>
<tr>
<td>(C) False Negative</td>
<td>(D) True Negative</td>
</tr>
</tbody>
</table>

In the specific area of predictive or presymptomatic genetic testing the error in data reporting represented by possible false positive respectively false negative test results yields additional risk to actually do more harm than good. It is in the nature of predictive genetic testing to provide the individual concerned with a percental risk or probability of developing a certain disease or condition in the future. However, due to environmental factors and the diagnostically less conclusive results the exact moment of the manifestation of this particular disease is unclear and even the mere subsistent existence of such a manifestation itself in the future unpredictable and unprovable (at the present point in time). At this point, the question, whether a nonexisting or misleading predictive genetic test embodies a range of possible false negative test results has also to be taken into consideration.

“As someone who is very familiar with the technologies involved I have to say that I am still indecisive if I would want to have my genome read and reported out. I already know most of my risk factors due to family medical histories. Most of the data I would obtain might tell me I have a somewhat increased risk of disease but […] in most cases it will still be a low absolute risk. I figure I should try to really just be as healthy as I can be regardless of what my genes tell me”
5.1 VALIDITY OF GENETIC TESTING

Genetic testing and the process of collecting, interpreting and reporting of genomic information in the clinical environment is liable to a plethora of components that aim for ensuring a methodically accurate approach in clinical decision-making. In this process, an individual's disorder (or disease) and the clinical setting are considered to occupy the centre stage. This central point is belted by a variety of important measurement tools respectively key figures, which are divided into analytical validity, clinical validity, clinical utility and - in an further step - personal utility.

The area of analytical validity - with key figures like analytic sensitivity, analytical specificity, quality control and assay - scrutinises the test's "ability to accurately and reliably measure the genotype of interest". This measurement tool allows to control whether a genetic test has truly identified the genetic information, that the test looked for in the first place. On the other hand, clinical validity - with key figures like clinical specificity, clinical sensitivity, prevalence, PPV's and NPV's and penetrance - looks for a "test's ability to detect or predict the associated disorder (phenotype)".

Atop of all the components of the discussed model process the measurement tool of clinical utility is applied in order to determine possible treatment methods and the form of patient management that is necessary in the specific case. As already mentioned, the value of the genetic test for personal and family choices in defined as the personal utility. Except for clinical and personal utility, all of the above discussed measurement tools are based on the calculation of various ratios between true positive respectively true negative and false positive respectively false negative results. Contrary to this, the value of a genetic test for determining medical treatment and patient management (clinical utility) and the value of such a test for personal choices of the patient (personal utility) are - based on the results of the preceding measurement results - subjectively determined. In the field of clinical utility evidence-based reports and economic benefits decide whether a test will be used, whereas the personal utility of a genetic test is defined based on the patients' personal perspective.

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
<th>Calculation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Analytical sensitivity</td>
<td>false-negative rate of the assay</td>
<td>true positives/(true positives + false negatives)</td>
</tr>
<tr>
<td>Concept</td>
<td>Formula</td>
<td></td>
</tr>
<tr>
<td>-------------------------------</td>
<td>------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>Analytical specificity</td>
<td>false-positive rate of assay</td>
<td></td>
</tr>
<tr>
<td>Clinical sensitivity</td>
<td>false-negative rate of diagnosis</td>
<td></td>
</tr>
<tr>
<td>Clinical specificity</td>
<td>false-positive rate of diagnosis</td>
<td></td>
</tr>
<tr>
<td>Positive predictive value (PPV)</td>
<td>likelihood that a patient has the disease given that the test result is positive</td>
<td></td>
</tr>
<tr>
<td>Negative predictive value (NPV)</td>
<td>likelihood that a patient does not have the disease given that the test result is negative</td>
<td></td>
</tr>
<tr>
<td>Clinical utility</td>
<td>value of the test for determining treatment, patient management and family planning</td>
<td></td>
</tr>
<tr>
<td>Personal utility</td>
<td>value of the test for personal and family choices</td>
<td></td>
</tr>
</tbody>
</table>

“Evaluating the validity of genetic tests” [58]

Given that genetic testing in the clinical environment is subject to the measurement tools discussed above and the decision whether a genetic test is required in the first place or diagnostically conclusive is based on the possible benefits (and threats) for the patient, the difference between clinical genomics and, for example, direct-to-consumer genetic testing - where no intermediary in the form of a health care professional is present - become evident (see table above).

“Genome sequencing can be informative or confusing, helpful to professional genetics counselors or misleading to subjects...they are not a useful tool if there is no cure for the diseases they pick up on, and there is no guarantee you will suffer from a disease uncovered by whole genome sequencing. It is too soon....”

Survey Participant - ID 157

5.2 GENOME-WIDE ASSOCIATION STUDIES (GWAS)

Personal genomics as a possibility for identifying the genetic profile of an individual for health-related reasons has not been around for a long time. The first milestone has been established after the completion of the first draft of the human genome with the help of the Human Genome Project. However, the collection of large amounts of genetic profiles has become possible only because of the rapid developments in the field of genomic sequencing technologies in the following years, which have been already discussed earlier in this thesis. Decreasing sequencing costs and the broadening availability of these sequencing technologies enabled researchers, scientists, students and also entrepreneurs to create unprecedented amounts of new databases containing large amounts of genetic information. This information forms the basis for a new method of studying varying components of the human genome and led to the increasing emergence of so called genome-wide association studies (GWAS). During the past decade years the total number of publications of genome-wide associations studies rose from less than 100 publications in the year 2005 to almost 2000 publications in the year 2013, and a further increase is expected. Due to the fact that the results that derive from GWA studies have enormous influence on clinical genomics, the current chapter discusses the fundamental principles of the most common study designs.
Most genome-wide association studies are based on three different study designs, which are case-control studies, cohort studies and the special case of trio design. Case control studies aim at looking for genetic variations between two groups, where the members of one group are considered to suffer from a certain condition or disease while the members of second group are considered to be free of complaints. After the sequencing process researchers look for differences in certain parts of the genome of both groups in order to discover genetic variants which can be associated with the existing condition or disease in the first group. On the other hand, cohort studies monitor a large group of individuals over a longer period of time, who - at the beginning of the study - do not suffer from a certain condition or disease. Members of this group, who develop a condition or disease during the observation period are - based on their genetic profile - continuously compared with members who have not experienced the same condition. As before, the goal of cohort studies is the discovery of associations between genetic variants and the incidence of a certain disease or condition. Lastly, the special form of the trio design has a unique approach when conducting a genome-wide association study by looking for genetic variants between parents and their children, who are considered to suffer from a certain condition or disease. This special design is seen as a suitable method to investigate children's conditions (for a detailed explanation of all three study designs see table on the next side).

"The potential for false-positive results, lack of information on gene function, insensitivity to rare variants and structural variants, requirement for large sample sizes, and possible biases due to case and control selection and genotyping errors, are important limitations of GWA studies."[60]

During the past ten years results derived from genome-wide association studies have been a driving factor for an increasing number of reports about genetically determined associations for a variety of human complex diseases, such as asthma and other pulmonary diseases, coronary heart diseases and even bipolar disorders and schizophrenia. As a matter of fact public opinion becomes potentially open for the idea of genetic determinism in an unquestioning way, where genetic predisposition is seen as a leading factor of influence for almost all human diseases and conditions

<table>
<thead>
<tr>
<th>Assumptions</th>
<th>Case-Control</th>
<th>Cohort</th>
<th>Trio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case and control participants are drawn from the same population</td>
<td>Participants under study are more representative of the population from which they are drawn</td>
<td>Disease-related alleles are transmitted in excess of 50% to affected offspring from heterozygous parents</td>
<td></td>
</tr>
</tbody>
</table>
representativeness are clearly specified Genomic and epidemiologic data are collected similarly in cases and controls. Differences in allele frequencies relate to the outcome of interest rather than differences in background population between cases and controls. Similarly in individuals with and without the gene variant.

### Advantages

**Short time frame**
Large numbers of case and control participants can be assembled
**Optimal epidemiologic design for studying rare diseases**
Cases are incident (developing during observation) and free of survival bias
Direct measure of risk
Fewer biases than case-control studies
Continuum of health-related measures available in population samples not selected for presence of disease
Controls for population structure; immune to population stratification
Allows checks for Mendelian inheritance patterns in genotyping quality control
Logistically simpler for studies of children's conditions
Does not require phenotyping of parents

### Disadvantages

**Prone to a number of biases including population stratification**
Cases are usually prevalent cases, may exclude fatal or short episodes, or mild or silent cases
Overestimate relative risk for common diseases
May be difficult to assemble both parents and offspring, especially in disorders with older ages of onset
Highly sensitive to genotyping error

Large sample size needed for genotyping if incidence is low
Expensive and lengthy follow-up
Existing consent may be insufficient for GWA genotyping or data sharing
Requires variation in trait being studied
Poorly suited for studying rare diseases

"Study designs used in genome-wide association studies" [61]

"The current state of knowledge is too unreliable for general use of predictive genetic testing save in specific areas. This, offering to wider public rather than on targeted basis is not appropriate at present."

Survey Participant - ID 196

5.3 GENETIC COUNSELING
In clinical genomics the practice of genetic counseling has experienced a deepening professionalisation during the second half of the 20th century. With the scientific understanding for and the medical detection of monogenic and chromosomal disorders becoming better and better it embodied new possibilities for medical attendance.[62] Genetic counseling is described as "the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease"[63] and was mainly considered a procedure to prevent inherited birth defects and genetic disorders.[64]

While genetic counseling in the clinical environment is primarily seen to represent a method of careful screening of a patients' individual medical history as a way to advise the patient on decisions in family planning and preventive measures in relation to the genetic profile it also emphasises on the sociocultural background of the persons concerned before doing so[65] Understandably the level of critical health literacy and the mere ability to deal with sensitive and/or unsettling medical information might not only vary between all strata of a homogenous society but between and within ethnic groups as well. Addressing these differences is considered to be one of the most difficult tasks in genetic counseling.[66]

"The responsibility to provide accessible, useful genetic counseling to individuals from many cultures and ethnicities arises from the increasing ethnocultural diversity of the populations served, coupled with the ethical goal of providing equal access and quality of services for all individuals. […]"[67]

As opposed to the practice of genetic testing in the clinical environment, the roles of genetic counsellors in relation to direct-to-consumer genetic testing have expanded, from an educating and mediating role as a risk interpreter to

“At this point, not enough is known, particularly by medical professionals (I could do a better job of analyzing my genome than my physician ever could), about causative relationships between specific genes and increased risk of disease to make sequencing and analysis worth the costs.”

Survey Participant - ID 346

PROJECTS AND COMMERCIAL SERVICES

The application of gene sequencing technologies has been reserved to research facilities, public authorities and hospitals for a long time. However, after and already before the completion of the publicly funded Human Genome Project a variety of ambitious entrepreneurs and start-ups joined the emerging race in personal genomics. One of the most salient examples for this trend was the establishment of the “Celera Corporation”, founded in 1998, only a few years before the completion of the Human Genome Project. The company was - amongst others - founded by Craig Venter, a biochemist and geneticist, who has been part of the international research team of the Human Genome Project. In consequence of a professional disagreement between Venter and the rest of the team, he decided to take another path by launching the privately funded company Celera. Eventually the company succeeded with its unique strategy in sequencing[68] the human genome at lower cost than the Human Genome Project and became a perfect example for a efficient private sector company in the emerging genomics markets.

While the availability of genetic tests was previously and at this stage limited and offered to
patients by intermediaries such as genetic researchers and health care professionals like physicians and geneticists, privately funded companies and new research initiatives mushroomed during the first decade of the new millennium, with a view to doing business and research on their own. Direct-to-consumer (DTC) genetic testing kits became a new method of acquiring genomic information from thousands of individuals, who would in return get an insight on their personal genome without the requirement of suffering from a certain disease or condition, solely based on the consumers personal interest and curiosity. Long term, large cohort studies started to collect genotypic and phenotypic information of interested volunteers in order to publicize this information for future use.

The following chapters outline the most well-known projects and commercial services in the field of personal genomics, namely the Harvard Personal Genome Project and its international network, the personal genomics 23andMe and the market for genomic sequencing and analysing technologies in general. Last but not least the efforts of national public health genomics programmes will be discussed in detail.

"I am located in the United States and have utilized the three major direct-to-consumer companies here for various family members: 23andme, ancestryDNA, and familytreedna. I have also used promethease, snpedia, runs of homozygosity, and various other tools including pubmed (searching for SNPs) as adjunctives to genetic testing. I have found it incredibly useful to myself and my family. I agree that it is a medical tool. I would still be angry to know it was being kept from me or that it was prohibitively expensive in order to limit the use to medical professional."

Survey Participant - ID 219

6.1 THE HARVARD PERSONAL GENOME PROJECT (GENOM AUSTRIA)

The Harvard Personal Genome Project was initiated[69] in 2005 at the Harvard University in the United States of America by the geneticist George M. Church. The project is referred to as "participatory public research"[70] because it aims for collecting the genomes of volunteers, who consent to give away their personal genetic profile for research purposes. The project established the Global Network of Personal Genome Projects which currently consists of project partners in Canada (PGP Canada, founded in 2012), the United Kingdom (PGP UK, founded in 2013) and Austria (Genom Austria, founded in 2014).

"The Personal Genome Project (PGP), an ambitious research study directed by faculty members in the Department of Genetics at Harvard Medical School, aims to recruit as many as 100 000 informed participants to contribute genomic sequence data, tissues, and extensive environmental trait, and other information to a publicly accessible and identifiable research database."[71]

As of August 2015, the project had more than 4.400 participants[72], who agreed to provide samples of saliva or blood for the purpose of genomic sequencing and - where applicable - to give away additional information on existing conditions, medications or possible allergies. Furthermore the project also asked participants for already existing genome data[73], which was often provided by other companies or projects, like 23andMe, Family Tree DNA, Promethease, AncestryDNA or Navigenics - all of them being big players in the field of personal genomics. However, the Personal Genome Project and its international network work as a not-for-profit association and the members of the network are not allowed to "sell or license participant data
or tissues".[74] In late 2014 the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, and the Medical University of Vienna announced the joint PGP-project "Genom Austria" with the goal to "explore the scientific, educational, ethical, and social implications of personal genome sequencing".[75]

"Am a biomedical scientist, I find molecular biology and genetics interesting and they are the future of medical therapeutics, so I would gladly read m[y] genome."

Survey Participant - ID 464

6.2 THE PERSONAL GENOMICS COMPANY 23ANDME

With the foundation of the personal genomics and biotechnology company 23andMe in early 2006 the practice of “direct-to-consumer” genetic testing began to emerge on a large scale. For the first time ever, a company - whose product has been named[76] the "retail DNA test" and priced as "invention of the year" in 2008 - offered a genetic testing kit for interested consumers. After ordering such a kit, consumers would receive a parcel by mail containing a "spit kit" enabling them to provide a saliva sample. A few weeks after having sent back the sample, consumers would be informed about their own genetic (health) profile and the associated genetic risks, based on their saliva sample provided earlier.

The price for a genetic testing kit from 23andMe has been falling dramatically over the past eight years, mirroring the general trend caused by cheaper and more cost-effective genomic sequencing technologies. As of late 2007 the testing kit was offered to US-consumers[77] for $999 and by late 2008 the price has fallen by more than 50% resulting in a total cost of $399. Meanwhile the product was also shipped to consumers from Canada and interested parties from more than 40 European countries.[78] In late 2010 the company reduced the cost for their product again, offering it for $199 and by early 2011 the price for a genetic testing kit dropped a last time, settling down at $99, unchanged up to the present day.

In early 2013 - seven years after its foundation - the company stated that the service has been provided to more than 250,000 costumers.[79] After having launched a large TV ad campaign in the same year - followed by a warning letter from the US Food and Drug Administration - the company announced their one millionth costumer at midyear 2015. Since all of the above mentioned developments play an important role in regards to the field of personal genomics, we discuss the product at issue and the events involving the FDA, which led to unprecedented results. 23andMe’s genetic testing service provided consumers with information about disease risks, personal traits, genetic carrier status and the individual drug response based on the individuals’ genetic profile. The health report revealed the absolute and relative risk[80] for 100 complex diseases like type 2 diabetes, prostate cancer or colorectal cancer by looking at certain variants in the consumers genome associated with the specific disease.[81]
Furthermore, the test results showed the genetic carrier status for approximately 24 inherited diseases, such as the BRCA cancer mutation (indication for genetically inherited breast cancer) or Cystic Fibrosis, and informed the consumer about her or his individual response to various drugs based on the genetic profile. Last but not least, the report contained basic information on traits like eye color or similar phenotypic characteristics. After having offered the health reports as part of the genetic testing kit, in late 2013 23andMe received a warning letter from the US Food and Drug Administration, forcing the company to immediately stop marketing the service and the related health reports.[83]

The Food and Drug Administration (FDA) complained about the missing clinical and analytical validity of the provided genetic testing service, addressed the threat deriving from possible false positive and false negative test results and the general danger of doing more harm than good when providing such a service without preceding approval and marketing authorisation from the FDA. Additionally, the FDA pointed out that the service provided by the company was believed to constitute a medical device which can be used for the purpose of medical diagnosis.[84]

"[...](your company’s) website at [www.23andme.com/health](http://www.23andme.com/health) (most recently viewed on November 6, 2013) markets the PGS for providing ‘health reports on 254 diseases and conditions’ [...] as a ‘first step in prevention’ that enables users to ‘take steps toward mitigating serious diseases’ such as diabetes, coronary heart disease, and breast cancer. [...] Most of these uses have not been classified and thus require premarket approval or de novo classification [...]”[85]

After having received the warning letter from the US Food and Drug Administration, the company additionally had to face a class action lawsuit from disgruntled customers. The lawsuit against 23andMe was based on the same concerns and arguments communicated in the warning letter from the FDA. The proposed class action alleged “that 23andMe, Inc. (‘Defendant’) falsely and misleadingly advertises their Saliva Collection Kit/Personal Genome Service (‘PGS’) as providing ‘health reports on 240+ conditions and traits’, ‘drug response’, ‘carrier status’, among other things, when there is no analytical or clinical validation for the PGS for its advertised uses.”[86]

Within a few days the company retracted their service, stopped the marketing campaign and undertook every effort to cooperate with the US Food and Drug Administration. At the present time the company does not offer any sort of health report for its US customers. However it does offer the same service - including health reports - in Canada and the UK, where - based on the diverging legal situation - no sanctions were imposed down to the present day.
The company’s effort to (successfully) collect more than one million genetic profiles from its customers during a time period of approximately eight years was observed by various parties, ranging from public authorities, research facilities, competitors and cooperation partners to the media and several academic disciplines. Proponents argued in favor of the mere abundance of genetic data that could be used for future research in the form of genome-wide association studies while the opponents voiced misgivings about data privacy, the (psychological and physical) wellbeing of the customers and the mere fact, that distinctly private (health) information was handed in to a privately funded company by unknowing consumers, who would in turn indeed pay in order to give away sensitive data. As a matter of fact the company has already announced its plans to share a considerable amount of the accumulated genetic data with large drug companies, even though the participants from this share have agreed on their genetic data to be circulated for research purposes.[87]

"Relationship between heritability, genetic complexity and predictive ability of personal genome testing" [88]

The direct-to-consumer product and the associated health report marketed by 23andMe has become the new starting point of a recurring discussion about fundamental and dissenting opinions and views on the future of personal genomics in the form of a directly distributed commodity. While regulations on direct-to-consumer genetic testing are demanded and welcomed by strict opponents, arguing in favour of the "protection of the ignorant"[89] (often discussed in relation to "the right not to know"), proponents see a fundamental violation of the right to learn and especially the "right to know" about the one’s genome.[90] However, in terms of the interpretation of genomic information for end-consumers, DTC-genetic testing is surely to face strong scrutiny in the near future.

"Given that numerous genetic and non-genetic risk factors interact in the causation of complex diseases, the predictive ability of genetic models will likely remain modest. Personal genome testing will have minimal benefits for individual consumers unless major breakthroughs are made in the near future."[91]

Although the company may earn a healthy profit by selling genetic information to third parties, it may not make a profit by selling genetic testing kits and the associated health reports anytime in the near future. The case of 23andMe - serving as an apple of discord - exemplifies the vast complexity of direct-to-consumer genetic testing in its entirety, whether it be the legal controversies, the discussion about data privacy or the fundamental difficulty in predictive genetic testing, especially for complex diseases (see figure on preceding page)
“I am worried that there is a potential for private companies to use the genome sequences and fear to hawk unnecessary products or treatments to scientifically illiterate folks.”

Survey Participant - ID 358

6.3 THE MARKET FOR GENOMIC SEQUENCING TECHNOLOGIES

Having discussed the tremendous developments of genomic sequencing technologies previously, we subsequently take a closer look at a few of the largest suppliers, some of them developing and manufacturing sequencing technologies for more than 15 years. The most well-known and most successful one is a US company with the telling name Illumina, founded in early 1998 and listed already on the stock exchange two years later. However, the company began offering genotyping services not until 2001. Illumina has been the main innovator in the field of genetic sequencing and sold its devices to research facilities from all over the world, holding more than two-thirds of the market for genomic sequencing technologies in 2013.[92] Revealingly the already mentioned personal genomics company 23andMe uses equipment from Illumina - the "Illumina HumanOmniExpress-24 format chip" - to genotype more than one million customers.

In 2014 the genetic sequencing company Illumina introduced its latest unit, the "HiSeq X Ten System", calling it "the most powerful sequencing platform ever created".[93] The new system was tailored for large research facilities and sold "as a combination of at least 10 HiSeq X systems, for a total cost of at least $10 million".[94] The new device kicked off a discussion about whether it would become possible to sequence a human genome for less than $1,000, which was seen as landmark for years.

"[...] scientists think that they will need to sequence hundreds of thousands or even millions of people to truly understand how genes influence disease so that better drugs and treatments can be developed. They say that $1,000 genomes are needed to enable the huge sequencing studies that could lead to breakthroughs in personalized medicine."[95]

Whatever the outcomes in the future may be, Illumina - with a revenue of $559 million in the 2015 second quarter[96] - is surely to continue being one of the most successful and influential supplier of genomic sequencing technology for the next years.

While Illumina is considered to be the largest supplier for genomic sequencing technologies for large-scale research efforts at the present time and its equipment is obviously exclusively affordable for high-end research facilities and well-financed for-profit companies, new
developments - making genomic sequencing available for a wider public, possibly even for interested end consumers - are already getting off the starting blocks. One of the most promising advancement has been established by the UK-based company Oxford Nanopore Technologies, founded in 2005. The company developed a portable USB device - which can be connected to a PC or laptop - that would use a specific sequencing method, called "nanopore sequencing" in order to allow users to sequence a genome in near real-time. The cost for device is currently at $1,000 - representing an access fee for the usage of the underlying computer programme MAP, which has also been developed by the company[97]

Even though the most part of the technological development identifies as a progress concerning the hardware, plenty of software applications for the purpose of analysing genomic data has been developed during the last years. These include, amongst others, OpenSNP and Promethease, both working in a similar way and accessible for little cost for the wider public already.

"Online genomics services enable everyone who is willing to contribute their genomic and lifestyle data to research to do so. OpenSNP is one such platform […] that offers consumers of online genetic and genomic testing services ways to 'publish their results, find others with similar genetic variations, learn more about their results, find the latest primary literature on their variations and help scientists to find new associations"[98]

At the same time large transnational technology companies like Apple[99] and Google[100] already try to gain a foothold in the genomics market by offering tailored services for the coming era of big data genomics. Before we dive into possible social and ethical implications, the following chapter discusses national efforts in genome sequencing.

“I am going to do this, but mainly for researching where my ancestors come from. I am not interested in the health aspects of the test. I have read that the companies offering this service cannot reliably make assumptions based on the data, and therefore I will choose to ignore the 'health' portion of the report. I assume that if these tests were useful diagnostically, health services would be using them already.”

Survey Participant - ID 701

6.4 NATIONAL PUBLIC HEALTH GENOMICS PROGRAMMES

National studies on the human genome and the sequencing and interpretation of larger populations have been a goal for several countries during the past decades. One of the earliest attempts was considered to be the Faroe Genome Project, with the unique goal of sequencing the genome of the Faroese population.[101] The project was carried out by the islandic-based and in the beginning privately funded company DeCode Genetics and was accompanied by harsh criticism about data privacy, which was to become a justified concern: the company filed bankruptcy and sold the aggregated genetic data to US investors.[102] A more up to date example for a national public health programme is the 100,000 Genomes Project established under the umbrella of the company Genomics England, being part of National Health Service (NHS) in the United Kingdom.

“Genomics England, a company wholly owned and funded by the [UK] Department of Health, was set up to deliver this flagship project which will sequence 100,000 whole genomes from NHS patients by 2017. Its four main aims are; to create an ethical and transparent programme based on consent; to bring benefit to patients and set up a genomic medicine service for the
NHS: to enable new scientific discovery and medical insights; and to kick start the development of a UK genomics industry.”[103]

The project not only aims to sequence the genome of (eligible) patients in the UK health care system but also focusses on the study of genomic structure of cancer and rare diseases. Another example of a nationally planned, genomic public health programme is the Precision Medicine Initiative, announced by the US administration in early 2015. Similar to the UK-programme, the initiative focusses mainly on cancer and oncological diseases. The initiative plans to assemble “a longitudinal ‘cohort’ of 1 million[sic!] or more Americans who have volunteered to participate [...]”, asking them to “give consent for extensive characterization of biologic specimens [...] and behavioral data, all linked to their electronic health records”. [104]

Even though both programmes, the 100,000 Genomes Project in UK and the US Precision Medicine Initiative, were established with the involvement of the respective government – and as such with the authorization of the public – the proper handling of genomic information is surely to be held under observation.

“This uncharted territory of genome research is really interesting and I believe it’s really good that it can help people have better lifestyles. However, I do think we have to thread carefully in this field, what some could use this knowledge for good purposes, some others could use it for their own benefit whether it is financial or else.”

Survey Participant - ID 518

SOCIAL AND ETHICAL IMPLICATIONS

Technological progress, scientific developments as revolutionary as the Human Genome Project, and the resulting insights into the human condition, which have led to continuing medical progress and the implementation of new practices for medical application, have always been accompanied by a questioning academic and - in further consequence - a broadening public dialogue. Whether it might have been a discourse about ownership of genetic information, inappropriate applications of genetic testing, the potential for discrimination, the limitations of genetic testing or patenting of genes to name but a few - the social and ethical implications of personal genomics are without any doubt manifold and complex. With genomic sequencing and interpretation technologies - both in terms of hardware and software - becoming more and more democratized utilities, the consistent continuation of these discussions become imperative and indispensable.

7.1 Data Privacy

Health data and information derived from clinical medical examination as well as the personal health history of every individual has ever since been handled in a strictly confidential way in every physician-patient relation. With advancing digitalisation of health-related data and the emergence of electronic health records concerns about data privacy gain centre stage all the more. Raw genetic data is by definition information, which can only be stored on a digital data network and therefore at all times prone to unauthorised access by malevolent parties.

Although genetic data is mostly stored in apseudonymised (“de-identified”) manner, where the option to draw inferences from the characteristics of the genetic information about a persons’ identity is made almost impossible or at least strictly limited, proof of concept experiments have already been able to identify individuals based on the seemingly encrypted genetic information.

In a "re-identification experiment" researchers from the Harvard University successfully
aggregated a variety of meta-information derived from the publicly available dataset of the already mentioned Personal Genome Project in order to identify participants.

"By linking demographics to public records such as voter lists, and mining for names hidden in attached documents, we correctly identified 84 to 97 percent of the profiles for which we provided names. Our ability to learn their names is based on their demographics, not their DNA, thereby revisiting an old vulnerability that could be easily thwarted with minimal loss of research value. [...]"[105]

However, the Personal Genome Project itself provides potential participants with the information that the participation on the project is not non-anonymous by default and states that it "cannot guarantee privacy".[106] By acknowledging the expressed risks and benefits linked with the participation, the project aims for the informed consent of all participants.

As a matter of fact, informed consent has been described to be "a cornerstone of the ethical conduct of research involving humans"[107], disemboguing in the strong conviction that study participants should - at all times - be fully aware of every possible risk and benefit associated with genomic research and their participation in such a research effort. By giving away blood or saliva samples, participants contribute to research and public health as well as to their own healthiness, but often they also place themselves in danger of staying uninformed about the further processing of their personal data.

In times of onward digitalisation of health data, data protection in genomic research has to take first priority in order to protect individuals from genetic discrimination and similar disadvantages resulting from unnecessary data leaks, both in clinical genomics and personal genomics provided by commercial services and not-for-profit research projects.

"Could be dangerous if this info goes to health insurance companies or government agencies."

Survey Participant - ID 216

7.2 GENETIC DISCRIMINATION

The fear from genetic discrimination is well-founded. The disputable and dubious reemergence and practice of eugenic ideology during the 20th century - not uncommonly demanded and promoted from all political colours[108] - laid the foundation for a critical reappraisal of genetic diversity, biological determinism and the use of genetic information. With genetic information becoming more and more commonplace, modern society is obligated to act cautiously and "to place the problem of genetic discrimination in a juridical framework".[109]

In the year 2005 the European Commission asked for the opinion of EU citizens on the "diagnostic and social uses of genetic data"[110] and the results showed a divergent attitude between the member states of the European Union. While the acceptability of uses of genetic data was relatively high in Belgium, Portugal and France, people from countries like Germany, Denmark and Austria showed to be less accepting of such uses.
The readiness to undergo genetic testing in order to detect possible (genome-based) diseases was lowest in Austria, with only 45% of all participants answering in the affirmative - while at least half of all participants from the remaining 24 EU member states stated they would take a health-related genetic test (EU25-average at 64%). However, when asked for the willingness to give away genetic information to the government or private insurance companies, most participants from all EU member states were far more reluctant: only an average of 25% was willing to disclose genetic information to governments and 14% were ready to disclose it to private insurance companies. These discrepancies clearly show that genetic information is seen as a two-edged sword, where the benefits of health-related genetic testing stand face-to-face with the risk of being exposed and potentially discriminated.

"As regards the relationship of genetic discrimination and racism, it bears stating that some genetic diseases are more prevalent in certain ethnic groups than in others. [...] Because certain ethnic groups are differently susceptible to particular genetic illnesses, there is a danger that members of minorities will be associated with such genes and treated as if they were ill, even if they do not bear the particular genetic mutation."[112]

Given that discrimination of every description - whether it occurs in the occupational socioenvironment, in family circles, the set of acquaintances or in the area of political and socio-cultural coexistence - can lead to disastrous consequences, like the acceptance of social inequality, inhuman social interaction and injustice, the specific phenomenon of genetic discrimination has the dangerous potential to add to such developments. Policymakers, educators, academics, researchers and scientists, health professionals, businesses and the civil society as a whole have an obligation towards every single individual to ensure that fundamental human rights are protected and human dignity is secured by a universal legal framework and - in the case of misconduct - effective sanction measures are imposed. With genomic information and a genomic economy on the rise the threat of genetic discrimination should continue to gain centre stage in the academic and political discourse and the domain of medical application of genomic data.

"If there were steps or treatment for particular disease processes that might develop, then it could be useful. If you could guarantee that insurance companies would not use the information to discriminate against you, it could also be useful."

Survey Participant - ID 298
7.3 KNOWLEDGE AND GENETIC IGNORANCE

The process of sequencing ("reading") a humans’ genome results in a huge amount of data, which should be carefully analysed and interpreted by health professionals, geneticists and genetic counsellors, in order to inform the patient (or consumer) about his genetic profile and its health-related meaningfulness. The question, whether this information is considered meaningful, desired by the patient and – most importantly – medically actionable at all, has been up for discussion for decades. During this time the “right to know” – respectively “the right not to know” (the right to remain ignorant) – about the own genetic profile and the advantages and disadvantages of both mindsets have been an apple of discord.[113]

The debate about genetic ignorance and the support of the underlying idea, by which every individual faced with possibly life-altering information about his or her own genetic profile – and consequently his or her own state of health – should have the ineradicable right to remain ignorant on such information, can be seen as being diametrically opposed to the process of autonomous decision-making, especially promoted in the context business-driven, commercial direct-to-consumer genetic testing.[114] Opponents of the idea of genetic ignorance argue against something they somehow see as an anachronistic paternalism towards patients and especially towards interested consumers. However, considering the fact that not every individual has the same level of (critical) health literacy and interest in learning something possibly undesirable in relation to his or her state of health, the right to remain ignorant about the own genetic profile, and the underlying mindset itself is well-founded. The most obvious reason to promote the idea of genetic ignorance – the right not to know – is the effort to protect individuals – often considered to be laypeople – from harmful effects of knowledge in terms of genetic information (“reasonable paternalism”).[115] Ultimately the “right” for genetic ignorance has already been considered and included in one of the most recognised legal frameworks[116], where it is stated that “the wishes of individuals not be so informed shall be observed” The subject area of genetic ignorance is surely to stay one of the most controversial subjects in the coming era of personal genomics, where personal genetic information might become easily available for almost everyone interested.

“Where ignorance is bliss, 'tis folly to be wise.”

Survey Participant - ID 471

EMPIRICAL STUDY

While the preceding part of this thesis dealt with the most important concepts and essentials on health literacy in association with genetic testing and the fundamental knowledge about the analysis of the human genome, the current and the following chapters bring the most important facts about the empirical observations made during the scientific process to light. The empirical study conducted for this thesis and its underlying hypothesis have been shortly introduced in the chapter research aims and objectives. The following chapters aim for shedding light on the most basic information of this study. First and foremost previous research and possibly existing findings will be discussed in detail, afterwards we will take a closer look at the steps that were made during the formation of the hypothesis. Thereafter the survey design and the main thoughts behind this design will be explained. Finally we will take a closer look at the recruitment method that was applied, before we dip into the results of the survey and the key findings.
8.1 PREVIOUS RESEARCH

Although the field of sociological research of applied personal genetic testing is naturally considered a rather new field of exploration there are plenty of studies with different approaches. Considering the fact that until today there is only a relative small number of people and population groups that have undergone genetic testing, it becomes clear that empirical observation in this field are not an easy task to manage. Even though almost none of the existent studies have a specific focus on health literacy they however do ask for a variety of different influencing factors, such as “perceived risks, psychological and behavioral impacts of genetic testing”[117] or “genetic risk and behavioral change”[118] in general. The most interesting study, that would also serve as an inspiration for the empirical study in hand, was a survey on “UK Public Interest in Internet-Based Personal Genome Testing”.

Given that at that time the field of commercial direct-to-consumer testing[119] had already taken off, the researchers asked for the personal opinion on such a service of a total of 4,050 “unselected adult volunteers from the UK-based TwinsUK register”.[120]

The survey questions, that asked for the personal interest in personal genetic screening, were split in three domains, namely the “awareness of personal genetic screening tests”, the “likelihood of ordering test if £250” and the “likelihood of ordering test if free”. Before answering the questions study participants were informed about the issue in the following manner:

“Since 2007 it has been possible to order a personal genetic screen over the internet. You send a sample of saliva to a commercial company who look at selected genetic markers. Based on these markers they estimate your personal lifetime genetic risk of developing around 20 common diseases (such as heart disease, Alzheimer’s disease, glaucoma or diabetes). Results are sent via an email alert to a private web-link.”[121]

Furthermore the survey would ask participants for their possible reasons to take a personal genetic screen. This section was split in a total of five sub-categories and the following options for an answer were given:

- “Encourage me to adopt a healthier lifestyle if found to be at high genetic risk of a disease”
- “Learn more about myself”
- “Convey genetic risk information to my children”
- “Doctor can monitor my health more closely”
- “Assist in financial planning for the future”

The questionnaire found that more than 93% of all survey participants agreed on one specific reason, namely to “encourage me to adopt a healthier lifestyle if found to be at high genetic risk of a disease.” This result may hint towards a general interest of the UK public for commercial personal-genome testing and is surely to have its impact on the methods of consumer-driven healthcare and prevention, even though the survey sample itself is stated not to be representative of the whole UK population. Interestingly enough, the willingness to undergo genetic testing was strongly tied to the estimated cost of the service. If the genetic test would be offered for a price of £ 250 the survey participants, who would be willing to undergo such a test in general, declined their interest in the procedure. If, however the test would be available at no cost, “nearly half of the respondents (48%) say[ing] they would be very or fairly likely to order
such a test''.[122]

“Our findings suggest that at the end of 2008 awareness of PGT among the general public was still low (13%), but this may have increased over the past year with significantly more exposure in media. […] However, the high level of interest in a free personal genetic test (48% said they were very of fairly likely to order PGT and 22% were undecided) suggests that uptake may increase when costs decrease.”[123]

Another interesting finding was highlighted, when discussing the differences in attitude between younger people (especially males) and older people (especially females) towards genetic testing. Younger people and males in general were described as the one group in the survey that would maintain a higher level of interest in the field of new health technologies, whereas older people and woman in general tend to be more skeptical when it comes to new technological developments in healthcare.

However, the study states a variety of different limitations, one of them being the already mentioned lack of representativeness of the sample. The participants of the study where part of the biggest UK adult twin registry and might – as a matter of fact – already been part of genetic studies before. Therefore, they might “be more aware of both the genetic and environmental contributions to disease than the general public”.[124] At the same time the researchers give account of the fact, that underprivileged social classes might be underrepresented in the study.

All in all the study does reveal a certain amount of public interest towards genetic screening, even though this study was limited to participants from the UK and dealt with commercial direct-to-consumer genetic testing kits, that were offered over the internet. In conclusion the study suggests that “health care systems must prepare for the potential ramifications of publicly-accessible genetic information on various aspects of health care delivery”.[125]

In contrast to the study discussed above several other approaches were used in order to find out more about the possible impact of genetic testing. Subsequently we discuss the study on “Motivations, concerns and preferences of personal genome sequencing research participants: Baseline findings from the HealthSeq project”.[126]

In this study published in the European Journal of Human Genetics researchers looked behind the motivations, concerns and preferences of 35 study participants, who had already undergone whole exome/genome sequencing, which is an extensive form of genetic analysis. When asking the 35 participants about their motivations and concerns in relation to the process of analyzing their own genetic blueprint, the researchers found eight distinct answer groups which were predicated by the participants:

· (1) “To learn personal disease risk or health-related information”
· (2) “Curiosity”
· (3) “To contribute to research”
· (4) “Interested in their ancestry”
· (5) “To provide disease risk information for their children and family members”
· (6) “Self-exploration”
· (7) “Novel opportunity”
Curiosity and the desire to obtain personal health-related information were found to be the most powerful motivators. In relation to possible concerns a third (34.3%) reported not having concerns about “receiving my personal results from whole genome sequencing”, whereas the rest of the group addressed a variety of concerns, as there are:

- “Concerns related to potential privacy issues about my data”
- “Concerns related to not knowing how I will feel about my results”
- “Concerns related to learning about my disease risk from genome sequencing”
- “Concerns related to the quality and reliability of the testing lab and the results”

One of the most evident limitations of the study was the small sample size of 35 participants. It is therefore stated, that the results “may not generalize to the general population.” It is however striking that the study examined the preferences of a group of individuals who already had close contact with the subject area. In conclusion the researchers comment that “[E]arly adopters of personal genome sequencing are unsurprisingly enthusiastic but nonetheless have concerns about how the results will affect them emotionally […]”

8.2 FORMATION OF HYPOTHESIS

The hypothesis that was constructed in this thesis is a means for examining the attitude towards and the opinion on genetic testing in relation to the personal assessment of the state of health literacy of the study participants. Considering the fact that health literate individuals tend to be more aware of health issues - both their own health and the health of others - and to have a lifestyle more adjusted to the possibilities of preventing and promoting a good health state it is stated that a health literate person is consequently more interested in undergoing a genetic test as compared to an individual with a lack of health literate behavior and attitude. Therefore the following hypothesis was established:

“Individuals with well-marked health literacy would consider genetic testing more often than individuals who are lacking a certain state of health literacy.”

Due to the immanent complexity of the concept of health literacy, where – as we learned – a distinction between the spheres of basic literacy, communicate/interactive health literacy and critical literacy is made, the hypothesis stated above is not indisputable. The reason behind that might be, that an individual with a high level of critical literacy has enough understanding and knowledge about genetic testing, so that these insights in return would put him in the situation of not wanting to undergo a genetic test because of several reasons, that seem important to him or her. Whether there are existing and well-founded concerns about the possible negative implications (genetic discrimination) or fears of learning about a genetic condition (especially non-actionable incidental findings). This problem is surely to have an impact on future decisions of whether an individual would or would not want to undergo a genetic test and can without doubt be described as a very personal mindset of the person concerned. With this in mind we want to think of another possible outcome, which is described as follows:
“Individuals with well-marked health literacy would not consider genetic testing more often than individuals who are lacking a certain state of health literacy.”

In order to find out about the attitude of the survey participants, the survey asked for their personal state of health literacy and their opinion on genetic testing. Before we dip into the findings of the survey, the survey design and the method of recruitment will be discussed subsequently.

8.3 SURVEY DESIGN

The first page of the online survey contained the basic information regarding the issue, such as three short introductory web-videos about health literacy, the process of human genome sequencing and an explanation on direct-to-consumer genetic testing. This measure was a conscious decision by the author of the study in order to make potential participants familiar with the following questions. Watching the introductory videos was however not a requirement for the completion of the online survey. A short insertion text informed about the main purpose of the survey and the approximate time, that was needed to answer the questions in the survey.

In order to learn about the specific state of health literacy of the survey participants and their attitude towards genetic testing a total of 21 questions were asked. Five questions asked for the demographic background, eleven questions tried to get to the bottom of the participants’ perception of his or her state of health literacy and another five questions dealt with their personal opinion on genetic testing and the willingness to undergo a genetic test. Subsequently the questions are listed and structured based on their main categories.

Demographic background

1. “How old are you?”
2. “What is your gender?”
3. “Where do you live?”
4. “Are you currently enrolled as a student?”
5. “What is your current level of study?” (subquestion)
The demographic background of the participants was considered an important part in order to understand differences between various socio-cultural groups. Individuals older than 14 years were considered eligible to answer the survey. Statements on locations were clustered into three sub-categories, so that participants could choose between the options “Austria”, “Outside Austria (EU)” and “Outside the European Union”. Since the educational background was considered a main issue, study participants were asked on whether they were enrolled as students or not. The question “What is your current level of study?” was constructed as a subquestion in relation to the preceding question “Are you currently enrolled as a student?” and the following answer choices were presented: (1) “Bachelor’s degree”, (2) “Master’s degree” and (3) “Other”

**Perception of personal health literacy**

- (6) “Are you familiar with the concept of health literacy?”
- (7) “I consider myself a health conscious person.” (likert scale)
- (8) “Healthy eating is important to me.” (likert scale)
- (9) “I enjoy my social life.” (likert scale)
- (10) “I am confident filling out any sort of forms - including this survey - by myself.” (likert scale)
- (11) “I do sports and/or work out every day in some way or the other.”
- (12) “I do sports and/or work out at least once a week in some way or other.” (subquestion)
- (13) “I engage in sportive activities up to a point.” (subquestion)
- (14) “Do you have regular medical check-ups or similar forms of preventive medical examination?”
- (15) “Why not?” (subquestion)

The first question on whether study participants were familiar with the concept of health literacy was listed in order to find out about their personal state of health literacy and their interest in health literacy. The answer options for the four following questions in the category “perception of own health-literacy” were designed in the form of a likert scale, where participants could choose between the options “strongly disagree”, “disagree”, “I don’t know”, “agree”, and “strongly agree”. Again, certain questions were constructed as subquestions that would be displayed only then, when the participant answered a preceding question in a particular manner. This is the case for question (12) “I do sports and/or work out at least once a week in some way or other”, for question (13) “I engage in sportive activities up to a point.” and question (15) “Why not?” in relation to the preceding question about regular medical check-ups.

**Attitude towards genetic testing**

- (16) “Are you familiar with the concept of genetic testing?”
- (17) “Were you aware of the possibility to read out your personal genome and thus learn more about your own genetic profile?”
(18) “Do you think that predictive and/or presymptomatic genetic testing should be offered to the wider public?”

(19) “Do you think that such a test should be provided only by healthcare professionals or also by private companies?”

(20) “Suppose that - after having your genes read out - you are thought to have a higher-than-average genetic risk for developing a certain disease in the future. Do you think this knowledge would encourage you to adopt a healthier respectively more adjusted lifestyle?”

(21) “After having answered the main questions of this survey: Would you consider "reading your genome", thus learning more about your own genetic profile?”

Similar to the introductory question in the section “perception of personal health literacy”, the first question when asking for the attitude towards and opinion on genetic testing was whether the participants were familiar with the concept of genetic testing at all. In a second step the questionnaire asked for the awareness about the possibility to read their genome. The following questions asked for the opinion on whether predictive genetic testing should be offered to the wider public and whether a genetic test should be provided by healthcare professionals (clinicians, researchers) or also by private companies like for example 23andMe. The question with the number (20) asked for behavioral changes after a person would undergo genetic testing and a certain risk would be found and the last question examined the general readiness to learn about the personal genetic profile.

8.4 METHOD OF RECRUITMENT

The participants of the online survey were recruited during a time period of exactly three months (11th of December – 11th of March 2015) with the assistance of the social media network Twitter and an automated mechanism, by which potential participants would receive an invitation to take the survey. This method is described as a type of non-probability sampling and referred to as convenience sampling. The most important elements of the used method will be covered in what follows.

The main unit for conducting the study was the group of social media users, who would interact in such a way, that an automated mechanism in the background would react based on their interaction and send an invitation for the online survey. For that, a variety of keywords were selected and the invitation would be triggered by the usage of these keywords during the interaction on the social media network Twitter. In order to automate this process the online service Zapier.com was used. The keywords, which would trigger an automatic invitation to participate in the survey are listed below. Keywords with the hash-symbol “#” – called Hashtags – in front of it are considered to be a common communication tool on Twitter. Hashtags are used as a sort of hyperlinked catchword in order to find more social media users that would write about the same topic.

<table>
<thead>
<tr>
<th>#healthliteracy</th>
<th>“Health Literacy”</th>
</tr>
</thead>
<tbody>
<tr>
<td>#publichealth</td>
<td>“Public Health”</td>
</tr>
<tr>
<td>#personalgenomics</td>
<td>“Personal Genomics”</td>
</tr>
<tr>
<td>#genomics</td>
<td>“Genomics”</td>
</tr>
</tbody>
</table>
“Hashtags” and Keywords used for trigger-oriented invitation to the survey

Every 15 minutes the automated mechanism would “crawl” through the Tweets of about 300 million active users[130] and send out an invitation to those, who had used one of the keyword. The invitation itself was a Tweet, which would mention the user concerned and call him or her to participate in the survey. The participants had – so to say – “opted in” to get an invitation for the survey by tweeting about one of the keywords listed above. As a matter of fact, the invited individual had to decide by himself whether he would want to participate or not.

Example for interaction by way of social media (Twitter) during the scientific process

One of the most distinctive characteristics of this recruitment method was the participants’ ability get in direct contact with the author of the study. In the same way the author was able to answer questions asked by the potential participants before or after they had answered the questionnaire (see picture above). On the other hand it was a matter of fact that this form of recruitment was prone to a self-selection bias, where only those users would be invited to take part in the study, that were somehow in a close relationship with the topic of the online survey. Nevertheless this recruitment method allowed to “harvest” an enormous amount of a total of 559 fully completed survey records.[131]

8.5 SURVEY RESULTS

Having discussed the most important issues on previous research, the formation of the hypothesis, the method of recruitment and the survey design we subsequently focus on the presentation of the survey results. Similar to the chapter “survey design”, there will be three main categories, namely demographic background, perception of personal health literacy and attitude towards genetic testing.

Demographic background

The survey had a total of 757 records (participants), of which 559 were recorded as fully completed and taken into account for the empirical analysis. The youngest participant was found to be 14 years and the oldest 85 years. Female participants were in the majority, with a total of 313 against 246 male participants.

<table>
<thead>
<tr>
<th>Female Participants</th>
<th>Male</th>
</tr>
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<td></td>
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</table>
More than half of all participants (54,38%) indicated their residence outside of the European Union, 159 participants lived "outside Austria, but within the EU" and 96 participants lived in Austria.

Only one participant out of four (24,87%) stated, that they were currently enrolled as a student, while the rest of 420 participants weren’t enrolled as a student at the time the survey was taken. Of the 139 individuals who were enrolled as students 53 participants pursued a bachelor’s degree, 36 participants a master’s degree and 50 were enrolled as another kind of student.

When being asked on whether or not the participant was familiar with the concept of health literacy a total of 438 stated "yes" and 121 participants said they were not familiar with this concept. The question on whether participants were familiar with the concept of genetic testing was answered with "yes" by 488 participants and only 71 participants stated that they were not familiar with this concept. A total of 416 out of 559 participants said that they were familiar with
both, the concept of health literacy and the concept of genetic testing.

**Perception of personal health literacy**

In order to find out more about the personal perception of the own health literacy participants were asked for their state of health consciousness, their eating behaviour, their social life, their ability to deal with (medical) forms in a confident way, sportive activities and whether they undergo medical check-ups or similar forms of preventive medical examination on a regular basis. The sample that was taken during the survey showed to have a well-marked state of health literacy when being measured by the posed questions.

**“I consider myself a health-conscious person.”**

<table>
<thead>
<tr>
<th>Degree of approval</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strongly disagree</td>
<td>10</td>
<td>1.79%</td>
</tr>
<tr>
<td>Disagree</td>
<td>28</td>
<td>5.01%</td>
</tr>
<tr>
<td>I don't know</td>
<td>12</td>
<td>2.15%</td>
</tr>
<tr>
<td>Agree</td>
<td>308</td>
<td>55.10%</td>
</tr>
<tr>
<td>Strongly agree</td>
<td>201</td>
<td>35.96%</td>
</tr>
</tbody>
</table>

More than 90% of all participants considered themselves "health-conscious" individuals, when they chose to *agree or strongly agree* on this statement made in the survey. In a similar way the sample consists mostly of individuals who state that healthy eating behaviour is a part of their lifestyle: more than 85% say that "healthy eating" is important to them.

**“Healthy eating is important to me.”**

<table>
<thead>
<tr>
<th>Degree of approval</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strongly disagree</td>
<td>12</td>
<td>2.15%</td>
</tr>
<tr>
<td>Disagree</td>
<td>27</td>
<td>4.83%</td>
</tr>
<tr>
<td>I don't know</td>
<td>22</td>
<td>3.94%</td>
</tr>
<tr>
<td>Agree</td>
<td>311</td>
<td>55.64%</td>
</tr>
<tr>
<td>Strongly agree</td>
<td>187</td>
<td>33.45%</td>
</tr>
</tbody>
</table>

The number of individuals in the sample who considered themselves *not* health-conscious is by comparison very small: only about 7% of all participants described themselves as *not* being health-conscious, a total of 12 people could neither say that they were nor that they were not health-conscious and opted for the answer option "I don't know."

The following question on whether the participants enjoy their social life yields similar results in terms of the distribution of the answers. More than 80% of all participants agree on the statement, only about 8% disagree on this matter and 46 individuals are in the dark about their social life.

**“I enjoy my social life.”**

<table>
<thead>
<tr>
<th>Degree of approval</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strongly disagree</td>
<td>16</td>
<td>2.86%</td>
</tr>
<tr>
<td>Disagree</td>
<td>31</td>
<td>5.55%</td>
</tr>
<tr>
<td>I don't know</td>
<td>46</td>
<td>8.23%</td>
</tr>
<tr>
<td>Agree</td>
<td>310</td>
<td>55.46%</td>
</tr>
</tbody>
</table>
The statement about the perception of the own confidence when filling out forms - including the survey that was filled out by the participants - was undoubtedly self-referential. Nevertheless a total of 19 participants or approximately 3% said that they were not confident when filling out such forms, whereas more than 90% of all participants said that they feel confident when filling out any sort of forms.

“I am confident filling out any sort of forms – including this survey – by myself.”

<table>
<thead>
<tr>
<th>Degree of approval</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strongly disagree</td>
<td>12</td>
<td>2.15%</td>
</tr>
<tr>
<td>Disagree</td>
<td>7</td>
<td>1.25%</td>
</tr>
<tr>
<td>I don’t know</td>
<td>25</td>
<td>4.47%</td>
</tr>
<tr>
<td>Agree</td>
<td>168</td>
<td>30.05%</td>
</tr>
<tr>
<td>Strongly agree</td>
<td>347</td>
<td>62.08%</td>
</tr>
</tbody>
</table>

Participants were asked for regular sportive activities and the amount of time they spend with such activities. About 30% of all participants stated that they do a lot of sports, which means the work out every day of the week in some way or the other.

“I do sports and/or work out every day in some way or the other.”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>163</td>
<td>29.16%</td>
</tr>
<tr>
<td>No</td>
<td>396</td>
<td>70.84%</td>
</tr>
</tbody>
</table>

For the rest of the sample that did not work out on a daily basis a subquestion was asked, where participants were able to report whether they work out at least once a week in some way or the other. Half of the remaining sample answered with “yes” and the following subquestion revealed that about 10% of all participants did not engage in sportive activities at all.

“I do sports and/or work out at least once a week in some way or the other.”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>280</td>
<td>50.09%</td>
</tr>
<tr>
<td>No</td>
<td>116</td>
<td>20.75%</td>
</tr>
<tr>
<td>Not displayed (subquestion)</td>
<td>163</td>
<td>29.16%</td>
</tr>
</tbody>
</table>

As a matter of fact the results regarding the sportive activities of the survey participants demonstrate another evidence for a rather health-literate respectively health-conscious sample, where only 53 out 559 individuals said, that they were absolutely not engaging in sportive activities of any sort.

“I engage in sportive activities up to a point.”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>63</td>
<td>11.27%</td>
</tr>
</tbody>
</table>
The last question in the category on the perception of the participants' state of health literacy asked for regular medical check-ups or similar forms of preventive medical examination and whether participants had such check-ups. More than 70% of the sample said that they were indeed having regular medical check-ups, whereas only about 30% said that they were not. These 163 individuals were asked why they did not have medical examinations on a regular basis and a variety of different answers emerged, ranging from “no time” to “I’m just not bothered” and “my insurance doesn’t cover it”. [132]

“Do you have regular medical check-ups or similar forms of preventive medical examination?”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>396</td>
<td>70.84%</td>
</tr>
<tr>
<td>No</td>
<td>163</td>
<td>29.16%</td>
</tr>
</tbody>
</table>

Attitude towards genetic testing

The category that asked for the participants' attitude towards genetic testing consisted of a total of six questions and one of them – whether participants were aware of the concept of genetic testing - has been shortly discussed in the first category “demographic background”. Subsequently the other five questions and their results will be discussed in detail.

“Were you aware of the possibility to read out your personal genome and thus learn more about your own genetic profile?”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>466</td>
<td>83.36%</td>
</tr>
<tr>
<td>No</td>
<td>93</td>
<td>16.64%</td>
</tr>
</tbody>
</table>

When asking the question on whether the participants knew about the possibility to read their own genetic profile, more than 80% answered with “yes”. A total of 93 individuals in the sample were unaware of such a procedure. Also, more than 75% of all participants think that predictive genetic testing should be offered to the wider public as against 134 out of 559 individuals said they don’t think that such a test should be offered to the general public.

“Do you think that predictive and/or presymptomatic genetic testing should be offered to the wider public?”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>425</td>
<td>76.03%</td>
</tr>
<tr>
<td>No</td>
<td>134</td>
<td>23.97%</td>
</tr>
</tbody>
</table>

“Do you think that such a test should be provided only by healthcare professionals or also by private companies?”

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<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Healthcare professionals only.”</td>
<td>214</td>
<td>38.28%</td>
</tr>
<tr>
<td>“Both healthcare professionals and private companies.”</td>
<td>189</td>
<td>33.81%</td>
</tr>
<tr>
<td>“I don’t know.”</td>
<td>22</td>
<td>3.94%</td>
</tr>
<tr>
<td>Not displayed (subquestion)</td>
<td>134</td>
<td>23.97%</td>
</tr>
</tbody>
</table>

The next question on whether a genetic test should be either provided by healthcare
professionals only or by both, healthcare professionals and private companies, was almost half split: 214 out of 559 participants said only healthcare professionals should be allowed to provide genetic testing, whereas 189 individuals said both parties should be allowed.

A small number of individuals stated that they did not know by which parties the test should be offered and 134 out 559 participants were excluded from this question based on their answer in the preceding question. The question on whether the results from a genetic test would possibly encourage individuals to adopt a healthier lifestyle in order reduce additional risk of developing a certain disease, was answered with “yes” by the majority of the sample: more than 80% think that a higher-than-average genetic risk, found in the process of a genetic test, would trigger a change in behaviour.

"Suppose that – after having your genes read out – you are thought to have a higher-than-average genetic risk for developing a certain disease in the future. Do you think this knowledge would encourage you to adopt a healthier respectively more adjusted lifestyle?"

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<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>453</td>
<td>81.04%</td>
</tr>
<tr>
<td>No</td>
<td>106</td>
<td>18.96%</td>
</tr>
</tbody>
</table>

The last question of the survey asked for the personal interest in a genetic tests and more than 60% of all participants said that they would be interested in learning about their own genetic information by undergoing a genetic test. About 20% stated that they are however not interested in genetic testing and about 18% stated that they were still indecisive and had not made up their mind on this decision yet.

“After having answered the main questions of this survey: Would you consider ‘reading your genome’, thus learning more about your own genetic profile?”

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>115</td>
<td>20.57%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>342</td>
<td>61.18%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>102</td>
<td>18.25%</td>
</tr>
</tbody>
</table>

All in all, it can be said that the sample that was drawn during the scientific process tends to be a generally more health literate or health-conscious one. The participants’ attitude towards genetic testing is mostly affirmative. However, the results show that contrary opinions on genetic testing do exist and that a portion of the sample is still unaware of all concepts and issues addressed in the survey. In order to find a possible confirmation for the hypothesis statement the following chapter looks for key findings of the survey.

8.6 KEY FINDINGS

While the results of the survey provide a comprehensive overview of the sample and the characteristics of those individuals who have participated in the survey, the hypothesis itself has not been examined yet. Based on the available data aggregated during the scientific process this chapter looks for possible answers on whether or not individuals with well-marked health literacy would consider genetic testing more often than those individuals who are lacking a certain state of health literacy. It is of the utmost importance to add, that an individuals’ state of health literacy in this thesis has been measured by a number of various questions that – when
answered in a certain manner – would give the opportunity to classify their behavior as “health literate” or health conscious.

However, those questions of the survey, which would ask for parameters regarding the level of health literacy focus mainly on fundamental health behavior like healthy eating or sportive activities, and are as such considered to bring the participants’ level of basic health literacy into question. The aspect of critical health literacy and the level of the skills associated with critical health literacy, was not brought into question explicitly.

**Hypothesis**

“Individuals with well-marked health literacy would consider genetic testing more often than individuals with lacking a certain state of health literacy.”

In order to find a possible answer for the hypothesis statement the analysis of the data strives for building several intersections and clusters between those participants who – based on their answers – are considered to have well-marked competencies and/or personal skills regarding health literacy and a positive attitude towards genetic testing at the same time. In the same way the analysis looks for those participants who – based on their answers – are considered to be less health literate and still willing respectively interested in undergoing genetic testing. Ultimately the ratios in the space of the two intersections are seen as a way to hint at a possible answer for the hypothesis.

**Cluster analysis 1**

As a first step the results of four questions in the category “perception of personal health literacy” - namely question (7), (8), (9) and (10) - will be intersected with one question from the category “attitude towards genetic testing”, which is the question with the number (21). Because they are considered to indicate a well-marked state of health literacy, the questions from the category “health literacy” that have been answered with the answer options “agree” and “strongly agree” serve as a first basic quantity or cluster. The first data inquiry shows that a total of 406 out of 559 participants answered the four mentioned questions in the category “health literacy” in a way, which makes them a part of the first basic quantity of individuals with well-marked health literacy. At the same time 255 participants out of those 406 form a cluster in which they also stated their general interest in genetic testing. In other words a total percentage of 62.81% of all participants from the first basic quantity, who are considered having well-marked health literacy skills by the terms of this inquiry, would consider to undergo a genetic test.

**Cluster 1 with well-marked health literacy (406 members)**

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>77</td>
<td>18.97%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>255</td>
<td>62.81%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>74</td>
<td>18.23%</td>
</tr>
</tbody>
</table>
In order to compare the group of people that are considered to have well-marked health literacy skills and the group of people with lack of health literacy a data inquiry for two separate partial quantities has been made. The first cluster consists of all people with well-marked health literacy skills irrespective of their attitude towards genetic testing totalling in 406 members (participants). The second quantity consists of all people with weak health literacy skills derived from their answers given in the category “perception of personal health literacy” totaling in 15 members.

Cluster 2 with weak health literacy skills (15 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>4</td>
<td>26.67%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>7</td>
<td>46.67%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>4</td>
<td>26.67%</td>
</tr>
</tbody>
</table>

The first cluster analysis compared two clusters from the dataset, which have been introduced above. The first cluster consists of 406 participants, who are defined as having well-marked health literacy skills. As seen in figure below almost two out of three from this group was generally interested in undergoing genetic testing, whereas one out of five (19%) was not interested. The rest of the participants in this cluster were still indecisive about the issue.

Cluster 1 (well-marked health literacy)

Cluster 2 (weak health literacy skills)

Cluster analysis 2

The second cluster consists of a total of 15 participants, who are defined to have weak health literacy skills based on the first inquiry. The conditions for this data inquiry were set in way, that only those participants, who answered the questions (7), (8), (9) and (10) with “strongly disagree”, “disagree” or “I don’t know”, would show up in this cluster. About one in two members from this cluster state that they would be interested in genetic testing, whereas about one out of four said they are not interested or still indecisive about genetic testing.

Cluster analysis 2

The second data inquiry looked for the state of the participants’ health literacy based on their answers given on question (11), which asked for daily sporting activities. A total of 163 out of
559 individuals stated that they were “doing sports and/or working out every day in some way or the other”. This group of individuals is considered to have well-marked health literacy in the second cluster analysis. A two-thirds majority of this cluster was generally interested in genetic testing, whereas about 16% were either not interested or indecisive.

**Cluster 3 with well-marked health literacy (163 members)**

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>27</td>
<td>16.56%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>110</td>
<td>67.48%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>26</td>
<td>15.95%</td>
</tr>
</tbody>
</table>

As opposed to this a total 396 out of 559 participants said that they were not engaging in sporting activities on a regular daily basis. This group of individuals is considered to have at least weaker health literacy than the first group in this cluster analysis. Nevertheless the majority of about 59% of all members of this cluster said that they were interested in genetic testing, whereas the rest of the individuals who were not interested or indecisive were almost evenly distributed.

**Cluster 4 with weak(er) health literacy skills (396 members)**

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>88</td>
<td>22.22%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>232</td>
<td>58.59%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>76</td>
<td>19.19%</td>
</tr>
</tbody>
</table>

The group of people who – based on the conditions of this data inquiry – had shown the weakest state of health literacy by answering the subquestion (13) with “no”, got similar results in regards of their interest in genetic testing: 30 individuals (56.60%) said they were interested in genetic testing, 16 individuals (30.19%) were not and seven individuals (13.21%) were indecisive.

**Cluster analysis 3**

The third cluster analysis focusses on question (14), when participants were asked on whether or not they had regular medical check-ups or similar forms of preventive medical examinations. In this particular case all participants who answered with “yes” were considered to have well-marked health literacy and those who answered with “no” represent the group of individuals with...
ocluster 5 with well-marked health literacy (396 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>74</td>
<td>18.69%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>251</td>
<td>63.38%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>71</td>
<td>17.93%</td>
</tr>
</tbody>
</table>

Again, more than two-thirds of all individuals from this cluster said that they were interested in genetic testing and about 18% were either not interested or still indecisive. In contrast to this, the cluster of people who did not have regular medical check-ups consisted of 163 individuals and were – based on the conditions of this data inquiry – considered to have weak health literacy skills. The results regarding the personal interest in genetic testing are similar in kind: the majority of this cluster stated that they would be interested in genetic testing, whereas one out of four said that they were not interested and 19% had not made up their mind on this issue.

ocluster 6 with weak health literacy skills (163 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>41</td>
<td>25.15%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>91</td>
<td>55.83%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>31</td>
<td>19.02%</td>
</tr>
</tbody>
</table>

The next cluster (cluster analysis 3) was formed by adding an additional condition to the data inquiry. This condition was represented by the question (20) and introduced a possible behavioural attitude, when having a genetic test. Participants should state whether or not they would adopt a healthier lifestyle when being exposed to a "higher-than-average genetic risk for developing a certain disease in the future". For the first time, the results shown in this cluster regarding the personal interest in genetic testing happen to be diverging from the preceding results so far.

ocluster analysis 3

The intersection between individuals who did have regular medical check-ups and – at the same time – were confident that a higher-than-average genetic risk provided by the results of a genetic test would encourage them to adopt a healthier and more adopted lifestyle showed primarily the same trends as the other clusters. More than 70% of all individuals of this cluster (329 members) said they were interested in genetic testing.

ocluster 7 with well-marked health literacy and confidence in
In contrast to this, the next cluster was build out of individuals who – like the preceding cluster – did have regular check-ups but, however, did not think that a higher-than-average genetic risk would encourage them to adopt a healthier lifestyle. Almost half of all individuals (67 members) from this cluster said that they were not interested in genetic testing and out of four said that they were either not interested or indecisive.

Cluster 8 with well-marked health literacy and non-existing confidence in personal behavioural change based on results from genetic testing (67 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>32</td>
<td>47.76%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>18</td>
<td>26.87%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>17</td>
<td>25.37%</td>
</tr>
</tbody>
</table>

For the first time the cluster analysis within cluster 8 lead to a different result, where almost half of all participants from this specially formed cluster stated that they were not interested in genetic testing. While the meaningfulness of this result is definitely open to debate, the next cluster with new conditions yields similar results. For this, the data inquiry asked for those individuals, who did have regular medical check-ups and – at the same time – objected to the idea of offering predictive and/or presymptomatic genetic testing to the wider public. Subsequently we discuss these results of the fourth cluster analysis.

Cluster analysis 4

Cluster 9 with well-marked health literacy and against the idea of offering predictive and/or presymptomatic genetic testing to the wider public (91 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>43</td>
<td>47.25%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>26</td>
<td>28.57%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>22</td>
<td>24.18%</td>
</tr>
</tbody>
</table>

Cluster 9 consists of a total of 91 individuals, is based on the conditions described above and showed results, which were similar to the preceding cluster. People with regular medical check-ups or similar forms of preventive medical examination, who were against the idea of offering predictive and/or presymptomatic genetic testing to the wider public are considered to be the basic quantity in this cluster. Almost half of all individuals from this group state that they are not interested in genetic testing, about 29% say they are interested and about 24% were still indecisive.

One last cluster was built by asking for individuals with weak health literacy (based on the
conditions of the data inquiry). This cluster included those individuals who did not have regular medical check-ups and who – at the same time – were against the idea of offering predictive or presymptomatic genetic testing to the wider public.

Cluster 10 with weak health literacy and against the idea of offering predictive and/or presymptomatic genetic testing to the wider public (43 members)

<table>
<thead>
<tr>
<th>Answer</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Not interested.”</td>
<td>26</td>
<td>60.47%</td>
</tr>
<tr>
<td>“Interested.”</td>
<td>8</td>
<td>18.60%</td>
</tr>
<tr>
<td>“Indecisive.”</td>
<td>9</td>
<td>20.93%</td>
</tr>
</tbody>
</table>

The results in this cluster were characterized by a majority of individuals who were generally not interested in genetic testing. Considering the conditions for this data inquiry the results, however, come as no surprise: individuals, who tend to be against population wide genetic testing – for whatever reason – may tend to be not interested in genetic testing for themselves in the first place.

CONCLUSION

The sample drawn during the scientific process shows to have characteristics which indicate that the majority of the participants consider themselves as health literate respectively health conscious. Given that the attitude towards genetic testing was affirmative for the predominant part, it can be stated that individuals with well-marked health literacy would generally rather consider genetic testing than being generally opposed to such a procedure. However, due to the fact that only a small proportion of the sample gave answers which would indicate a generally low state of health literacy - and even these partial quantities showed to be rather willing to undergo genetic testing than being opposed to it - the data analysis of a larger group of individuals with assumed lack of health literacy in relation to their attitude towards genetic testing - which by definition would possibly lead to falsification or to evidence in favour of the stated hypothesis - was not possible. In this context it is important to add that the recruitment of participants who would consider themselves as being rather not health literate has to be considered as a difficult task, especially when the particular recruitment method applied in this study is taken into account. Social media users, who served as a basic quantity for this sort of social research are possibly inscrutable in the way they behave and it is difficult to determine whether the given answers can truly be considered to be a reflection of their real behaviour. Given that a satisfactory validation of the insufficient data in relation to the stated hypothesis is not possible within the scope of this thesis, further research is recommended.

8.7 Limitations of the Study

Even though the results revealed interesting tendencies, opinions and attitudes in relation to participants’ level of health literacy and genetic testing, the scientific method applied in order to find a possible evidence for the confirmation or falsification of the stated hypothesis can be considered as inadequate. First and foremost the recruitment method applied in the study led to a possible self-selection bias, by which individuals would somehow select themself into the group of participants. Due to this sampling method - referred to accidental or convenience
sampling - individuals may have had a close relationship to the issue of health literacy and/or genetic testing, leading to potentially onesided tendencies. For the same reason (method of recruitment) the results presented in this study are not representative. Further research is needed.

DISCUSSION

The thesis in hand is an attempt to merge two distinct issues with the aim to stress the importance of both topics in relation to one another. The concept and the possible implications of personal genetic testing cannot be completely understood without the basic understanding of the concept of health literacy. In addition to that health literacy itself is not in the least to be considered as simple adherence to simplistic guidelines promoting a healthy lifestyle, but influenced by a complex interaction of environmental, social and personal factors and differentiated in diverging intermediate stages (functional, interactive and critical health literacy).

The exceptional feature of genetic testing, and the potential wish to undergo genetic testing for the purpose of health-related decision-making lies in the predictive character of such a process. However, the luring promise of knowing about a future disease without actually suffering from the specific disease at the present moment in time becomes less attractive when the possible consequences, such as the psychological burden or the lack of power to act when faced with difficult-to-treat conditions, become apparent. With genomic sequencing technologies and services for genetic analysis and interpretation becoming more and more democratised and potentially independent from professional supervision, it remains open whether genetic literacy – the knowledge about the personal genomic profile – will become a crucial part of the personal state of health literacy of every individual.

The right to remain ignorant about one’s genetic profile might undeniably become subject to pressure, when the interpretation and analysis of the human genome becomes even more sophisticated and available in the near future. The mere status of not knowing about the health information purported by the data derived from one’s genome could be seen as a form of inconsiderate carelessness towards the own health status and possibly even towards the health status of others (offspring). On the other hand, the mere access to genetic testing technologies might be limited to socially and therefore economically advantaged populations, leaving disadvantaged groups behind and thereby creating a new, contrasting form of genetic discrimination. Whatever the outcomes of future developments might be, the professional management of every aspect associated with genetic information derived from patients and all interested individuals is imperative.

LIST OF LITERATURE*

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28. Fourth amendment of the Austrian Gene Technology Act,

29. ("Änderung des Gentechnikgesetzes, BGBl. I Nr.127/2005)


36. Pearson, Manolio; "How to interprete a genome-wide association study", JAMA Vol. 299(11), p. 1335 - 1344, 2008 [footnote 60, 61]


68]

44. Venter et al., "The Sequence of the Human Genome", Science Vol. 291, 2001 [footnote 69]

45. Church, "The Personal Genome Project", Mol Syst Biol. v.1, 2005 [footnote 70]


86. Sanderson et al., "Motivations, concerns and preferences of personal genome sequencing participants: Baseline findings from the HealthSeq project" Eur J Hum Genet., 2015 [footnote 127, 128, 129]


· listed according to the chronological order of their appearance, including graphic representations