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EXPECTATIONS, FRAMES AND PRACTICES OF GENETIC COUNSELLING IN DIFFERENT CONTEXTS OF GENETIC TESTING

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To my family

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Expectations, frames and practices of genetic counselling in different contexts of genetic testing

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Abstract

Genetic counselling is a process in which the counsellee receives information and support concerning a genetic disease. This study examines the genetic counselling attached to genetic testing. Since genetic information is increasing alongside new testing technologies and the situations faced at the genetic clinics will therefore be more diverse, it is essential to assess what the expectations directed at genetic counselling are. It is also important to compare how they face the current counselling practices.

In this study, the expectations, frames and practices of genetic counselling in different contexts of genetic testing were examined from three different perspectives: First, international guidelines covering genetic counselling were analysed to summarise what is expected from genetic counselling and to study how genetic information is framed. Second, national experts in European countries were asked about the regulations and practices of genetic counselling in their country. Finally, ten counsellees who had visited a genetic clinic were interviewed to analyse their expectations and experiences. The counsellees' perspective was also approached through the background review of the previous studies on counsellees' experiences.

On the basis of the study, there are basic elements that are expected to be covered in genetic counselling from all perspectives. However, the views concerning bioethics, genetic exceptionalism and psychosocial aspects vary depending on the perspective and on the individual situation. Since there are sometimes more differences than similarities between genetic tests, no universal recommendations for counselling can be applied. The practices of genetic counselling should be defined situationally, emphasising the individual needs over the genes.

Keywords: Genetic counselling, genetic testing, expectations, frames, practices, experiences, genetic information

Elina Rantanen

Perinnöllisyysneuvontaan liittyvät odotukset, kehykset ja käytännöt geenitestien erilaisissa konteksteissa

Turun yliopisto, Lääketieteellinen tiedekunta, Biolääketieteen laitos, Lääketieteellinen biokemia ja genetiikka, Turun molekyylilääketieteen tohtoriohjelma (TuDMM)

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Tiivistelmä

Tässä työssä käsitellään geenitestauksen yhteydessä tapahtuvaa perinnöllisyysneuvonta, jossa neuvottava saa tietoa ja tukea geneettiseen sairauteen tai vammaan liittyen. Testausteknologioiden kehittyessä geneettinen tieto lisääntyy ja perinnöllisyysklinikoilla kohdattavat tilanteet moninaistuvat. Siksi on tärkeää arvioida, minkälaisia perinnöllisyysneuvontaan kohdistuvat odotukset ovat ja miten ne vastaavat perinnöllisyysneuvonnan nykyisiä käytäntöjä.

Tässä tutkimuksessa tarkasteltiin perinnöllisyysneuvontaan kohdistettavia odotuksia sekä sen kehyksiä ja käytäntöjä kolmesta eri näkökulmasta. Perinnöllisyysneuvontaa käsittelevien kansainvälisten ohjeistojen analyysilla selvitettiin, mitä ideaalilta perinnöllisyysneuvonnalta odotetaan ja miten geneettistä tietoa kehystetään. Kansallisilta asiantuntijoilta kysyttiin Euroopan maiden perinnöllisyysneuvontaan liittyvästä lainsäädännöstä ja käytännöistä. Lopuksi kymmentä perinnöllisyysneuvonnassa käynyttä neuvottavaa haastateltiin heidän odotuksistaan ja kokemuksistaan. Neuvottavien näkökulmaa tarkasteltiin lisäksi aiempien neuvottavien kokemuksia käsittelevien tutkimusten tausta-analyysilla.

Tutkimuksen perusteella perinnöllisyysneuvonnalta odotetaan tiettyjä peruselementtejä kaikista näkökulmista tarkasteltuna. Bioetiikkaa, geneettistä ekseptionalismia ja psykososiaalisia tekijöitä koskevat näkemykset kuitenkin vaihtelevat riippuen perspektiivistä ja yksittäisestä tilanteesta. Koska geenitestien välillä on toisinaan enemmän eroavaisuuksia kuin yhtäläisyyksiä, universaaleja suosituksia ei kannata soveltaa kaikessa neuvonnassa. Perinnöllisyysneuvonnan käytännöt pitäisi määritellä tilanteisesti ja perustella geenien sijaan yksilöllisillä tarpeilla.

Avainsanat: Perinnöllisyysneuvonta, geenitestaus, odotukset, kehykset, käytännöt, kokemukset, geneettinen tieto

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List of original publications

This doctoral thesis is based on the following original articles referred to in the text by their Roman numerals I-IV.

I Rantanen E, Hietala M, Kristoffersson U, Nippert I, Schmidtke J, Sequeiros J, Kääriäinen H: What is ideal genetic counselling? A survey of current international guidelines. European Journal of Human Genetics 2008(16): 445-452.

II Rantanen E, Pöntinen S, Nippert I, Sequeiros J, Kääriäinen H: Expertise, empathy and ethical awareness: ideals of genetic counselling based on framing of genetic information in international guidelines. New Genetics and Society 2009(28): 301-316.

III Rantanen E, Hietala M, Kristoffersson U, Nippert I, Schmidtke J, Sequeiros J, Kääriäinen H: Regulations and practices of genetic counselling in 38 European countries: the perspective of national representatives. European Journal of Human Genetics 2008(16):1208-1216.

IV Rantanen E, Hietala M, Kääriäinen H: From a prerequisite for a gene test to important support. A frame analysis of genetic counseling in counselees' interviews. Submitted.

1. Introduction

The world of genetics has always fascinated people. The typical characteristics of the family are eagerly sought in every newborn, and, on the other hand, genetic manipulation, cloning and selecting foetuses on basis of genetic information provoke fears and suspicions and provide inspiration for science-fiction. Since the early 20th century, genetics has served the purposes of medicine; in medical genetics, genetic information is used to study medical conditions that are or may be genetic. This information is gained via clinical examination of the patient, including family history, but recently more and more often by a genetic test. Usually, when a genetic test is performed, the meanings and the implications of genetic information are discussed with the person in a process called genetic counselling. As there is a diversity of different genetic tests, genetic counselling is offered in many different contexts: to an affected patient or parents of an affected child, a healthy person considering predictive or carrier testing on the basis of the family history, or a couple who are expecting a child or planning a family and have a risk of having an affected child.

Genetic counselling in all these diverse situations includes similar elements relating to the appropriate information and support needed. The most often cited definition of genetic counselling was published in the American Journal of Medical Genetics in 1974:

> Genetic counselling is a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained person to help the individual or the family to comprehend the medical facts, including the diagnosis, the probable course of the disorder and available management, to appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives, to understand the options for dealing with the risk of recurrence, to choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision, and to make the best possible adjustment to the disorder in an affected family member and to the risk of recurrence of that disorder. (Fraser 1974.)

Newer definitions of genetic counselling have also been approved. The European Society of Human Genetics defined genetic counselling in 2003 in a rather similar way to the Amercian Society in 1974:

> Genetic counselling is a communication process, which deals with the occurrence, or risk of occurrence, of a genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to 1) understand the medical facts of the disorder; 2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives; 3) understand the options of dealing with the disorder; 4) choose the course of action which seems appropriate to them in the view of their risk and their family goals and act in accordance with that decision; and 5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. In addition, genetic information may be given by a range of healthcare practitioners in the context of genetic testing or screening. (Godard et al. 2003.)

In consideration of the need for a new definition of genetic counselling in parallel with the development of genetic testing and the associated changes in the practice of counselling, the (US) National Society of Genetic Counsellors defined genetic counselling in 2005 as follows:

> Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:
>
> - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
> - Education about inheritance, testing, management, prevention, resources and research.
> - Counselling to promote informed choices and adaptation to the risk or condition. (Resta et al. 2006.)

The development of genetic medicine has been rapid. The advance of molecular genetic testing in laboratories has led to the introduction of tests for hundreds of genetic disorders (OECD 2000, Aymé and Rodwell 2013). As medical research has increasingly focused on the genetic basis of diseases and the use of genetic tests in clinical medicine has simultaneously increased (Ibarreta et al. 2003), so has the need for counselling associated. Thus, it is interesting and important to consider what these services are like today, what they are expected to be, how they are expected to develop, what kinds of conceptions of genetic information lie behind the ideals of genetic counselling, and how this all affects people living in the world of increasing genetic information. The aim of this dissertation was to bring together different perspectives in the field of genetic counselling. The purpose was to study what is expected of genetic counselling, and to analyse what the conceptions of genetic information behind those expectations are like. Another purpose was to study how the expectations correspond to the current practices of genetic

counselling in Europe. The study was part of the EuroGentest Network of Excellence that aimed at improving and harmonising the quality of genetic testing, including the quality of the genetic counselling services associated with it (see www.eurogentest.org). Genetic services develop rapidly, and the European professionals have recognised the need for a common discussion. This study served as one basis for the discussion and for the development of new recommendations.

2. Literature review

2.1 Development of genetic counselling

2.1.1 The eugenic history of genetic counselling

Human genetics is a rather young field of science. Although the first descriptions of inherited conditions were made in the 18th century, it was only in the early 20th century that scientists began the in-depth study of modern genetics. In the 19th century, the Austrian monk Gregor Mendel worked on plant hybridisation, which is regarded as pioneering in the development of human genetics. Mendel's work proved to have such general relevance to medical genetics that it is often regarded as its foundation. His work was re-discovered in the early 20th century when the development of modern medical genetics began. (Harper 2008, Resta 2005, Skirton and Patch 2002.)

On the basis of separate discoveries in human genetics in the first half of the 20th century, medical genetics started to become a defined speciality in the middle of the 20th century, and by the 1980s, it was a rapidly developing field of medicine in North America and Europe (Harper 2008). Genetic advice has been provided to families for centuries (Begleiter 2002). Genetic counselling, as it is known today, began to emerge in accordance with the development of medical genetics. In 1955, when Sheldon Reed's book *Counselling in Medical genetics*, the first of its kind, appeared, there were 13 genetic counselling clinics in North America, and by the 1970s, genetic counselling had already been recognised as an important component of medical genetics in many countries (Harper 2008).

Families' reasons for seeking genetic counselling were in the first counselling clinics partly the same as they are today, for example understanding the causes and risks concerning mental handicaps and Huntington's disease, but there were also differences: some of the conditions have been reduced by preventive measures, and one of the most common reasons for visiting a genetic clinic in the USA in 1950s, skin colour, has disappeared from genetic counselling (Harper 2008, Reed 1955). In addition to the technological development, the greatest change has been the increased number of options from which to choose. During the later decades of the 20th century, the development of medical genetics affected the elements of genetic counselling: accurate diagnosis, risk estimation and pedigree details became the essential cornerstones of genetic counselling. In the 1970s, communication and psychological elements were emphasised. The counselling

sessions became long in comparison with other clinical specialities, and it included a letter summarising the consultation. These practices have dominated genetic counselling for decades. (Harper 2008, Resta 2005.)

Early genetic counselling was closely linked to the eugenic movement: the objective of counselling was to improve the society through racial hygiene. The eugenic movement aimed at improving human qualities through directed breeding practices. Its rise was connected to the idea that genetic knowledge could be used to eliminate social problems. The first person to approach eugenics systematically was Francis Galton, who had made a strong contribution in the development of the study of inheritance. He first used the term 'eugenics' in 1865 when establishing whether mental characteristics were largely based on heredity. At the end of the 19th century, eugenics was turned into action: people with high social standing and intelligence were encouraged to increase their family sizes to increase the "good traits" in society. (Harper 2008, Walker 2010, Resta 2005, Raz 2009, Clarke 1997, Galton 1909.)

Eugenics was practised in several different forms, from obligatory sterilisations to more sophisticated practices to encourage middle class to have more children (Kerr and Shakespeare 2002). It was felt that by encouraging people with "good" traits to have more children and by encouraging people with "bad" traits to not have children, social problems could be solved (Resta 2005). Eugenics was practiced in many countries including the USA and Nordic countries, but the movement became world-known by Hitler and the Nazi party. In the 1920s, even the supporters of the eugenic movement began to have suspicions concerning the scientific and the social inadequacies of eugenics, but despite this, the greatest abuses committed in its name appeared only later. Since the horrors of Nazi Germany, eugenics has been opposed strongly. (Harper 2008, Resta 2005.)

The post-war generation of human geneticists and generations afterwards have been strongly anti-eugenic (Harper 2008). As the eugenic goals of genetic counselling began to disappear the interests of the families became central to the counselling sessions (Resta 2005). Counselling changed in the 1960s and 1970s, not only due to increasing medical knowledge, but also because of the need to dissociate it from eugenics: medical, technological, professional and political factors led to the status of non-directive genetic counselling (Weil 2003). However, it is still discussed whether genetic counselling can ever be free from eugenics. It may not be eugenic purposely, but it has been argued that eugenics is always an essential character of it (Kerr and Shakespeare 2002).

The eugenic history of genetic medicine in Western countries reflects on the discussion on today's genetic counselling. Many counselling situations relate to

reproduction, most directly those in the context of prenatal genetic testing. In addition, carrier testing is usually performed in order to determine the risk status of any future offspring. In some cases, diagnostic and predictive genetic testing may affect the counsellee's decision on having children, too. Since reproductive decisions essentially belong to the realm of genetic counselling, discussing the selection of "good" and "bad" traits in the context of genetic testing has not disappeared, although it has changed its nature.

2.1.2 Regulating genetic counselling

The need to dissociate genetic counselling from its eugenic past has led to the development of ethical principles and regulations for genetic testing. The salient ethical principles include the right to autonomy, justice, beneficence and non-maleficence as in all medical practice (Beauchamps and Childress 2001). There are also regulations particularly applied to genetic counselling that emphasise, for instance, the neutral presentation of the available options and the individual choice (WMA 2009, UNESCO 2003, Meincke 2001). These regulative documents have been criticised as the production of representatives of certain professions only and as disappointing because of their balancing role between different interests (Kerr and Shakespeare 2002). However, these kinds of guidelines have been considered necessary to fill the gaps in the legislation of genetic testing because the field of genetics is developing more rapidly than the legislation, and as legislation might not be the solution that is best suited to all issues in genetic testing and counselling (Lötjönen 2004, Rantanen et al. 2008a).

Although the ideals of genetic counselling are expressed rather uniformly in international guidelines (see Rantanen et al. 2008a), they may be applied differently in different countries. In Wertz and Fletcher's studies (2004, 1988), the autonomy of the counsellee, the non-directive approach, confidentiality, and the privacy of genetic information were emphasised more in the English-speaking nations and in Western Europe than anywhere else. Furthermore, the variation in the training of the providers of genetic counselling has been seen to lead to differences in the practices among countries (Biesecker and Marteau 1999, Rantanen et al. 2008b).

The practices of genetic counselling have been argued to differ even within countries. The best practices have been said to be in use in the main centres, but not all centres in the same country operate in the same manner (Godard et al. 2003, Rantanen et al. 2008b). There are laboratories that do not have direct links to counselling services, although it is not recommended that they be separated from the testing procedure (Ibarreta et al. 2003, Rantanen et al. 2008b). In many countries, there are also local factors that limit

access to genetic services, such as geography, religion, healthcare system, or the lack of understanding of genetics (Harris 1997).

2.1.3 Different models of genetic counselling

Non-directiveness has served as a central ethos for genetic counselling at least since the 1970s, providing both practical and ethical guidance to counsellors (Weil 2003, Bartels et al. 1997). It promotes the autonomy and the personal control of the client (Biesecker 2000), and there are several reasons why genetic counsellors wish to claim that their work is non-directive. Respecting the patient's autonomy is a dominant principle in healthcare in general (Beauchamps and Childress 2001), there is also a will to distance the present from the past of eugenics, and finally, the counsellors may not want to commit to the decisions of the patients (Kessler 1997, Clarke 1997, Elwyn et al. 2000, Weil 2003, Rantanen et al. 2008a).

However, non-directiveness is not always supported as the best model of genetic counselling. Counsellees may not accept non-directiveness – they may interpret it as indifference and they may not wish to make decisions on their own (Shiloh 1996, Furu et al. 1993). The counsellor may be asked to become a facilitator in decision-making instead of being just an information provider. It has been argued that non-directiveness limits the counsellor's ability to effectively serve the counsellee (Weil 2003, Biesecker 2000, Kessler 1997, Rantanen et al. 2008a).

Non-directiveness derives from the fear of eugenics and has originally been applied in reproductive genetic counselling. Therefore, it has been questioned whether it is the best counselling model in other areas of genetic counselling in which medical recommendations may be essential, such as cancer risk counselling (Clarke 1997, Trepanier et al. 2004, Biesecker 2001). It may also be asked whether non-directive counselling even is possible. It is based on the division between knowledge and values, but the division is more problematic; experts choose the appropriate knowledge and the ways to express it. The values of the genetic counsellor may be present despite their effort to remain neutral, and the mere institutional context of providing the tests may imply the preferable course of action (Clarke 1997, Rantanen et al. 2008a).

Due to the central role of non-directiveness in genetic counselling, considerable debate has centred around it. There have been demands that the role of non-directiveness be clarified. Biesecker (2000, 2001) and Kessler (1997) believe that it could serve as a component of the ethical basis for clinical practice and as a way of seeing the relationship between the counsellee and the counsellor, but regarding it as a goal of genetic counselling or as a

theory of clinical practice does not provide the best possible genetic counselling. While the traditional definition of non-directiveness has been seen as limiting, the concept has evolved: it has been defined as a strategy directing counsellees towards their own decisions, which is seen to require interactive, skill-based counselling (Kessler 1997, Weil et al. 2006, Rantanen et al. 2009). Despite the acknowledged difficulties in fulfilling non-directiveness, genetic counselling is still constituted as providing objective information (Latimer 2007).

The dominant models of genetic counselling have changed over time. Walker (2010) explains that after the eugenic model of genetic counselling, there was a preventive model period, when information about the risks was in the central role, although not many tests were yet available. In Walker's timeline, the decision-making model started to appear in the 1960s when, alongside technological possibilities, new options and help in deciding about them were attached to genetic counselling. During the past decades, the psychotherapeutic model, where ever more psychological and emotional elements are involved, has been dominant (Walker 2010). In Biesecker's (2001) analysis on the different goals of genetic counselling, this similar change in counselling models is also present, although she notes that different goals coexisted, too. In the 1970s, the first school of thought emphasised the preventive goals of genetic counselling and later, the second school the psychological well-being of the client (Biesecker 2001).

Thus, psychological elements, emotional support and shared decision-making have weighed heavily in the realm of genetic counselling since the 1970s (Harper 2008). Weil (2003) argues that the central ethos of genetic counselling should be to bring the psychosocial component into every aspect of the work instead of the emphasis on non-directiveness. In his opinion, the fundamental role of genetic counselling is to help individuals to use the information of medical genetics to meet difficult situations (Rantanen et al. 2008a). Decruyenaere et al. (2000) offer a combination of information-oriented and psychological counselling. Elwyn et al. (2000) believe that the shared decision-making model provides a useful framework for the complex interactions that occur in genetic consultations. In the shared decision-making model, the counsellor and the counsellee share information, on the grounds of which the decision is to be made. In all these models, the genetic counsellor is expected to respect the counsellee's views, to make professional opinion available and to empower the counsellee in the decision-making (Wüstner 2003, Gwyn and Elwyn 1999, Rantanen et al. 2009). Charles et al. (1997) see that the benefit of integrating the patient's emotions and personal values with the facts is that it offers a potential middle choice between the two polar extremes of paternalistic medicine and non-directive counselling where the counsellee is left alone to make the decisions.

In order to define the current practice of genetic counselling, MacCarty Veach et al. (2007) published a reciprocal-engagement model of genetic counselling practice. It was defined in 2004 by 23 North American genetic counselling program leaders who described the genetic counselling practice that is taught today. The reciprocal-engagement model consists of three components: educating genetic information, which is unique in genetic counselling, individuating counselling, which refers to the relationship between the counsellor and the counsellee, and the outcomes of genetic counselling, which means that the counsellee understands the information and applies it autonomously. Wang et al. (2004), too, emphasise the categorisation of the areas of counselling to identify the goals and to examine the process. Their categories resemble those of the reciprocal-engagement model: in their analysis the goals of genetic counselling are educating and informing counsellees about the genetic condition, providing support and helping them to cope, and facilitating informed decision-making (Wang et al. 2004).

Different models of genetic counselling have been developed and studied widely alongside the development of genetic testing and changes in counselling practice. Both the process variables of genetic counselling, such as counsellor competencies, communication and support, and the outcomes of genetic counselling, such as risk comprehension, satisfaction, distress and decision-making have been stressed (e.g. Biesecker and Peters 2001, Pilnick and Dingwall 2001, Biesecker 2001, Bernhardt et al. 2000, Soldan et al. 2000, Wang et al. 2004). Over the decades, many professional shifts and changes in the roles of counsellees have occurred that affect the models and the ideals of counselling. Since the definition of genetic counselling, published in 1974 (see introduction), the realm of counselling has expanded from referring mainly to reproductive counselling to counselling in the contexts of more complex and diverse situations and diseases (Walker 2010). A stronger emphasis on genetic predisposition to common complex diseases has been described as an important professional shift (Pagon 2002). Another important shift is expected in the "genomic era" along with genome-wide testing, personalised medicine and the increase in direct-to-consumer genetic testing (Harris et al. 2013, Walker 2010, Guttmacher and Collins 2003). These changes have led to the need for a new definition of genetic counselling (Resta et al. 2006, Walker 2010). Greater flexibility in genetic counselling protocols has also been demanded in parallel with the ever more diverse counselling situations (Guimarães et al. 2013).

2.2 Genetic test types and genetic exceptionalism

The reasons why people receive genetic counselling are diverse; thus, their expectations and experiences may vary considerably depending on the condition in question, test

type and their personal life situation (Pilnick 2002). Although all tests that provide information about the health, future health or the health of the offspring through the DNA or the chromosomes are called genetic tests, and the counselling provided in the context called genetic counselling, they cannot be bundled. Situations encountered at the counselling clinics are varied.

The situations of genetic counselling differ specifically due to the type of genetic test in question. In the European project EuroGentest – Harmonising genetic testing across Europe (www.eurogentest.org), recommendations for genetic counselling related to genetic testing were created. In the recommendations, different genetic test types were defined as follows:

> **Diagnostic testing** means a genetic test performed in a symptomatic individual to confirm or exclude a genetic condition. This is usually not very different from other medical tests performed in order to achieve a diagnosis, except for the possible involvement of relatives or implications concerning them. **Predictive testing** refers to genetic testing in a healthy high-risk relative for a specific later-onset monogenic disorder. The mutation in the family leads to the disease or a considerably high risk for the disease. **Susceptibility testing** (sometimes referred to as risk profiling) means a genetic test of a marker or simultaneous testing of several genetic markers with the aim to detect an increased or decreased risk for a multifactorial condition in a healthy individual. **Pharmacogenetic testing** means testing for a genetic susceptibility for adverse drug reactions or for the efficacy of a drug treatment in an individual with a given genotype. **Carrier testing** means a genetic test that detects a gene mutation that will generally have limited or no consequence to the health of that individual. However, it may confer a high risk of disease in the offspring, if inherited, from one parent (in case of X-linked inheritance, autosomal dominant premutation or chromosomal translocation) or in combination with the same or another mutation in the same gene from the other parent (in case of autosomal recessive inheritance). **Prenatal testing** refers to a genetic test (either to detect a mutation, linked haplotype or chromosomal change) performed during a pregnancy, where there is increased risk for a certain condition in the fetus. **Preimplantation genetic** diagnosis (PGD) means testing the presence of a mutation, linked haplotype or chromosomal change in one or two cells of an embryo in a family with a previously known risk for a Mendelian or chromosomal disorder, in order to select the unaffected embryos to be implanted. **Genetic screening** means testing where the target is not high risk individuals or families, but where the test is systematically offered to the general population or a part of it (e.g. newborns, young adults, an ethnic group, etc.). (Kääriäinen et al. 2009.)

As the term 'genetic test' encompasses a wide range of possible situations in terms of the type of test, identifying a common goal for all genetic counselling may not even be possible. It has been argued that the practices of genetic counselling, evolved in the

reproductive context, should not be transferred directly to other testing situations as this would treat genetic information differently from other health information (Chadwick 1999). The emphasis on non-directiveness in genetic counselling has derived from the desire to avoid eugenics in the reproductive decision-making situations, but it may have failed to recognise the differences between genetic testing situations. While the goal of reproductive counselling could be to promote the counsellee's self-determination in exercising choices, the goal of genetic counselling for common diseases resembles that of other health education (Biesecker 2001).

It can also be asked whether diagnostic genetic tests should be separated from other medical tests at all. They confirm or exclude a condition in a symptomatic individual like any other tests. They are part of ordinary medical practice in that a symptomatic patient is diagnosed and possibly treated, whereas in predictive, carrier and prenatal testing the subjects concerned are usually not patients at all, but healthy people, tested for future health or for the health of their offspring (Harper 1997). While diagnostic genetic tests can be compared to any other medical tests, predictive genetic tests have been regarded as more "exceptional" (Green and Botkin 2003). Genetic exceptionalism refers to the use of genetic information differently from other health information (Rothstein 2005). It has been suggested that genetic information is special, because it is immutable through lifecourse, predicts future illness, has implications for family members and may lead to social discrimination and psychological anxiety (Human Genetics Commission 2002, Launis 2003, Armstrong et al. 1998). Genetic testing has also been considered exceptional because many hopes and fears have been directed at genetic information. The gene has been described as having the status of a cultural icon (Nelkin and Lindee 1995). Shiloh (1996) writes that even though the decisions made in different testing situations may vary greatly, all decisions regarding genetic information have something in common: their seriousness, the relevance for the person in question, for family members and even future-generations, the irreversibility, and the significance of the basic values of life. It has also been suggested that patients feel greater responsibility for their condition when it has a genetic basis (Hollands et al. 2012, Hallowell 1999, Novas and Rose 2000).

Rothstein (2005) considers that genetic exceptionalism is a legislative question: because more far-reaching laws are often politically infeasible, genetic-specific laws are regarded as better than nothing. There have been different views on whether genetic testing should be regulated separately from other medical practice. One perspective is based on the view that it does not differ from other medical testing and therefore does not need separate regulation, whereas the opposite view is that it should be seen as completely removed from medical regulation. The middle way between these is that genetic testing should be

partly involved in general medical regulation and partly have its own regulation (Meincke 2001).

Evans and Burke (2008) state that the history of genetic information, where it has mainly been used to diagnose conditions for which treatment has been limited, has justified genetic exceptionalism, but the future looks different now the treatments are developing, genetic information in drug treatment increasing, and classification of diseases changing. It has also been argued that genetic exceptionalism has served to maintain a privileged position for genetic information in the clinic, requiring safeguards in its communication to patients (Ross 2001, Will et al. 2010). Recently, the discussion has turned increasingly towards the non-exceptional trend in genetics. It has been argued that other than genetic tests may as well predict future illness, affect family members and increase psychological anxiety (Green and Botkin 2003, Launis 2003). Genetic exceptionalism has also been opposed as potentially leading to genetic determinism (Murray 1997). As genetic testing is increasing, specifically in diagnostic settings and in the risk assessments of individual treatments for common diseases, the ideal genetic counselling seems to be slightly changing (NSGC 2010, Saukko et al. 2006). Instead of genes, the implications of the test and the individual situation in which the counsellee is are emphasised as the grounds for counselling ideals (Saukko et al. 2006, Guimarães et al. 2013, Wang et al. 2004, Will et al., 2010).

Although uniform ideals for all genetic counselling may not be realistic or reasonable given the different situations in which people seek services, Biesecker (2001) considers that there should be a consensus among providers, at least within the same country, in order to standardise the practice, deliver a clear message to counsellees and to serve as guides in the training of genetic counsellors. The requirements of standardised practices and, on the other hand, considering the variation of counselling situations, suggest that the practices of genetic counselling need common discussion and evaluation, especially considering the possibilities of genome-wide testing and the increasing interest of people in genetics and its implications for health.

2.3 Planning life on the basis of genetic information

2.3.1 Understanding risks, making choices

An element of risk essentially belongs to genetic counselling. Genetic counselling does not usually provide a certain answer, yes or no, but a probability figure of having the mutation, becoming ill or passing the mutation to one's offspring. This needs

to be translated into a decision in genetic counselling. Even if a genetic mutation is found, it necessarily does not lead to the disease and if it did, there is no certainty when and how. The genetic map has been thus described as a gamble – it does not predict certainties but risks (Davison 1996). This risk information can be used to make choices and plan life, and the growing number of predictive tests will make this possible for more and more people. It has been argued that those who know their genetic risks can prepare themselves and make their lives longer and better (Beck-Gernsheim 1996, Rose 2001). However, knowing genetic risks may also increase anxiety and decrease the quality of life (Helén 2004). Hence, genetic information cannot be regarded as simply good or bad, but needs to be considered from different perspectives in genetic counselling.

It has been argued that a new model of medicine, based on the surveillance of the healthy population, not only sick patients, has developed (Ettorre 2002). Predictive information and risks are named as the key elements in this new medicine (Clarke et al. 2003, Scott et al. 2005, Skolbekken 1995), and providing personalised risk information about one's health, genetic testing plays an important role. Helén (2004) describes medical genetics as a display window of the new medicine since it requires sophisticated technology, makes individuals aware of their future diseases and focuses strongly on individuals.

In Ulrich Beck's theory of reflexive modernisation, the practice of risk management is an essential feature of the new society (Rantanen et al. 2009). He refers to the systematic way of dealing with hazards and insecurities that are introduced by modernisation (Beck 1992). Individualisation is a key process in the risk society: individual choices are made obligatory and dealing with risks an essential cultural skill (Beck 1992). Hence, genetics has been described as a science of reflexive modernity (Kerr and Cunningham-Burley 2000, Rantanen et al. 2009). Beck states that the focus on risks comes together with the rise of technologies. Therefore medicine – and especially genetics – has a strong role in the risk society. Health risks are to be managed through precisely these technologies. Genetic counselling is an immediate situation in which risk estimations are made and possible ways to manage the risks are dealt with. It can be thus regarded as a central stage of the risk society.

Understanding genetic risk is not always easy. The figures of risks and probabilities need to be transformed into an answer (Kerr and Shakespeare 2002). It is known that the way in which the risk is presented in genetic counselling affects the decision-making (Michie and Marteau 1996, Alaszewski and Horlick-Jones 2003, Edwards et al. 2002). Therefore, presenting risk information and turning it into social implications may direct decision-making despite all the efforts of the counsellor to remain neutral.

Decision-making is another central component of genetic testing and counselling: on the basis of the risk, a decision needs to be made whether to have the gene test or not, and how to use the genetic information received via the test. These decisions are made either before genetic counselling or during the counselling process, and they are always expressed and discussed in the counselling session. In the case of diagnostic genetic tests, the decisions are usually rather simple: the symptoms already exist, so the gene test is usually performed just to verify or exclude a certain condition. However, these tests may also affect future plans: whether to have children who have a risk to have the same condition as the parent, or in the case of a diagnostic test performed on a child, whether to have siblings who have a risk of having the same condition; carrier testing and prenatal testing directly affect reproductive decision-making. Carrier tests are performed in order to discover if there is a risk of passing the disease in the family to future offspring. Prenatal tests are performed if there is a recognised risk of this, whether found in prenatal screening or on the basis of the family history. In these cases, especially in prenatal testing when the baby already is expected, big social and ethical questions are dealt with and the decisions made may have a fundamental effect on the life of the person or the family.

Predictive genetic testing involves a set of different questions. The result of a predictive gene test, too, may affect the decision on whether or not to have offspring, but the main purpose of such a test is to discover whether there is a risk that the person tested will have a genetic condition in future. Knowledge gained via a gene test about a disease that will or may appear later may dominate people's lives and affect future plans – sometimes more than it needed to (Kerr and Shakespeare 2002). Therefore, predictive genetic testing is usually considered very carefully before the decision on the test. The individual's right to know, but also the right not to know, about the genetic risk is a basic principle of genetic testing (e.g. Council of Europe 1997, 2009).

The individualisation that Beck (1992) described as an essential process of the risk society has been seen to characterise the practices of modern medicine. Everyone is asked to advance their health and establish control over one's life through lifestyle and preventive measures (Petersen et al. 2010). Genetics represents well the individual-focused healthcare in which citizens make rational decisions on the basis of full information about their risks. Rose (2001) states that, unlike other health practices, in the context of genetic testing people can reinterpret genetic information rather than just follow doctor's orders and make choices about their own genetic futures. The rhetoric of the individual's right to know and informed choice has been argued to reflect the expectations that individuals govern themselves and make rational decisions based on the available information (Petersen 1999, 1998). Jallinoja (2002) points out, however, that even after receiving all

the information, the individual has a set of open questions about life that need to be thought through in the light of the information but also in the light of expectations, hopes and interpretations. The genetic counsellor's role is to empower the consumer in free but informed decision-making (Jallinoja 2002).

An individual making informed and free decisions is at the centre of the neo-liberal consumer society (Kerr 2004). It is being argued that the neo-liberal practices of governance utilise the active citizenship as a strategy to accomplish regulated autonomy: the rational decision-maker fulfils the responsibilities of the citizenship by undergoing genetic tests and counselling (Petersen 1999). In addition to the option of choosing the test, or the procedures after the test, individuals are obliged to do so (Rose 2001, Helén 2001, Petersen 1999). In the light of neo-liberal practice, every citizen's duty is to acquire information about genetic risk, to take an active role in health, and to independently inform family members (Polzer et al. 2002, Van Hoyweghen et al. 2006).

The individualised nature of modern medicine, including confidential decision-making, makes the involvement of the family in genetic testing difficult to handle. The concept of the family as a patient does not fit into individual-focused healthcare. Genetic testing, however, always includes the element of kinship: every result received from a gene test makes other family members aware of their risks (Sarangi et al. 2003). In genetic counselling, familial emotions and possible conflicts need to be considered: hearing the results of a gene test may awaken feelings of guilt for not having the disease, strong defences or an urge to blame the spouse whose family carries the gene (Richards 1996). The contradiction between confidential, individual genetic information and the responsibility of the individual to deliver genetic risk information to family members is widely discussed (Aktan-Collan et al. 2011, Forrest et al. 2007, Gaff et al. 2007, Koch and Nordahl Svendsen 2005, de Wert 2005, Claes et al. 2003).

There are already thousands of families who receive genetic information via genetic tests and are both capable of and forced to make decisions with this information – sometimes big decisions such as starting a family or choosing a profession. For most people, genetic information still does not play a role in life, but the group of people who are aware of their genetic risks or the genetic basis of their disease is growing in proportion to the genetic research into common complex diseases and as tests become cheaper with new technologies. Commercial companies that offer direct-to-consumer genetic tests also increase the proportion of the population aware of and interested in the genetic basis of their health (Harris et al. 2013). Decisions based on genetic information can therefore be clearly seen to be on the increase.

2.3.2 Exercising life politics within consumerist health services

Since genetic information is information about future risks and chances, it is a basis for planning life (Rantanen et al. 2009). Anthony Giddens (1991) has proposed that modern life includes an increasing amount of chances and individual choices to be made, which leads to the exercise of life politics. By life politics, Giddens means politics dealing with identity, lifestyle and self-reflection: it is politics about decisions concerning one's life, through which a unique personality superseding the social basis for identification may be built (Giddens 1991). Individualised lifestyles and individual knowledge about risks are cornerstones of life politics.

The shift of power towards the individual has also been considered in the theories of bio-politics. Michel Foucault, whose ideas on biopower and biopolitics have been productively used by social and political scientists (e.g. Rose 2001, Rose and Novas 2004, Rabinow and Rose 2006, Petersen and Bunton 2002, Petersen and Lupton 1996, Polzer et al. 2002, Lemke 2004, Kaufman and Morgan 2005), argued that a new form of power emerged along with modernity: biopower that operates through specialised knowledge and self-governance replaced sovereign power (Foucault 1978). Bio-politics is the politics of population operating through its self-governing individual members (Foucault 2003, Dean 1999).

Influenced by the ideas of Foucault, Rose (2001) suggested that the molecule has come to replace the population in later biopower. In his argument, we are living in an era where life, the life sciences, their institutions and practices as well as politics are becoming increasingly molecularised (Raman and Tutton 2010). Now, the individual members of the population are both capable of taking and encouraged to take responsibility for managing their own lifestyles and improving their lives (Rose 2001). They are also stated as being enabled to take greater responsibility for their own vitality (Rabinow and Rose 2006). In this new form of bio-politics, biology is to be worked on and to be changed into part of the "economy of hope" (Rose and Novas 2004, Raman and Tutton 2010). This refers to the use of biomedicine in improving vitality and life. Helén (2004) conceives of today's business of advanced medical technology as a life enhancement economy. Biomedicine has changed the market of possibilities and increased the available options.

Alongside the technological development, people certainly now have many more opportunities to exercise life politics and make individual choices than they used to. Hence, it has been argued that we are living within consumerist, neo-liberal health services (Petersen et al. 2010, Petersen 1999, 1998, Tupasela 2010, Rose 1999, Bradby 2009, Gabe et al. 2004). Petersen et al. (2010) argue that consumerism is a manifestation

of the market fundamentalism that characterises neo-liberal rule. The neo-liberal rule requires people to take responsibility for their health which is seen as leading to freedom to pursue their goals (Rose 1999). Individual citizens are assumed to act responsibly, take appropriate preventive actions and exercise informed choice in the biomedical marketplace (Petersen et al. 2010).

In the healthcare characterised by consumerism, patients are regarded more often as clients who want urgent care, tailored to their personal needs. Awareness and self-care have grown, while the sources of medical information have increased, and patients have, in many cases, become experts in their own diseases (Bradby 2009, Gabe et al. 2004). New technologies have created new arenas to disseminate products and services and increased the development of the expert patient (Tupasela 2010, Brown and Zavestoski 2004, Fox et al. 2006). Bradby (2009) states that people's expectations of quality of life and health services have increased as well as an increase in life expectancy, which leads to increased consumerism. Medical information has become more popular, widely accessible and provided not only at the onset of the illness, but also as entertainment. This change has also made patients more active in the use of health information and services, and they often offer their own diagnoses on the basis of information collected from the Internet (Bradby 2009).

In the new medicine, where risks and predictive information are emphasised as tools to make choices, act responsibly, govern oneself and exercise life politics, new identities may also be created. The category of being-at-risk has been said to constitute a new source of social identity (Novas and Rose 2000, Scott et al. 2005). There is a class of healthy ill whose life choices may be based on risk information and future orientation. Nordahl Svendsen (2006) argues that by talking about what happened in the past and what might happen in the future, genetic counsellees explore social identities and generate perspective on one's life. They connect genetic risk profiles to other frameworks making genetic information meaningful in terms of their own lives (Nordahl Svendsen 2006). These social identities also serve as bases for new social groups. Patient groups are playing an increasingly important role as social groups and also as formulators of health policies (Novas 2007, Tupasela 2010, Petersen et al. 2010). Rabinow (1992) calls such grouping a form of new "biosociality".

The increased use of predictive genetic tests provides people increasingly more information about their future health. This information may be used to plan a lifestyle to avoid becoming ill – such life politics may relate to everyday habits such as eating and smoking, but also to big and unique decisions such as having children or selecting

their profession. Genetic information concerning one's future may give people tools to handle their lives, increase their anxiety, leave a set of open questions in front of them, or equally, be totally irrelevant for them. All these approaches need to be considered in genetic counselling where this information is delivered. As Rose (2001) has pointed out, the thought about the gene becomes more complex and fragmented as we move into the post-genomic era. The contexts of genetic information may become more ambiguous and the meanings of this information more varied among the counsellees.

2.4 Criticism towards geneticisation and new eugenics

The increasing role of genetics in medicine has inspired social scientists, many of whom have warned of the dangers that the increasing quantity of genetic information may bring (e.g. Lippman 1991, Nelkin and Lindee 1995, Fukuyama 2002, Ettorre 2002, Kerr and Shakespeare 2002, Rothman 2001). In the social sciences, the institutions and practices of medicine have often been analysed critically. In their famous book about medicalisation, Conrad and Schneider (1980) wrote about the growing role of medicine as the institution that maintains the social control of defining the deviance and normality. In the strictest views of medicalisation, the increased medical power has been seen to cause abuse of power in the name of healthcare (Illich 1995, Árnason and Hjórleifsson 2007). According to the medicalisation theorists, the medical sector has become more dominating, and ever more phenomena have been conceptualised through medicine, which affects the behaviour and attitudes of people. This view has also been criticised. The medicalisation critique has been based on the Foucauldian interpretation in which medicalisation does not need to be seen as repressive and be opposed. In this perspective, we nonetheless live in a medicalised society that serves to keep social order and producing health (Lupton 1998). Modern medicine can be seen as striving to realise our wishes to achieve health and live longer and better lives (Helén 2002, Foucault 1978).

As genetic technologies and knowledge have increased, fear of the power of medicine gained a new form in the fear of the power of genes. Abby Lippman launched the concept *geneticisation* in 1991. She referred to "genetic colonisation of the body" by genetic mapping projects, the benefits of which she considered doubtful since in the case of many common disorders knowing the genes does not provide new ways to promote health (Lippman 1991, 1992, Rantanen et al. 2009). Geneticisation refers to the trend of explaining more and more diseases and characteristics through genetics and can, thus, be also referred to as genetic determinism or genetic essentialism (Árnason and Hjórleifsson 2007).

Genetics has created great expectations. It has been expected to change the conceptions of health and illness, allowing identification of those at risk and the use of drugs tailored to the individual (Petersen 2006). Although Petersen (2006) considers it doubtful that many of the promised benefits of genetic research will be delivered, he argues that an increasingly pervasive genetic worldview nonetheless profoundly shapes conceptions of health and illness and priorities in healthcare. Several authors have, especially at the turn of the millennium, argued that the gene has become a cultural icon that has a remarkable role in present thinking, and which becomes visible in the ways that the genome and the DNA are discussed (Nelkin and Lindee 1995, Rothman 2001, Ettorre 2002, Kerr and Shakespeare 2002). Geneticisation has thereby been seen to take place both in medicine and in the society and culture.

There has been concern that genetic determinism may lead to discrimination on the basis of genes (Morgan 1996). The fear of eugenics remains. While the old eugenics focused on populations and coercive control, the new eugenics is considered to be found in the focus on counselling of individuals (Petersen 1999, Raz 2009, Raman and Tutton 2010). New eugenics has been called "backdoor eugenics", as if sneaking in through backdoor (Duster 2003). This, in addition to the concept of "consumer eugenics", refers to the individual choice in the reproductive genetic counselling situations that are not purposely eugenic, but may lead to change in the composition of population, and, despite the decision, always have a eugenic quality (Kerr and Shakespeare 2002, Lemke 2002). Rothman (2001) calls this 'micro eugenics': "good" and "bad" genes are selected and named socially and politically, which reflects values concerning diseases, disorders and deviances. Raman and Tutton (2010) ask that if individuals are now obliged to be responsible, as argued, does this not suggest that disciplinary power remains: although coercive eugenics is no longer a part of state intervention, there are still normalised population-based interventions coordinated by the biopolitics of the state.

In clinical genetic counselling settings, questions of power and choice are always immanent. It has been stated that genetic counselling can never be objective: the way that the alternatives are presented is said to encourage people to behave in a certain manner, and, thus, the eugenic practices in the context of prenatal genetic testing cannot be seen to have completely disappeared (Kerr and Shakespeare 2002). Clarke (1991) sees that the attempt to measure the results of genetic counselling through outcome audit and other means of evaluation represents a neo-eugenic policy of the state in the search for improved public health at the cost of the wishes of individual patients. He sees non-directiveness and individual choice as safeguards for this (Koch and Nordahl Svendsen 2005).

The concept of medicalisation was created to criticise the optimism related to the increasing power of medicine. It was followed by the concept of geneticisation, criticising the power of genes. In the same way that medicalisation was opposed as not recognising the enabling role of medicine and as being misused as a polemical device rather than a scientific construct, so too has the concept of geneticisation been criticised (Árnason and Hjórleifsson 2007). Ettorre et al. (2006) consider it interesting that a rearticulation of life through geneticisation has occurred, though the science of genetics has not reached the expectations attributed to it. It has also been stated that the use of the concept of geneticisation is unqualified and ambiguous, and that it emphasises too strongly the role of heredity as a source of self-identity, or, on the other hand, completely fails to recognise the role of heredity in the constitution of all living things (Cox and Starzomski 2004, Condit and Williams 1997, Novas and Rose 2000, Rantanen et al. 2009). Zwart (2007) demands that the debate on genetic information be moved into the 21st century, instead of addressing challenges genetic determinism that were popular in the 1990s but are now outdated (Rantanen et al. 2009). Kerr (2004) does this by declaring that the death of genetic determinism is widely accepted, and that genes are now regarded as complex, interactive and contested entities.

Raz (2009) describes a similar change in the debate concerning eugenics, moving from authoritarian to liberal. In the recent literature, the question has been raised of how we can employ genetics while avoiding the evils historically associated with it, emphasising a liberal eugenics that focuses on the individual's own values and conceptions of life (Raz 2009, Buchanan et al. 2000, Agar 2004, Gavaghan 2007). Green (2007) suggests that the fears of the new eugenics movement are overblown and that genetic interventions could improve parents' ability to enhance children's lives and promote social justice (Raz 2009). It has also been argued that the opposition between directiveness and individual choice does not fit into the practice of genetic counselling: directive information is considered necessary to create autonomous individuals (Koch and Nordahl Svendsen 2005). Jallinoja (2002) writes that the ethics needs to be negotiated situationally, since no universal rules can be applied to all genetic counselling situations, and since strict rules could produce a fear of eugenics if someone else made the decisions for us.

The social and ethical discussion concerning genetic testing and counselling has been on-going ever since the old eugenic practices of genetics. The discussion was very vivid especially in the 1990s when the Human Genome Project to sequence the entire human genome began, and then again at the beginning of 2000s as the project reached its end (National Human Genome Research Institute 2012, The International Human Genome Mapping Consortium 2001, Collins et al. 2003). Threats associated with the

increased knowledge of our genetic selves have been analysed widely, and the concepts of geneticisation, genetic determinism and new eugenics have been employed. Recently, the discussion has focused more on the possibilities that the increasing genetic information may bring, the "economy of hope" and "life enhancement technologies" (Rose and Novas 2004, Helén 2004). Increasing genetic information has both dystopian and utopian sides (Raz 2009). Genetic information is now more complex and fragmented, and between the threats and the possibilities there is an individual seeking genetic information via a gene test who may experience them both or ignore both the dystopias and the utopias and regard the information as ordinary health information.

2.5 Studying genetic counselling from different perspectives

Genetic counselling's different aspects have been studied widely, and professionals of genetic counselling have defined the best and the dominant practices and models of genetic counselling (e.g. Weil 2003, Biesecker 2000, 2001, Kessler 1997, Walker 2010, Elwyn et al. 2000, Wüstner 2003, Gwyn and Elwyn 1999, MacCarty Veach et al. 2007, Wang et al. 2004, Resta et al. 2006). The professionals have also been asked about specific issues related to genetic testing and counselling, such as the genetic testing of children (Brierley et al. 2014, Mackoff et al. 2010), non-directiveness, confidentiality or privacy of genetic information (Wertz and Flethcer 1988, 2004) or direct-to-consumer genetic testing (Howard and Borry 2013).

The perspective of the counsellees has also been studied widely. The focus has usually been on a specific situation, condition or a group of people. A big portion of studies on counsellees' expectations, knowledge and experiences relates to counselling in the context of genetic testing of cancer (Kosonen et al. 2008). In the studies examining counsellees' satisfaction, some elements of information have been emphasized as the most important aspect of genetic counselling (Salemink et al. 2013, Collins et al. 2000, Pieterse et al. 2005, Wertz and Gregg 2000). Counsellees have also appreciated sensitive communication as well as psychological support and the engagement of the counselor (Salemink et al. 2013, Evans et al. 2004, Skirton 2001, Pieterse et al. 2007, Bjorvatn et al. 2007). Counsellees have often not expected genetic counselling, but having received it, have appreciated the time spent on it (Bernhardt et al. 2000, Barr and Millar 2003, Metcalfe et al. 2007). The studies show that counsellees are usually very satisfied with the genetic counselling received, even if they lack understanding of the genetic information provided (Metcalfe et al. 2000, Liede et al. 2000, Davey et al. 2005, Holloway et al. 2005, Josephson et al. 2000, DeMarco et al. 2004, Hopwood et al. 2004, Elliott et al. 2014).

Since the research performed has focused more on the specific issues related to genetic testing and the associated counselling, the purpose of this study was to bring together different perspectives on genetic counselling and to examine them through the same research frame: what genetic counselling is expected to be, how it is discussed and how well the expectations correspond to the current practices. Guidelines covering genetic counselling were considered essential material, because they define what genetic counselling should be like. Since there is no research examining the guidelines concerning genetic testing from the perspective of counselling, this approach was also seen to fill in the gap in research and to serve as a tool for further development of guidelines concerning genetic counselling. Analyses of the guidelines concerning genetic testing have been made, but they have studied specific topics, e.g. testing of minors (Borry et al. 2006) or listed the legislation and the guidelines of testing and counselling in different countries (Matthiessen-Guyader 2005, Borry et al. 2007, Soini 2006).

Genetic counselling professionals have been asked about their views on the practices of genetic counselling, but a conclusive perspective was missing, and, therefore, it was seen as useful to assess the practices on the national level by the representatives of the national genetic associations. This perspective has not been studied before, although national surveys have been made to examine the legislation and recommendations on genetic testing (Matthiessen-Guyader 2005). It was considered necessary to involve counsellees' views, too, to answer the same research questions. Since counsellees' opinions have already been gathered in many studies, a background review of the existing studies was performed to gain a broad overview of the experiences. These were further discussed in interviews with counsellees from the perspective of this research frame.

3. The aims of the study

The objective of this research was to review the kinds of expectations that are concentrated on genetic counselling, and the different conceptions of genetic information that lie behind those expectations. Another purpose was to study how the expectations correspond to the current practices of genetic counselling in Europe. The research aimed at examining the perspectives of the different parties involved in the field of genetic counselling: bodies that have created written guidelines for genetic counselling, the National Societies of Human Genetics in European countries and people attending genetic counselling were defined as such parties.

The research questions were:

1) **What is expected of genetic counselling**
   a) in the written guidelines produced by different parties?
   b) from the perspective of people attending genetic counselling?

2) **How is genetic information and genetic counselling framed**
   a) in the written guidelines produced by different parties?
   b) by people attending genetic counselling?

3) **What are the practices of genetic counselling like, and how do they meet the expectations**
   a) from the perspective of the National Societies of Human Genetics in European countries?
   b) in the experiences of the people attending genetic counselling?

4. Materials and methods

4.1 Perspective of the international guidelines

Genetic counselling practices are guided by different international guidelines and recommendations, produced mostly by international organisations such as the WHO and professional associations. These guidelines are naturally occurring data that interestingly reflect the ideals of genetic counselling. The guidelines were regarded as essential material in defining what genetic counselling is expected to be, how it is actually defined and what kinds of constructions of genetic information are produced. International guidelines concerning genetic counselling were thus decided as constituting the key material in the analysis of the expected genetic counselling.

4.1.1 Data collection

Available guidelines covering counselling related to genetic testing were sought on the Internet. In addition to official guidelines, guidelines included written statements, recommendations and reports addressing issues related to genetic counselling. Papers produced by the relevant global, European and American organisations, as well as by organisations from other continents were included in the search. National guidelines were not included due to their large number and the diversity of languages; the exceptions were guidelines produced by the Nuffield Council on Bioethics and the Genetic Interest Group (both from UK), because they were referenced on many occasions. Guidelines were sought by browsing the websites of different organisations, either previously known or found with the help of the systematically examined references listed in other studies and websites, and, in the end, using the keywords "guidelines", "recommendations" and "genetic counselling" in the Google search engine. These guidelines were collected during 2005. The data collection process of the international guidelines is also described in Articles I and II.

The 56 documents collected were produced by 29 different organisations: UNESCO, the WHO and the OECD, European political bodies, ethical committees, organisations of genetic professionals, organisations of other medical professionals and patient associations. The guidelines are listed in Appendix 1. Few of the 56 guidelines collected were specifically written to address genetic counselling, but the topic was covered in all of them. Their backgrounds differed as some were official "soft-law" statements, while others were bioethical declarations of principles, or still less official recommendations and reports.

4.1.2 Data analysis

The documents were coded for different topics and sub-topics with the assistance of the software package QSR NUD*IST (Qualitative Solutions and Research for Non-numerical Unstructured Data Indexing Searching and Theorising). It is designed to support the processes of coding data, searching for text and patterns of coding, and theorising about the data (Barata et al. 2006, Alexander 2006, Hueifang et al. 2006, Scott et al. 2005). With the assistance of the QSR NUD*IST, two different analyses were performed for the data. First, the expectations of genetic counselling were studied by searching for those topics most often mentioned in the guidelines. The most common topics related to genetic counselling were identified, their main contents reviewed, and the similarities and differences between the documents analysed. This analysis is also presented in Article I.

The second analysis performed for the data was frame analysis. It belongs to the tradition of social constructionism, which aims to analyse the processes through which the common conceptions of reality are established (Berger and Luckmann 1971). Frame analysis resembles discourse analytical studies (see Potter and Wetherell 1989): both the content of the constructions and the ways that have been used to produce them are examined. The focus was on the dynamics of framing the meanings of genetic information (Rantanen et al. 2009).

Originally, the concept of framing was used by Erving Goffman, who investigated it as a process through which societies reproduce meanings. Goffman (1974) argued that cultures generate "primary frameworks" that offer meanings through which information can be digested. These frameworks enable us to identify, locate, perceive, and label occurrences (Fisher 1997). There is no consensus on what frames actually are, due to the different methodological interests for which the concept has been employed, but frame analysis has been argued to supply a common reference language for analytical points (Jackson 2005). Fisher (1997) defines cultural frames as "loose, socially-generated structures in discourse which people use to organise information, and around which groups develop ideological and policy arguments". In this study, 'frame' was also defined as a cultural structure around which the different constructions of genetic information, detected in the documents analysed, were organised (Rantanen et al. 2009).

The frames, through which genetic information was discussed in the guidelines, were defined by identifying topics through which the constructions of genetic information could be analysed. These 14 topics were identified in the coding performed with the QSR NUD*IST: (1) training of professionals providing genetic counselling, (2) the role of the family, (3) testing of vulnerable people, (4) practical issues of counselling, (5) the type of

information that should be provided, (6) the counsellee's understanding, (7) psychological support, (8) counselling in the context of prenatal diagnosis, (9) counselling in the context of predictive testing, (10) the right to know and not to know genetic information, (11) the autonomy of the counsellee, (12) non-directiveness, (13) confidentiality, and (14) the ethical issues involved in genetic testing. Text units of each of these topics were combined and reduced to more general statements. Finally, all the statements on each of the topics were reviewed together. The different constructions of genetic information found through this coding were extracted and sorted as three frames. This analysis is also presented in Article II.

4.2 Perspective of the national experts

In order to analyse how the ideals of the guidelines correspond to the practices of genetic counselling, an overview of genetic counselling in European countries was considered useful. The National Societies of Human Genetics were assumed to have a general overview of the practices of genetic counselling in their country, and, therefore, were selected as respondents. As the interest was in the general situation of genetic counselling in European countries, no individual practitioners were surveyed, instead the societies were asked to describe the practices considering the whole country. The collection and the analysis of this data are also presented in Article III.

4.2.1 Data collection

In 2005 and 2006, an electronic survey was performed in all European countries, with the exception of Andorra, Liechtenstein, Monaco, San Marino and the Vatican, which were assumed to co-operate with their neighbouring countries in the field of genetic counselling. The questionnaire was sent to the president of the NSHG in the 29 European countries that had such a society. They were asked to fill in the questionnaire within a group that had experience in practical genetic counselling. All these societies provided answers, written either by the board or the president of the society, or a designated expert. An appropriate contact person was sought with the assistance of the EuroGentest network and the European Society of Human Genetics in the countries found not to have a NSHG. The contact persons in nine additional countries answered the questionnaire. Hence, answers representing 38 countries were received (the countries are listed in Table 1 in Article III). The survey was piloted in Finland and Belgium.

Genetic counselling was defined for the respondents in a covering letter according to the definition of the European Society of Human Genetics (Godard et al. 2003).

Questions about legislation, guidelines and generally applied practices of genetic counselling were formulated on the basis of the topics that emerged in international guidelines analysed. 'Legislation' referred to official laws passed by the parliament or government, or additional legally binding documents. 'Guidelines' meant professional or other best practice recommendation papers. 'Generally applied practices' referred to those practices that, though not necessarily written, are recognised as the common manners of practice.

Respondents were asked whether their country had legislation, guidelines or generally applied practices for counselling in different genetic testing situations. The testing situations that were asked about were counselling in the contexts of diagnostic, carrier, predictive, prenatal and preimplantation testing, and susceptibility testing for multifactorial diseases. The respondents were also asked whether their countries had legislation, guidelines or generally applied practices for different aspects of genetic counselling. These were: by whom counselling may be performed, asking for consent, confidentiality and the need to breach it, non-directiveness of counselling, offering psychological support to the counsellee, informing at-risk relatives, the duty to recontact the patient, counselling minors or those with diminished mental capacity, and counselling members of ethnic minorities. In addition, respondents were asked to describe how the existence or lack of legislation and guidelines affected the practical genetic counselling work, how well genetic counselling services were organised in their country, and how they predicted the situation would change in the coming years. These were open questions, but the answers could be classified into different categories. The questionnaire is in Appendix 2.

4.2.2 Data analysis

Answers from the NSHGs and the contact persons were analysed with the help of the SPSS (Statistical Package for Social Scientists) to gain an overall view of the regulations and practices of genetic counselling in Europe. This analysis, however, was not and did not aim to be a quantitative analysis of the European practices, but a description of the situation from the perspective of an expert from each country. Thus, no statistical data was produced, but with the help of the SPSS the answers from different countries were compared and an overall picture was gained of the situation in each country, as well as concerning each topic asked about. The answers for the open questions were classified while coding on the basis of the answers given. The results and the discussion of this analysis are presented in Article III.

4.3 Perspective of the people attending genetic counselling

The perspective of people attending genetic counselling was regarded as the third cornerstone in the definition of the expectations, frames and practices of genetic counselling. Since it was desired that the themes of genetic counselling be examined profoundly, it was decided that interviews would be conducted of people who had gone through a genetic testing process in a genetic counselling clinic. This also allowed a frame analysis that could be compared to the frame analysis of the guidelines. As one aim of the dissertation was to study international and European ideals and practices of genetic counselling, it was originally planned to study the perspective of people who had attended genetic counselling on the international level. However, an international survey or interviews were considered problematic for many practical reasons, such as the diversity of languages and healthcare systems to be considered, timetable and budget. On the basis of the recommendations from the international EuroGentest NoE expert group on genetic counselling, the international perspective was instead examined by searching through a large number of existing studies on patients' expectations and experiences of genetic counselling, performed in different countries. A review of these previous studies was then performed to serve as background information for the interviews. On the basis of the review, the main topics describing the ideals and practices of genetic counselling were discussed in the 10 interviews with counsellees who had visited a genetic clinic in Turku, Finland.

4.3.1 Data collection for the background review

The search for articles on genetic counselling from the counsellees' perspective was conducted in 2007 on the databases PubMed, Cinahl, and PsycINFO for literature from 1997 to June 2007, using the term *genetic counselling* in combination with the following terms: *counsellee, expectation(s), perception(s), needs, satisfaction* and *knowledge*. Papers were excluded from the review if the study was not related to genetic counselling situations where genetic testing was actually offered, if genetic counselling was conducted in the context of a genetic screening programme, or if they were reviews. Studies dealing with prenatal genetic counselling were excluded if the reason for referral for counselling was not a genetic disorder in the family or a positive/ambiguous prenatal diagnostic test result. (Kosonen et al. 2008.)

102 studies were identified that examined patients' expectations of counselling related to genetic testing, their experiences on genetic counselling, or their knowledge after genetic counselling. These studies are listed in Article IV. The studies were performed in several different countries, mostly the UK and USA. In 21 studies, the research method used

was an interview, in 55 a questionnaire and a combination of these two in the remaining cases. The number of research participants ranged from 6 to 833. In 30 studies the research participants were relatives at risk; in four studies, those affected; in four studies, parents; in one study, support persons; in one study, spouses; and in one study, pregnant women. The remaining studies included combinations of various types of participants. In 31 studies, the type of the genetic test concerned was predictive testing; in eight studies, diagnostic testing; in three studies, prenatal testing; and in two studies, carrier testing. In the remaining studies, genetic counselling concerned other various types of testing. A great majority of the studies (72) related to genetic counselling in the context of cancer. (Kosonen et al. 2008.)

4.3.2 Data analysis of the studies on counsellees' experiences and expectations

As the number of the studies included in the background review was large, the statistical software package SPSS was used as a tool to assist the analysis. The topics examined in at least five studies were coded into SPSS. In the studies, the topics were either only introduced, or it was mentioned that a certain percentage of the respondents were of this opinion, or in the case of a small number of topics, the results were expressed on a scale. The topics that were only introduced were coded as yes/no, and the topics that expressed the percentages were coded using 10% intervals. The topics that were expressed on a scale were converted into a shared comparable scale. After coding, the data was systematically reviewed in order to discover what the main expectations and experiences of the counsellees were. In addition, the expectations and experiences expressed in each study were further reviewed in order to collect results that possibly did not fit into the analysis performed with SPSS. The topics related to knowledge after genetic counselling were excluded at this stage of analysis because it was thought that measuring the knowledge was not directly linked to the experience on ideal genetic counselling. The background information of the review data was published on the EuroGentest website (Kosonen et al. 2008). The results of the background review have not been published, but they have been used to ground counsellees' perspectives in the interview study presented in Article IV.

4.3.3 Collection of the interview data

The collection of the interview data is described in Article IV. After reviewing previous studies on counsellees' experiences on genetic counselling and forming an idea of the expectations and experiences on genetic counselling from the counsellees' perspective, interviews of counsellees were performed in 2007 and 2008. A letter suggesting an interview was sent to 24 persons who had visited the genetic clinic of the University

Hospital of Turku in Finland (letter in Appendix 3). A letter was sent to eight persons who had visited the clinic because of a diagnostic genetic test of their child or themselves, to eight persons who had visited the clinic because of a carrier test, and to eight persons who had visited the clinic because of a predictive genetic test. The focus of the study was ideal genetic counselling. It was considered that the three different test types would bring enough of perspective on the topic, and, thus, it was decided that letters were not to be sent to people having visited the genetic clinic due to prenatal testing, because it was not wished to study the sensitive issues and instant decision-making related to prenatal testing. The letters were sent to persons who had visited the clinic at least three months ago and from this time back until all letters were sent. None of the persons had visited the clinic longer than a year ago. The persons had visited the same genetic clinic, but discussed with different clinical geneticists, who were not aware of the following interviews at the time of the counselling.

Ten counsellees agreed to be interviewed: two parents (from different families) who had visited the clinic because of their child's diagnostic genetic test (Connexin-26 related hearing loss and hereditary motor and sensory neuropathy), one who had had a diagnostic test for herself (haemochromatosis), four who had had a carrier test for serious childhood-onset autosomal recessive diseases on the basis of the disease in the family, and three who had considered a predictive test (two for Huntington's disease and one for Lynch syndrome), of whom two had had the test.

The interviews were performed at the University or the University Hospital of Turku, or in counsellee's home, however they wished. The interviews lasted from 45 to 90 minutes. The counsellees were asked about their expectations and experiences of genetic counselling. The topics of the previous studies guided the framework of the interviews. In addition, general questions about genetic testing, family history and the life and future of the counsellees were asked. The interview framework is in Appendix 4.

4.3.4 Analysis of the interview data

Frame analysis was performed for the interview data. First, the transcripts of the interviews were examined with the assistance of the software package QSR NUD*IST (Qualitative Solutions and Research for Non-numerical Unstructured Data Indexing Searching and Theorizing). The text was coded into different topics and sub-topics. The topics were based on the themes dealt with in the interviews and completed with the discoveries during the coding process. The topics were 1) accessing the genetic clinic, 2) expectations of genetic counselling, 3) practical issues, 4) evaluations of genetic counselling, 5) the

decision on testing, 6) receiving the test result, 7) evaluations of the counsellor, 8) genetic testing in general, 9) family, 10) the counsellee's situation and future, and 11) the disease.

Each topic was examined more closely and the key categories were determined within the topics: 1) history and family background, 2) expectations, 3) no expectations, 4) genetic counsellor, 5) positive experiences, 6) negative experiences, 7) most important factors in genetic counselling, 8) differences to other healthcare services, 9) psychosocial issues, 10) ethical issues, 11) everyday living, and 12) future planning. Each of these categories was examined separately and sorted into different perspectives. Finally, all the perspectives were compared and combined into main frames, through which the interviewees described their experiences on genetic counselling. Frame analysis as a method is described in more detail in Chapter 4.1.2. The frame analysis of the interviews is also presented in Article IV.

5. Results

There were three different objectives in this study: to define what is expected from genetic counselling from different perspectives, to analyse how genetic information and counselling are framed, and to study what the practices of genetic counselling are like in Europe and how they correspond to the expectations. In this chapter, answers are provided to each of these. First, the expectations attached to genetic counselling, defined in international guidelines and determined from the experiences of people who have attended genetic counselling, are presented. Second, the results are introduced of the analysis on how genetic information and its provision in genetic counselling are framed in international guidelines and in the interviews of the persons who have attended genetic counselling. The practices of genetic counselling in European countries, described by the national experts and assessed by the people who have attended genetic counselling, are presented in the third part.

5.1 Elements expected from genetic counselling

Listing the elements of expected genetic counselling reveals how genetic counselling is regarded as a part of the healthcare services, and how genetic information is regarded as a part of the health information that people receive. The ideal elements were sought by reviewing the international guidelines discussing genetic counselling and by examining the counsellees' perspectives in the interviews and in the background review.

5.1.1 Ideal genetic counselling in international guidelines

Nine topics appeared as the elements of ideal genetic counselling more often than the others in the guidelines analysed. All of these topics were mentioned in at least 30 of the 56 documents and are here regarded as the most salient aspects through which ideal genetic counselling is defined. The requirements most often mentioned belonging to genetic counselling in the guidelines were *the education and training of professionals*, *content of the information to be provided, the counsellee's understanding of genetic information, informed consent, autonomy of the counsellee, psychological support*, *the implications for the family, confidentiality* and *genetic discrimination*. These are presented in Article I.

Given the complexity of the genetic information to be delivered, the guidelines considered that only specialists in the field of genetics should perform genetic counselling. It was

considered inevitable that the number of non-geneticists requesting genetic tests and disclosing test results would increase as the use of genetic tests increases, and, therefore, training in medical genetics as an integral part of healthcare training was considered necessary. Good communication skills as well as understanding of the ethical complexities of disclosing genetic information were seen as important parts in the training of professionals in genetics.

The information that was expected to be covered in genetic counselling included information about the condition in question, treatment options, the risk of having the condition, the purpose, nature and consequences of the genetic testing in question, the risks involved in the procedure, the limitations of testing, the alternatives the counsellee should consider, practical information on what will happen next, the potential harm of testing, the risks to family members, and information on the support groups available. The information was expected to be objective, adequate, balanced, understandable and adapted to the counsellee's circumstances.

Counsellors were expected to aid the counsellees in making their own informed choices and to encourage the independence of their decision-making. Some guidelines specified that free choice means more than the absence of coercion: it was described as meaning the practical ability to act afterwards on the decision that has been made. Some guidelines stressed the importance of psychological support, stating that it is at least as important as the informational aspect, and that responding to counsellee's emotional reactions may even take priority.

Genetic information that is both personal and familial was seen as often raising questions about the moral obligations of the professional towards the relatives who are at risk of a genetic disease. Different documents emphasised disclosure of information differently. All agreed that the counsellee has a moral obligation to share the genetic information with the family members that are at risk, and that if this is relevant to other relatives it should be recommended that the patient disclose it – or, more strongly put, that the patient should be persuaded to disclose it. Some guidelines went further to suggest that the counsellor should be able to warn the at-risk relatives when the disorder is serious and there is prevention or treatment available. The guidelines advised that the social impact that genetic information has on the family should be considered in counselling as well. It was considered important that genetic information be shared with the partner, especially if it affects the children or decisions regarding family planning, but also when there are no such issues, because the result of a genetic test was seen in any case as having a considerable impact on family life.

The confidentiality of genetic information was introduced as a general principle that should be respected in genetic counselling. Two kinds of situations where confidentiality may be jeopardised were defined. First, insurance companies, employers and school admission boards were regarded as possibly interested in the results of a gene test. The guidelines stated that privacy of information must be assured. Second, the fact that genetic information concerns family members was seen as raising potential conflicts between confidentiality and the duty to warn the relatives. Therefore, confidentiality as an absolute principle was considered problematic in many guidelines. However, breaching confidentiality was seen as an exception which the professional should consider carefully.

Discrimination against a person on the grounds of his or her genetic heritage was regarded as the major ethical question related to genetic testing that must recognised in genetic counselling. The guidelines prohibited such discrimination and suggested that society should support genetic differences, which means that indirect action, such as limiting healthcare access for the patients with genetic diseases, should also not take place.

5.1.2 Counsellees' expectations of genetic counselling

Counsellees expectations of genetic counselling that were listed on the basis of the background review of the previous studies on counsellees' experiences are presented in Table 1. This was used to ground the interviews of the counsellees but has not been published. The first column shows the number of the studies in which the issue was either offered as an option or introduced by the respondents. The second column indicates in how many studies the issue was introduced in general but without measurement of how many respondents expected it, or how much it was expected. In the third column is listed the number of the studies in which a certain percentage of the respondents expressed the expectation. This can be compared to the other number in the column, indicating the number of studies in which the majority of the respondents felt that way. The last column shows the number of studies in which a scale was used to measure the expectations. The summary of the values in scale from 0 (lowest value) to 1 (highest value) is expressed after the number of the study.

Table 1. Expectations of genetic counselling.

Expectations of genetic counselling	Total number of studies in which the issue offered as an option or introduced by the respondent (N=102)	Number of studies where the issue was expected in general	Number of studies in which it was expected by a percentage of respondents / in how many of those expected by more than half of the respondents	Number of studies in which it was expressed on a scale / the summary of the average values in scale 0-1
Information about one's own risk	23	7	14/9	2 / both above 0.7
Information about the condition in question	12	4	7/5	1 / 0.5-0.6
Information about inheritance	12	2	7/5	3 / all above 0.5
Information about risk management	12	3	8/4	1 / 0.9-1
Information about treatment and the prevention	12	1	10/5	1 / 0.9-1
Reassurance	11	1	9/2	1 / 0.8-0.9
Information about the risk of other family members	10	2	8/2	0
Access to genetic testing	10	2	7/1	1 / 0.9-1
Surveillance	9	2	7/1	0
Information about the risk to children	8	2	5/2	1 / 0.8-0.9
General information	8	1	6/3	1 / 0.5-0.6
Information about the procedure	8	2	5/1	1 / 0.9-1
Aid in decision-making	7	2	3/0	2 / both above 0.7
Dealing with the emotional aspects	6	2	2/2	2 / both above 0.6
No expectations	6	4	2/1	0
Information about the family history	5	0	5/3	0

The expectations that most frequently appeared in the studies reviewed – either because of being asked of the respondents or introduced by them – were those concerning information about one's own risk, the condition in question, inheritance and risk management, prevention and treatment. The issues expected from genetic counselling by the majority of the respondents in most studies were information about inheritance, information about the condition in question, information about one's own risk, information about the family history, and dealing with emotional aspects. The issues not relating directly to information, such as reassurance, surveillance and help in decision-making were not expected by as

many counsellees in most studies. When asked about the expectations on a scale, all of the topics asked about were expected above the middle value.

Following the trend in the background review, information was the only element that was expected beforehand of genetic counselling by the counsellees interviewed. They expected the counsellor to answer their questions and to deliver information about the condition in question and about the ways to manage it. In addition, they wanted practical information about the testing process. Many interviewees also mentioned that they did not expect anything special from genetic counselling, but mainly the test result. Several interviewees expected the counselling session to be rather fast and practical. Some of them had not expected genetic counselling at all.

5.2 Framing genetic information and counselling

As the expectations of genetic counselling, sought in the guidelines and from the perspective of counsellees, are a rather superficial lists of elements that should be involved in the counselling session, the topic was approached also through frame analysis as this provides tools for deeper analysis of the conceptions of genetic information that lie behind the ideals of genetic counselling. Framing genetic information and counselling was studied in the international guidelines and in the interviews of the counsellees. Because of the differing nature of these two data sets, the question was posed in different manners for both of them. In the case of the guidelines, the framing of genetic information was studied, and, on basis of this, the consequences for genetic counselling were analysed. In the case of the interviews, where genetic counselling was discussed quite practically, the setup was the opposite: framing the actual counselling was studied first and the conceptions of genetic information then considered on this basis. This is presented in Article IV. This analysis was widened for this conclusion back to the framing of genetic information, which causes consequences for the ideal counselling practices. For this, the frames were partially renamed.

5.2.1 Framing genetic information in the guidelines and its effects on genetic counselling

The texts concerning counselling in the context of genetic testing in the international guidelines could be classified into three different frames, *diagnostics, life planning* and *threat*, each of which reflects different perspectives on genetic information and puts a different emphasis on genetic counselling. Each of them is here presented separately and summarised in Table 2. The frames were originally published in Article II.

Table 2. Frames of genetic information used in the 56 guidelines studied.

	The frame of diagnostics	The frame of life planning	The frame of threat
Conception of genetic information	Practical-medical that is used to improve health	Social-psychological that is used to plan future life, to prepare and to adjust	Historical-bioethical that is used to promote perfect health and offspring
Consequences on genetic counselling	Orientation towards information and clear presentation	Orientation towards emotion expression and coping strategies	Protection from misuse of genetics, safeguarding freedom of choice
Main users of the frame	Political bodies, professional organisations	Professional organisations, political bodies	Ethical and political bodies, patient associations
Citation	"It is recommended that all counselling be carried out under the responsibility of a qualified physician."	"Genetic tests differ from other medical tests. The results can have serious implications for an individual."	"The value of life must not be reduced to a matter of genetic inheritance."

5.2.1.1 The frame of diagnostics

In the frame of diagnostics, genetic testing was not considered to differ from other medical examinations. It was not seen as revealing deep knowledge about identity, but as serving the health purposes of an individual and the family. In the frame of diagnostics, the persons attending genetic counselling were first and foremost expected to receive appropriate information about the mode of inheritance, the disease which they are tested for, and the testing procedure. As long as genetic information is used for the purpose of medical application, and appropriately trained professionals are involved in delivering it, the possible problems were considered avoidable. Non-directiveness of genetic counselling was seen as extremely important since it was expected to guarantee the appropriate manner with which to provide the genetic information.

> "Non-directive counselling must be the rule. The main goal of genetic counselling is to help individuals or families understand or cope with genetic disease, not to decrease the incidence of genetic disease." (European Commission: 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing by an Expert Group.)

The professional who provides counselling was regarded as a translator of genetic information who needs to present the information clearly since it is considered very complex. The training of the professionals was therefore emphasised. Boundaries between different professions were constructed because genetic information was often seen as too complicated to be presented by a general practitioner. In some documents, the role of genetic information was expected to grow in medicine in general, hence the need for training in genetics for all physicians was considered all the more important.

> "It is recommended that all counselling be carried out under the responsibility of a qualified physician. Counselling of common problems by appropriately trained non-physician health care providers or non-genetic specialist physicians is acceptable." (European Society of Human Reproduction and Embryology: Best Practice Guidelines for Clinical Preimplantation Genetic Diagnosis and Preimplantation Genetic Screening.)

5.2.1.2 The frame of life planning

In the frame of life planning, genetic information was constructed as exceptional compared to other medical information because it does not add something new to the existing identity, but reveals something that has always been uniquely tied to the person. Also, the future-oriented nature of genetic information was seen as distinguishing it from other health information. Receiving genetic information was expected to have an effect on choices affecting the future.

> "Medical genetics' main concerns, however, extend beyond those of the traditional structure of medicine and the physician-patient relationship. For example: (…) (b) genetic discoveries may be predictive of future adverse events in an individual's or family member's health; (c) genetic information and the choices of the present may affect future generations (…) (World Health Organisation: Review of Ethical Issues in Medical Genetics.)

Since genetic information was considered to profoundly affect the individual's identity, the psychosocial element was expected to be included in all counselling situations where decisions concerning future life are made, and it was seen essential that the counsellor concentrates on the emotional reactions of the counsellee. Adapting information to the personal situation of each patient, and not abandoning them to make difficult decisions on their own were seen as good practices of counselling.

> "Genetic information often has a profound impact on an individual and the family. This needs to be acknowledged in the counselling process and clients given a safe environment in which to express their emotional/psychological responses. It may be appropriate that responding to an individual's emotional reactions takes priority over discussing further the genetic issues." (Human Genetic Society of Australasia: Guidelines for the Practice of Genetic Counselling.)

5.2.1.3 The frame of threat

The past of eugenics was expressed in some of the guidelines as a reminder of where the incorrect use of genetics has led before. Even though the era of eugenics was seen to have passed in most of the documents, there were suspicions in the frame of threat that the old eugenic ideas are still prevalent in genetic medicine. Education for the public was considered

important, so that people would have the knowledge to oppose the genetic information that they actually do not want, and the eugenic thoughts that may be hidden in the healthcare practices. Particularly in the guidelines of the Disabled People's International, it was clearly stated that persons with disabilities are the victims of the eugenic practices of genetics, since the value of their life is not taken into account in genetic counselling.

> "This has all happened before. It must not be allowed to happen again. (…) The genetic threat to us is a threat to everyone. The value of life must not be reduced to a matter of genetic inheritance." (Disabled People's International: Disabled People Speak on the New Genetics. DPI Europe Position Statement on Bioethics and Human Rights.)

Discrimination on the basis of genes was introduced also pertaining to insurance companies and employers, who were expected to be interested in knowing what genetic risks people have. Therefore, the confidentiality of genetic information was considered extremely important in the guidelines. Genetic information was seen as a real threat to individuals, particularly when it is predictive. Knowing the risk of a future illness was not always considered beneficial: it may increase anxiety, and choices may, at worst, be based on information that is not certain. Too much reliance on genetic information was seen as leading to genetic determinism.

> "The development of predictive medicine may indeed have positive results for public health and prevention in quite a number of cases, but its importance should not be exaggerated. It should not be presented as the key to future medicine. The risk here would arise from placing a naive reliance on the absolute power of science and genetics, or illusory expectations about the benefits they can bring." (Conference of European Churches, Commission for Church and Society: Genetic Testing and Predictive Medicine.)

In the frame of threat, even though it was considered that genetic counselling could never be non-directive and the information provided objective, counselling was considered important since without appropriate genetic counselling genetic information would be even more dangerous. The counsellor was expected to have training in bioethics, in addition to the training in genetics. The professionals who provide counselling were expected to recognise the dilemmas involved, act as gatekeepers against the misuse of science, and protect counsellees from the threats of genetic information.

> "Our society must be willing to provide for counselling when appropriate and needed, because without it the provision of diagnostic, highly predictive genetic tests is potentially harmful." (The European Association for Bioindustries: Human Medical Genetic Testing. A EuropaBio Position Paper.)

5.2.2 Framing genetic information and counselling in counsellees' interviews

The conceptions of genetic counselling in the interviews were first classified into three frames: *the frame of information, the frame of atmosphere* and *the frame of irrelevance.* These are presented in Article IV. These frames are here widened to describe the meanings attributed to genetic information, not only the counselling situation. Thus, the frames are partly renamed as the *frame of information, the frame of emotions* and *the frame of ignorance and distancing*. They are each presented separately and summarised in Table 3.

Table 3. Framing genetic information in counsellees' interviews.

	The frame of information	**The frame of emotions**	**The frame of ignorance and distancing**
Conception of genetic information	Non-exceptional health information	Deeper and more final than other health information, includes emotional burden	Not particularly influential information
Consequences of genetic counselling	No differences to other healthcare visits, emphasis on information	Emphasis on the human approach and psychological issues, unhurried and warm atmosphere	No expectations or knowledge about genetic counselling: assuming to go directly to take the test
Users of the frame	All interviewees; main frame of most of them	Nearly all interviewees, especially those having discussed a predictive genetic test	Majority of the interviewees having had a diagnostic or a carrier test
Citation	"Most important was to know what it was all about." "It was like any other blood test."	"I was regarded as an individual person and I felt very good about it."	"It was just a compulsory part of the whole process. I did not need or want it."

5.2.2.1 The frame of information

In the frame of information, genetic information was seen as comparable to any other health information that may affect the future. Thus, genetic counselling was not seen as differing from other encounters with healthcare professionals.

> "Blood is taken and it is analysed for the purposes needed, as usual." (mother of a child having had a diagnostic genetic test)

The interviewees emphasised the informational nature of genetic counselling. Some interviewees talked about a blood test, not a gene test, about which the physician first gave information. All of the interviewees used the frame of information, and some of them mainly spoke about genetic counselling through this frame. When asked to specify the most important element in genetic counselling, several interviewees named some aspect of information: a conception of what was going on, the risk figure or the actual test

result. The interviewees expected some new information, as most of them already had information on the condition, gathered from family members, the Internet or a patient association.

In the frame of information, the conception of genetic counselling was rather practical. It was expected to offer useful information about one's health in the case of diagnostic and predictive testing, and about one's genes that may affect the health of future offspring in the case of carrier testing.

> "It was of utmost importance just to know what it was all about." (a woman having had a carrier test)

5.2.2.2 The frame of emotions

In the frame of emotions, the emotional burden of genetic information was central. The ones who had had a predictive genetic test described their self-observation, getting prepared to hear the test result and contradictory feelings when facing the test result. On the one hand, the result was expected, on the other, it was feared. The feelings that the interviewees described as belonging to their testing process were the fear for the condition of the child, the anxiety of the risk for other relatives, and the exhaustion when hearing about another identical diagnosis in the family.

> "I was feeling rather down (when getting another identical diagnosis in the family) in a way, the same process was to start all over again (...) I don't think anyone claps their hands in that situation." (father of a child having had a diagnostic genetic test)

The anxiety about children's future was mentioned: who will counsel them, will the disease get worse, and will those who have not been tested develop the disease? One interviewee also described the genetic condition as a taboo in the family: she had experienced and recognised the feelings of denial and guilt and the fear of stigmatisation because of the situation. For one interviewee, the diagnosis had been a surprise. She felt that the information had not been delivered from generation to generation.

Some interviewees described the condition as dramatic because it cannot be cured and taken away – it has always been and it always will be there. The result of a genetic test was described as final. One interviewee had not wanted a predictive genetic test because she feared that it would have changed her attitude towards herself and attitudes towards her at work. Another interviewee who had undergone genetic counselling because of a predictive test had wanted to have the test because of the desire to know for sure and to make the decision and not just always think about it.

> "It does affect one more because it is oneself in question and you know it is true (the test result)." (a woman having had a predictive genetic test)

Interviewees described how the genetic testing process had affected their feelings on their future life. One said that the anxiety had increased and many practical issues like whether to move abroad or to have children had to be considered. Others, however, stressed the increased control over life and the clarity about what to do in the future. Because of a negative result concerning a predictive test for Huntington's disease, one interviewee said that she finally had the courage to think about having a family. For some, the feelings did not change after the result, as the disease and information about it had been in the family already before and it had been part of their life before the actual diagnosis.

In the frame of emotions, the most important element of genetic counselling was the warm and human atmosphere faced at the genetic clinic. Most of the interviewees felt that their feelings had been taken into account. They described the counsellor as a sympathetic person, who paid attention to the psychological issues of genetic testing. Many of them had experienced informal, listening and comprehensive counselling where their needs had been taken into account. Some interviewees felt that attending genetic counselling was different from other medical encounters because of this unhurried and warm atmosphere. The counsellees received more attention and support than they had been used to.

> "Most important was to notice how human the physician was. Of course information also is very important, but maybe even more the attitude." (a woman having had a predictive genetic test)

5.2.2.3 The frame of ignorance and distancing

In the frame of ignorance and distancing, genetic information was comparable to other health information in the same way as in the frame of information. Some counsellees did not have expectations about genetic counselling. The result of the gene test did not affect their life in a very significant way.

Some counsellees had not thought that they would receive genetic counselling, but expected to go directly to the gene test. One called counselling a compulsory part of the process, because she felt that she already knew everything needed before and just wanted to know the test result. Some counsellees were surprised that so much time and attention was paid to them. Many of the interviewees mentioned that they did not have expectations about counselling, but were "all ears". Thus, the expectations were fulfilled and often superseded.

> "I was surprised how much it was discussed, how big process it was (…) I had not thought that so much resources would be put into it, I thought I would go straight to the lab and get the problem sorted." (a woman having had a carrier test)

Some of the interviewees distanced themselves from the test result and the disease in question. As the result was negative in the case of the carrier tests, it did not have any effect on the lives of those who had had the test. They also mentioned that they had not thought about the disease even before the test, because they had trusted in the low probability. Carrier tests are performed in order to discover the risk to the children; thus, reproductive questions are always immanent. These interviewees said that selecting characteristics for the future offspring or abortion of the foetus with a genetic mutation would be difficult questions, but they had not needed to think about these because of the negative test results. Thus, they did not consider the questions relating to genetic testing to affect their own lives at all.

The interviewees who had had a diagnostic or a predictive genetic test also used the frame of ignorance and distancing. Two interviewees whose children had been diagnosed as having Connexin-26 related hearing loss and hereditary motor and sensory neuropathy talked about the diagnoses in a careless way. They made the symptoms or the disease in their families sound easy and unproblematic and called it "small potatoes". The life plans of the interviewees had in most cases already been made, and the result of the gene test did not make a big difference in this respect. The risk was not considered to particularly affect the life and choices to be made.

> "One could get cancer or get into a car accident or whatever that might affect future plans". (a woman having had a predictive genetic test)

5.2.2.4 Different framing from different counsellees

All interviewees used at least the frames of information and emotions, but their weight was different depending on the test situation of the interviewee. The frame of ignorance and distancing was used by the majority of the interviewees who had visited the genetic clinic because of a diagnostic or a carrier test, but less by those who had received genetic counselling due to a predictive genetic test. Depending on the interviewees' reason for visiting the genetic clinic, their needs for counselling were different. Those who had visited the clinic because of a predictive genetic test, had needed and received profound counselling with the discussion on the implications of the test, the future of the counsellee and issues to be considered when making the decision to have the gene test.

Interviewees who had visited the clinic because of a carrier test or a diagnostic test expressed rather different feelings. For them, counselling was mostly the situation that necessarily preceded the actual test, simply because it was a part of the protocol. They had made the decision to have the gene test beforehand and expected to have practical information before the test and the result afterwards. These interviewees did not regard genetic counselling as a very important experience in their life. Some of these interviewees wondered at the high level of the service: the expectations of the interviewees were not as profound as the service they received. The committed and involved approach faced at the genetic clinic made them feel that they were receiving good service, but it also may have increased their anxiety.

The conceptions of genetic information also differed between the interviewees. Some, mainly those who had had a diagnostic test, did not distinguish between genetic information and other health information. Those who had had a carrier test did not regard the information as actual health information at all, because the tests proved not to affect their lives. The conceptions of genetic information held by those who had had a predictive test varied. Depending on their life situation and family history, they talked about genetic information as deep information about oneself or as practical health information that may or may not affect one's life.

5.3 Practices of genetic counselling

In addition to studying the expectations directed at genetic counselling, another purpose of this study was to examine what the existing genetic counselling practices are like in European countries, and to examine how these relate to the expectations studied. The practices were investigated via a survey sent to the representatives of the National Societies of Human Genetics or other experts in European countries. Counsellees' experiences on genetic counselling were examined in the interviews and in the background review of the previous studies. The study of the experiences was intended to describe the practice of counselling in comparison to the expectations reflecting the ideals.

5.3.1 Practices described by the national experts

In the survey performed among the representatives of the National Societies of Human Genetics, or other experts of genetic testing in the countries that did not have a professional society, the respondents were asked about the legislation, guidelines and generally applied practices relating to genetic counselling. It was assumed that if legislation or official guidelines existed, these would guide the practice. It was known, however, that in most countries genetic counselling is not regulated, and, hence, generally

applied practices were enquired into (see Matthiessen-Guyader 2005, Soini 2006, Borry et al. 2007). Because the survey was directed at the societies of human genetics and they were assumed to answer in a manner reflecting the situation in the country as a whole, separate practices varying from clinic to another were not looked into.

According to the survey, in 2006 no legislation or professional guidelines existed specifically regulating genetic counselling in 13 European countries. In 9 countries, there were both legislation and guidelines, in 4 countries only legislation, and in 12 countries only guidelines. In addition, in most of the countries, there was more general legislation or guidelines that affected genetic counselling, relating for example to confidentiality or patient rights. Respondents from 35 countries reported at least some generally applied practices of genetic counselling. The results presented here were originally published in Article III.

5.3.1.1 Counselling in the context of different testing situations

Counselling in the context of prenatal testing was mentioned more frequently than other counselling situations as being covered both by legislation and national guidelines, and guided by a generally applied practice. Counselling in the context of diagnostic testing was the second most considered to be covered by regulations, and together with counselling in the context of carrier testing, it was regarded as being guided by generally applied practices as often as counselling in prenatal testing. Counselling in the context of susceptibility testing for multifactorial diseases was least often covered by legislation, guidelines or generally applied practice. The figures are presented in Article III.

The respondents who described their generally applied practices explained that counselling may take place both before and after testing, or, in the context of diagnostic testing, sometimes only before or after a test, as needed. Five respondents mentioned that counselling was performed only in appropriate settings, for instance within clinical genetics services. In the context of predictive genetic testing, offering psychological support to the counsellee was mentioned, as well as not performing such tests on children.

5.3.1.2 Different aspects of genetic counselling

The aspects relating to genetic counselling that the respondents most often mentioned their national legislation as covering were informed consent and confidentiality. These were also mentioned in the answers from some of the countries with no specific legislation for genetic counselling. Topics that were most often covered in the national guidelines were non-directiveness, confidentiality, and by whom genetic counselling may be provided. The figures are presented in Article III.

Non-directiveness and the qualifications required to perform genetic counselling were most often regarded also as being guided by generally applied practices. Respondents from 19 countries reported that only specialised medical geneticists or genetic counsellors may perform genetic counselling, whereas eight respondents said that sometimes other medical professionals provide counselling as well. Representatives from six countries thought that the non-directiveness of genetic counselling is a golden rule in their country, but nine respondents felt that this is applied only among professionals specialised in medical genetics.

Respondents from 26 countries considered that there is a generally applied practice of contacting relatives at risk in their country. Some of them explained that the relatives are contacted only through the index patient, while the others mentioned that it is possible for the professional to contact the relatives directly, with the consent of the index patient. Respondents from 22 countries regarded offering psychological support in connection with counselling as a generally applied practice. Respondents from 19 countries thought that there are generally applied practices concerning the counselling of minors or persons with diminished mental capacity. Some respondents mentioned the presence of parents, an appropriate level of counselling and counselling in clinical genetics settings as such practices. Representatives of 20 countries said that there was a generally applied practice of asking for consent for every genetic test, while in 15 countries it was reported as being applied only for some testing situations. Half of these respondents answered that written consent was required for some cases, while in other cases only verbal consent was asked for. Most often, written consent was required in the contexts of prenatal and predictive testing. In 12 countries, written consent was required for all types of genetic testing, while in four countries verbal consent was always sufficient.

There were also topics that were seldom covered in national legislation and guidelines, or regarded as guided by a generally applied practice. These were, in particular, counselling individuals from ethnic minorities and the duty to recontact the patient afterwards, and also breaching confidentiality. It was mentioned that recontacting, if there is new knowledge, occurs whenever possible. Only one representative mentioned that there are registers through which patients are contacted, if needed. Respondents from four countries said that breaching confidentiality is forbidden by the law or the guidelines, while another four mentioned that it is done only in exceptional cases and never without consent. Only three respondents mentioned having generally applied practices related to counselling individuals from ethnic minorities. They described using interpreters and involving cultural differences in the training of professionals.

5.3.1.3 Respondents' assessment of the regulation, current status and future of genetic counselling in their country

Respondents from nine countries thought that the effect of the existing regulation was positive in their country. They were satisfied with the regulation they had. "The existence of such legislation helps to standardise practical work", explained one respondent. However, respondents from 18 countries wanted more or better regulation; for example, it was stated that "national legislation should be more detailed" and that the lack of regulation had a negative effect "due to different considerations among specialists and patients as well". Respondents from ten countries thought that national regulation was not necessary, even though five of them had legislation or some guidelines. "Most guidelines which exist are helpful, but lack of legislation allows flexible practice", explained one of them. Some of these ten respondents thought that there was no need for national guidelines, because "the professional staff follows international guidelines". Those respondents who wanted more regulation of genetic counselling came from all over Europe, but especially from the new and candidate EU member states, or from countries not involved in the EU.

Respondents were asked to describe how well genetic counselling was organised in their country. Only three respondents were completely satisfied with the situation. "There is good access throughout the country and there is a sufficient number of well-trained clinical geneticists", explained one. Most of the respondents were fairly satisfied with their situation, but thought that some improvements were needed. Positive aspects that were mentioned were the access to and good infrastructure of counselling services, and the appropriate level of training and expertise. However, the gaps that were described also related to training and service provision. "There is a lack of provision in relation to the demand on genetic counselling", commented one respondent. 14 respondents considered that genetic counselling was not at all optimal in their country. It was stated that genetic counselling was not organised, or that a lot of improvement was still needed. It was also explained that there was a lack of specialists, a need for better training, failures in networking, and a lack of formal recognition of the field. Representatives of four countries thought that the organisation of counselling varied. One explained that "genetic counselling is well organised only in the capital".

More than 70% of the respondents remarked upon the lack of general genetic knowledge among healthcare professionals and the prioritisation of healthcare resources as problems in the organisation of genetic counselling in their country. More than 60% referred to the lack of legislation and guidelines and the lack of trained professionals. Representatives from ten countries thought that unequal access to counselling, sometimes related to geographical distances, was a problem. Only representatives from four countries

considered that there were language or cultural problems in counselling. Outside the list, the reasons that were mentioned included the poor quality control of practices, lack of medical insurance, lack of corporate identity among geneticists, and problems specific to a small population.

The respondents were asked to predict the changes that might happen in genetic counselling practices in their country in the near future. Even though this was an open question, 17 respondents predicted or hoped for more legally binding regulations or written professional guidelines. Some of these respondents described why this development was needed. "The new legislation I hope will make the provision of medical genetic services more uniform over the country", explained one. In some of the countries, the changes were expected to formalise the position of medical genetics. It was also thought that genetics would be more integrated with other fields of healthcare, both in primary care, and in other speciality fields. An increase in the number of professionals in the field, general improvements, and regional development were also mentioned. Two respondents were afraid that there would be no development in genetic counselling in their country in the near future. Issues that only one respondent mentioned as a future prediction were improvements in the definition of cases that needed counselling, an increase in continuous training, and that development would take place only through the private sector, or when the next generation of physicians entered in the field. It was also predicted that the patient journey through the genetic service system would become more difficult.

5.3.2 Experiences of the counsellees

The practices of genetic counselling, experienced by the counsellees, were first sought in the background review of the previous studies, in which many aspects covered in genetic counselling had been studied. They are listed in Table 4, which has been used to ground the interviews of the counsellees, but has not been published. The first column shows the number of the studies in which the issue was either offered or introduced by the respondents. The second column states in how many studies the issue was introduced in general, but the number of respondents that were satisfied with it, or how satisfied they were, was not measured. In the third column, the number of the studies in which a certain percentage of the respondents expressed the experience in question is listed. This can be compared to the other number in the column, which states in how many studies the majority of the respondents were satisfied with the experience. The last column shows the number of the studies in which a scale was used to measure the experiences. The summary of the values in scale from 0 (lowest value) to 1 (highest value) is expressed after the number of the study.

Table 4. Experiences on genetic counselling.

Experiences of genetic counselling	Total number of studies in which the issue offered as an option or introduced by the respondent (N=102)	Number of studies where the issue was introduced in general	Number of studies in which experienced by a percentage of respondents / in how many of those experienced by more than half of the respondents	Number of studies in which it was expressed on a scale / the summary of the average values in scale 0-1
Negative experiences	34	26	8/0	0
Satisfaction with information	29	9	13/12	7 / all above 0.7
General satisfaction	24	2	10/10	12 / all above 0.7
Satisfaction with the emotional aspects	17	5	4/2	8 / all above 0.7
Satisfaction with the procedural aspects	15	7	5/5	3 / all above 0.7
Lack of information	15	3	11/0	1 / 0.4-0.5
Reassuring counselling	15	5	10/5	0
Expectations met	13	2	9/9	4 / in three studies above 0.9, in one 0.3-0.4
Useful counselling	12	2	9/8	1 / 0.9-1
Clarifying counselling	11	3	7/4	1 / 0.9-1
Clear communication	11	3	8/6	0
Satisfaction with the information about the condition	10	6	3/2	1 / 0.8-0.9
Felt listened to	10	0	9/9	1 / 0.9-1
Satisfaction with the time used	9	1	7/6	1 / 0.9-1
Supportive counsellor	9	6	2/2	1 / 0.8-0.9
Received aid in decision-making	9	3	5/4	1 / 0.9-1
Satisfaction with the information about the inheritance	8	6	1/0	1 / 0.8-0.9
Competent counsellor	8	3	3/3	2 / both 0.8-0.9
Informative counselling	8	2	5/4	1 / 0.9-1
Satisfaction with the atmosphere	7	5	1/1	1 / 0.9-1
Helped to cope better	7	1	6/5	0
Difficulties in understanding	7	2	5/0	0
Dedicated counsellor	6	1	5/4	0
Satisfaction with the information about one's own risk	6	2	3/3	1 / 0.9-1
Satisfaction with the information about the risk of family members	5	4	1/1	0
Satisfaction with the information about the risk management	5	1	4/2	0
Reassuring counsellor	5	1	4/3	0
Understanding counsellor	5	0	4/4	1 / 0.9-1
Counsellor showed personal interest	5	3	2/1	0
Expectations exceeded	5	1	4/2	0
Satisfaction with the communication style	5	3	2/2	0

In the studies reviewed, the counsellees were very satisfied with the genetic counselling received. Although negative experiences were asked about in many studies and expressed, too, the majority of the counsellees did not have such experiences in any of the studies in which percentages were used. The respondents were in all studies satisfied with the counselling in general and most of them particularly satisfied with the information, communication style, procedural aspects, feeling of being listened to, usefulness of the counselling, aid in decision-making, time used, help in coping and competence, supportiveness, dedication and understanding nature of the counsellor, whereas the opinions on satisfaction with the emotional aspects, reassurance, information about the risk management and inheritance, and counsellor showing personal interest were more scattered. The studies using the scale showed a very high satisfaction with all aspects asked about.

In counsellees' interviews, the satisfaction with the counselling was also expressed. The counselling session had been experienced as positive, warm, unhurried and informative. Counsellees had felt that they were being treated as individuals whom the counsellor cared for. Many of the interviewees had received support and understanding in their situation and expressed exceptional satisfaction with a profound level of service that they had not expected. When asked about negative experiences or suggestions for improvement, two counsellees admitted that they had not understood everything that had been said or at least did not remember it afterwards. Thus, written information was wished for. Some counsellees expressed wishes that they realised were impossible: more information about prevention, faster test results and more convincing about the authenticity of the results.

Some ambiguities were also apparent in the interviews when evaluating the practices of genetic counselling. Although the counsellees appreciated the profound attitude of the counsellor that made them feel worthy of receiving genetic counselling, it also increased the anxiety of at least one counsellee who had not regarded the disease in question as very serious, but who reconsidered this due to the serious approach of the counsellor. Another counsellee was dissatisfied with the way the counsellor expressed the test result, feeling sorry about it, while the counsellee had hoped for clarification of the diagnosis and was hence happy about the positive test result.

6. Discussion

6.1 Expectations, frames and practices of genetic counselling

The main results concerning expectations, frames and practices of genetic counselling are here discussed, combining the perspectives of international guidelines, national experts and counsellees.

6.1.1 What is genetic counselling expected to be altogether?

The elements expected from genetic counselling that were emphasised in all data sets studied were related to information provided in genetic counselling. The expectations that were most often discussed in the interviews of the counsellees as well as in the background review of the counsellees' perspective related mainly to information: about one's own risk, the condition in question, inheritance, risk management and practical issues. The expected ideal genetic counselling, from the counsellees' perspective, was, thus, information-oriented. In the interviews, no other expectations were mentioned. In the review, dealing with emotional aspects did not appear as often as the expectations concerning the receiving of information.

Providing appropriate information was, of course, also one of the key expectations set by the international guidelines. They introduced the same topics to be discussed in counselling as did the counsellees in their expectations and, in addition to these, some other points, such as the limitations of the test or the existence of support groups, which the majority of the counsellees did not expect. However, in the guidelines, the main emphasis was not on information only, but issues of autonomy, consent and confidentiality were also discussed in most of them, as well as psychological sides of counselling and ethical issues, such as genetic discrimination and familial implications. These were introduced in most guidelines. In this sense, expectations differed between the data sets.

As a result, from the counsellees' perspective genetic counselling was expected to be information-oriented and practical, and, in addition to these points, from the perspective of the guidelines also social, ethical and psychological. The interviews of the counsellees also addressed a view that did not become visible in the background review or in the guidelines: some of the interviewees actually did not expect any counselling at all related to a genetic test. The differences in the expectations towards genetic counselling from the perspectives of the official documents and counsellees is understandable due to the

differing purpose and nature of the data, but, interestingly, it does demonstrate that for most counsellees genetic counselling is a rather practical event that does not require any more ethical or psychosocial considerations than do discussions with healthcare professionals in general.

6.1.2 How is genetic information framed in the counselling speech?

In order to summarise how genetic information was framed in the guidelines, produced mostly by expert bodies, and in the speech of the counsellees interviewed, the two frame analyses were compared. Both data sets included the co-existence and the confrontation between the informational emphasis in which genetic information can be compared to any other health information and the emotional involvement that includes the particular feelings that the persons visiting genetic counselling may face and the ways in which these are taken into account in genetic counselling.

The frame of diagnostics in the guidelines and the frame of information in counsellees' interviews closely resembled each other. Genetic information was constructed as non-exceptional and genetic counselling mainly regarded as an ordinary encounter between the healthcare professional and the patient in both of them. The frame of life planning in the guidelines and the frame of emotions in the interviews could also be compared. In both of these, genetic information was seen to raise specific issues regarding counsellees' feelings and plans about future life that need to be taken into account in the profound and human genetic counselling.

The third frames in the two data differed, however, strongly. In the international guidelines, ethical aspects were seen as necessary in genetic counselling. In the interviews of the counsellees, they were not articulated, unless specifically asked about, and even then they were usually ignored. Genetic testing and counselling was a rather practical event for the counsellees that did not include ethical considerations. Conversely, the irrelevant attitude towards genetic information and its delivery came up in some interviews. This, of course, was not involved in the guidelines that are created to highlight the issues attached to genetic testing and counselling. Although many elements were similar from the perspectives of the guidelines and the counsellees, the guidelines focused more on ethics and the counsellees more on practice. This is natural due to the different nature of the two data sets, but it also shows that genetic information can be given varying meanings in the lived lives of the counsellees, which is not usually expressed in the ideals of the guidelines. The communication of these two perspectives should provide some further points to consider in the development of new guidelines and practices for genetic counselling.

6.1.3 How do the practices of genetic counselling face the expectations attached to them?

According to the analyses, the most frequently highlighted and also the most apparent practice of genetic counselling was information reception. The counsellees expected information concerning different sides of genetic testing and were also satisfied with the information received. The content of the information expected to be provided in genetic counselling was expressed in the international guidelines, too. In the case of information-giving, the ideals and practices of genetic counselling seemed to correspond.

There were also other practices expected in the guidelines and reported by the national experts to be generally applied in their country, such as asking for consent and the confidentiality of genetic information. In the guidelines, the expertise and the training of the counsellor was highlighted, and this corresponded to the answers of the national experts. They explained that the practice of who may provide genetic counselling is generally applied in their country. It was also stated that non-directiveness is applied only among training specialists. The lack of trained clinical geneticists and the lack of genetic training of medical practitioners in general were mentioned as weaknesses in providing genetic counselling. Thus, both in the professional guidelines and in the professional societies, the boundaries of the profession were built and the lack of expertise in providing genetic counselling disapproved of.

According to the analysis of the guidelines, considering discrimination and familial implications belong to ideal genetic counselling. These did not appear exclusively in the analysis of the existing practices, but it does not mean that they are not discussed. In the survey sent to the national experts, familial implications were present only via the topic of confidentiality. Some of the interviewees mentioned that familial implications had been discussed. They were not, however, among the key expectations of the counsellees, and, therefore, were not often described as practices. In addition to information, psychological support was also highlighted strongly both as an expectation and as a practice. It was expected in the guidelines, but not by the majority of the counsellees. However, when describing their experiences, many of the interviewees emphasised the support and the warm approach as a whole. Most of the national experts reported psychological support as being a generally applied practice in their country.

In the future development of the genetic counselling practices, new trends in genetic testing may have to be considered. In the survey conducted in European countries, the duty to recontact the patient, breaching confidentiality and counselling persons from ethnic minorities were the topics least often regarded as regulated or guided by generally

applied practices. Alongside development in genetic research, recontacting may become more relevant in the future. Breaching confidentiality is also widely discussed in the context of informing at-risk relatives. Counselling persons from ethnic minorities will increase as immigration grows. All of these situations, in addition to all the other new or increasing situations of genetic counselling, indicate the need to consider the appropriate practices regularly.

The national experts hoped for more guidelines and standardised practice. The lack of these was regarded as an obstacle for the development of the field, and their appearance in the near future was predicted in many countries. However, one of the respondents noticed that there is no need for more guidelines since without them the practices are more flexible. The experiences of the counsellees proved that in some cases more individual consideration of the service would be useful. Sometimes, a "light" version of genetic counselling could take place. It probably would not entail all the aspects required in the guidelines, but it could serve the purpose without increasing the anxiety of the counsellee unnecessarily.

6.2 Examining the cornerstones of genetic counselling: information, support and ethics

Information and support have traditionally been considered to form the core of genetic counselling (see Fraser 1974). Although counselling has become more exact alongside the increase in genetic tests and as the practices have developed in time, this core has not changed over the decades according to this study. Information and support were repeatedly named as both the expectations and practices of genetic counselling in the data. Counsellees particularly emphasised information.

The orientation towards information partly supports the notion of the non-exceptional trend of genetic testing (e.g. Saukko et al. 2006, Green and Botkin 2003). Counsellees in particular often regarded genetic information like any other health information that is expected from medical professionals. Some of the interviewees mainly wished to have the test conducted and to receive the result and did not expect any additional service underlining their specific situation. This has also been noted in previous studies concerning counsellees' experiences (Bernhardt et al. 2000, Barr and Millar 2003, Metcalfe et al. 2007). The wish to have just the result of a genetic test without particular counselling attached also reflects patients' increased awareness and self-care (see Bradby 2009). Counsellees have often sought information about their condition

beforehand and in cases of a known family history already know a lot about the disease and its inheritance.

Although information was emphasised in counsellees' expectations, the counsellor's support and warm reception were appreciated in their experiences. The warmth of genetic counselling and the support provided formed for many counsellees exceptionally good service compared to other encounters with healthcare professionals. In previous studies on counsellees' opinions, satisfaction with the service and especially with the engagement of the genetic counsellor has also been reported (Metcalfe et al. 2000, Liede et al. 2000, Davey et al. 2005, Holloway et al. 2005, Josephson et al. 2000, DeMarco et al. 2004, Hopwood et al. 2004, Elliott et al. 2014).

While counsellees did not expect psychological support, in most of the guidelines it was one of the key expectations of genetic counselling. The introduction of psychological support in the guidelines proves that the status of the psychological model of genetic counselling is strong on the level of recommendations. Many writers (Walker 2010, Harper 2008, Weil 2003, Biesecker 2001) have argued that during the recent decades both the psychological model and consideration of the emotions of the counsellee have entered genetic counselling, forming the most essential element of it. This is apparent in the frame of life planning used in the guidelines analysed. Some of the guidelines emphasised support over information, paying attention to the specific features of genetic testing that oblige careful genetic counselling.

In addition to the practical elements of information and support necessarily belonging to the process of genetic counselling, ethical discussion has also always been strongly related to genetic testing. The most obvious ethical questions derive from the eugenic past of genetics that dominated the practice of genetic counselling in the first half of the 20th century (see Harper 2008). Due to the need to distance itself from eugenics, the model of non-directive genetic counselling was born in the latter part of the 20th century and has served as the dominant model of genetic counselling for the past decades (Harper 2008, Clarke 1997, Weil 2003). Genetic counselling has focused on the needs of the individuals as opposed to state direction, but critics have stated that the new eugenics can be found precisely in the counselling of individuals (Petersen 1999, Raz 2009, Raman and Tutton 2010). The counselling practices are not purposely eugenic, but it has been argued that the individual's choices may lead to eugenic consequences, and leaving the responsibility purely to the counsellee falsely diminishes the responsibility of the professionals, the healthcare system and the society (Kerr and Shakespeare 2002). In addition to the ethical considerations relating to reproductive decision-making, other ethical debates, too, have

centred on genetic testing. Geneticisation and genetic determinism have been used to refer to the (overly strong) control of genes over health, illness and life in general in our thinking (e.g. Lippman 1991). The confidentiality of genetic information, discrimination on its basis and the power of predictive genetic information have also been discussed. These all stress the possible threat introduced by the delivery and the use of genetic information and by choices based on it.

Whereas information and support constitute the core of the genetic counselling session rather consistently from all perspectives analysed in this study, the bioethical discussion around genetic testing varies according to the context in which genetic counselling is discussed. In the guidelines, the frame of threat, emphasising the bioethical aspects of genetics, was strongly present. They partly shared the discourse presented in the literature, introducing a variation of ethical aspects to be considered in genetic counselling.

The most frequently mentioned challenge in the frame of threat of the guidelines was the familial nature of genetic information. It was seen that counsellors should not only be able to have a confidential relationship with the counsellees, but they should also consider the impact that the genetic information may have on the family. The balance between confidentiality and the duty to warn the at-risk relatives is a subject of continuous discussion (Aktan-Collan et al. 2011, Gaff et al. 2007, Koch and Nordahl Svendsen 2005, Claes et al. 2003). Therefore, genetic counsellors are expected to have sufficient training in the particularities of genetic information. Another ethical challenge particularly addressed in the guidelines of the professionals was the autonomy of the counsellee, which was expected to be improved with counsellee's consent. The concept of autonomy closely relates to the concept of non-directiveness that is also expected from genetic counselling, but not as often as autonomy in the guidelines. The reason why autonomy is a more general ideal than non-directiveness may derive from the ambiguous concept of non-directiveness (e.g. Biesecker 2000, 2001, Kessler 1997). According to the analysis of the guidelines, non-directiveness is still a commonly shared view, but it is not as often regarded as clinical guidance as the autonomy of the counsellee. The "non-directive era" of genetic counselling, during which non-directiveness was emphasised over other aspects (Weil 2003) seems to have slightly passed, although the principle of non-directiveness may still be valid.

Eugenics was also mentioned in the guidelines. They expected the counsellors to understand the ethical questions posed by counselling, especially in the context of prenatal diagnostics. It was considered important that genetic professionals actively avoid discrimination and eugenic thinking and, for this, training in bioethics was

considered desirable. The Disabled People's International especially produced the frame of threat in which eugenic thinking was linked to genetic counselling. In some of the guidelines, fear of discrimination on a genetic basis and reliance on the power of genetics in defining people's lives were seen as ethical challenges posed by genetics. Although not as commonly used as the frames of diagnostics and life planning, the frame of threat was very strong in the documents in which it was employed. Although co-existing, the frames were emphasised differently in the guidelines: while the professional organisations focused more on practical issues, such as communication and the content of information, ethical boards and the organisation of the disabled put the emphasis on ethical questions, such as autonomy and discrimination.

In other data, bioethics was not articulated. It is natural that in the quantitative approach in which the practices of genetic counselling were studied in the survey to the national experts and in the background review of counsellees' perspective, the ethical issues were not actually introduced, although it was reported in the survey that confidentiality and consent are usually guided by legislation or guidelines and applied as general practices. However, the ethical issues were not further detailed. The ethical discourse was also lacking in the counsellees' interviews. Since prenatal diagnosis was excluded from the study, precisely because of the specific ethical issues clearly related, the ethical discourse was expected to diminish and change. However, it was not expected to be completely missing, although the theme was specifically introduced in the questions. The counsellees did not regard it as an important frame of genetic counselling at all. At least they distanced it from their own particular situations.

The discourse on bioethics concerning genetic testing has been described as changed. Geneticisation has been opposed similarly than medicalisation some years earlier (Árnason and Hjórleifsson 2007). The discourse on genetic market of possibilities, life enhancement technologies (Helén 2004), economy of hope (Novas and Rose 2004) and fragmented gene (Rose 2001) seems to have overcome the discourse on genetic determination and eugenics. Kerr (2004) has declared that genetic determinism is dead and the discussion should move on. Raz (2009) argues that the discussion over new eugenics has turned to emphasise individual's own values and conceptions of life. As the practices of genetic testing have become more ordinary, the ethical discussion around genetic information and its use and delivery is getting more fragmented.

However, a more fragmented ethical discourse should not mean the end of ethical discussion. While genetic testing increases, we are forced to consider the new threats that may be related to the practice. It is worth asking whether we recognise the possible

future misuses of genetics. It is clear from the past that the ideas that are now disapproved strongly were once approved and employed very rapidly while the political and the social environment were appropriate. The old eugenic practices of state direction in reproductive decision-making are now rejected, but along with the increase in genetic testing, individuals may be required to take more responsibility in their health behaviour on the basis of their personal genetic information. Genetic determinism may be given new forms that can be applied in sociopolitical decision-making if there is no appropriate ethical discussion attached to the development of genetic technologies.

As the practice of genetic counselling is changing in tandem with technological and social changes, so, too, are the expectations directed at it. The changes in the bioethical discussion reflect this. Different aspects concerning information are expected to be delivered in all genetic counselling situations, but support and especially ethical aspects are not and it is not felt necessary to consider these in all situations. Genetic information has become more ordinary and the expectations, practices and the bioethics of genetic counselling are now more dispersed. Discussion around these topics is still very much needed.

6.3 From ordinary to life-shaking genetic information: genetic exceptionalism and life politics re-visited

The genetic counselling expected, as detected in the guidelines analysed, was rather uniform, independent of the origin of the guidelines. The same aspects were repeated from one guideline to another, forming a quite consistent vision of what may be seen as ideal genetic counselling. It might mean that genetic counselling is not a very complex activity, but actually a rather simple encounter between the professional and the counsellee. It could equally mean that the problematic issues have not been examined properly in the guidelines as the uniform view is so strong (Rantanen et al. 2008a). Kerr and Shakespeare (2002) argue that the international guidelines for genetics have created a global discourse of bioethics that is disappointing in its content as the regulations seek to balance different interests. It could also be that seeking the general expectations concerning genetic counselling does not allow the recognition of the differences among particular testing situations.

In the survey about the regulations and practices of genetic counselling in European countries, the differences between testing situations were expressed. Different testing situations were asked about separately. Prenatal diagnosis, usually involving the most

challenging psychological and ethical elements, was most often regarded as regulated or generally agreed on. Thus, there has been a need to discuss these topics together. Also, counselling in the contexts of diagnostic testing, carrier testing and predictive testing was quite often considered to be covered by regulations or generally applied practices. The practices described in some answers of performing genetic counselling only before or after a genetic test in the context of diagnostic testing, or offering psychological support particularly in the context of a predictive test, show that there are specific practices related to specific counselling situations. These cannot be gathered as the answers in the study differed in their accuracy, but it can be concluded that they exist.

In the interviews of the counsellees, the differences between the counselling situations was evident. Depending on interviewees' reason for visiting the genetic clinic, their needs for counselling were different. The counsellees who had visited the clinic because of a predictive genetic test had needed and received profound counselling with discussion on the implications of the test, the future of the counsellee and issues to be considered when making the decision on the gene test. For interviewees who had visited the clinic because of a carrier test or a diagnostic test, counselling was a situation that necessarily preceded the actual test, simply because it was part of the protocol. They had made the decision of having the gene test beforehand and expected to have practical information before the test and the result afterwards. These interviewees used the frame of ignorance and distancing; they did not regard genetic counselling as a very important experience in their life. Some of these interviewees even wondered at the high level of the service. In the background review of the counsellees' perspective, the importance of different expectations and experiences could have varied between and within the studies due to the disease tested for, life situation, the research frame and the culture, but these could not be studied in the review as the number of the studies in each category was too small. However, most of the counsellees in most of the studies expected certain basic issues from counselling, related mainly to information. In addition, there are counsellees who expect more profound counselling, reassurance and support. Hence, for some counsellees, basic counselling serves the purpose, and could even be regarded as the ideal, whereas some counsellees require more a profound and more psychological approach.

Even though the decisions made in different genetic testing situations vary, it has been written that their seriousness, their relevance for the person in question, for family members and future-generations and their irreversibility connect all these situations (Shiloh 1996). This has been questioned since the discussion has recently represented more the non-exceptional trend of genetic testing (Green and Botkin 2003). The changing nature of genetic testing has also changed the discussion on genetic exceptionalism. As

the treatments are developing, genetic information is becoming more ordinary due to technology, and genetic testing is beginning to be used in more diverse contexts, genetic exceptionalism can hardly be justified in the way that it has been understood (see Evans and Burke 2008). Since genetic information may be more ordinary or more life-shaking, depending on the situation in which it is delivered, it is problematic to treat counselling situations under the same umbrella of genetic testing.

Now, instead of emphasising genes, the implications of the genetic test and the individual situation in which the counsellee is are emphasised as the basis for best counselling practices (Saukko et al. 2006, Guimarães et al. 2013, Wang et al. 2004, Will et al., 2010). Thus, counsellees receiving genetic counselling due to predictive testing, for instance, may indeed need more "exceptional" service because of the difficult situation they may face in case of a positive test result. They may have to carefully consider whether to want to know about the future risk of becoming ill and the effects this knowledge has on living one's life. However, the experiences of the interviewees who had discussed having a predictive test varied, too: one of them had been more certain about what she wanted and what sort of counselling she needed than the other two. Those who had had a diagnostic or a carrier test did not regard the process as exceptional, but this might have been different in the case of different conditions, or if the carrier tests of both the person with a family history and the spouse had been positive. Thus, the test type implies what is required from genetic counselling, but the expectations also need to be defined situationally.

The variation between genetic testing situations needs to be considered in the future when more and more genetic information will be available. Huntington's disease is a common example of the devastating effects of genetic information. It is a progressive, genetic brain disorder that causes uncontrolled movements and leads to emotional problems and cognitive decline (Genetics Home Reference 2013). A person having a predictive test for Huntington's disease may find out that there is a certainty of becoming ill at some point of his or her life. Since no one can tell when this will happen and in which way, all life plans may be affected by this information, as was expressed by a counsellee in the interview study. In the case of diagnosing a disease that is already causing clinical symptoms, this also affects life plans but no more if the diagnosis is based on a gene test than other type of medical test. However, diagnosing a hereditary condition may change the relations and the dynamics in the family. Prenatal testing also often forces the consideration of life plans. If a family expecting a baby finds out that the baby has, for instance, chromosomal abnormalities, they will have to consider the future in the light of this information when making the decision on whether to continue the pregnancy.

On the other hand, in many situations the decisions based on genetic information do not affect life plans, or there may be no need to make decisions in the first place. In case of a condition that is not regarded as serious or life affecting, genetic information does not have a significant role in life. In the interview study conducted, hereditary motor and sensory neuropathy was regarded as such condition. It includes slowly progressive distal weakness, muscle atrophy, and sensory loss due to an inherited peripheral neuropathy (National Institution of Neurological Disorders and Stroke 2014). It was described as being of no great consequnce, and the parent interviewed did not see that knowledge of the hereditary condition would affect him having more children or decisions on his children's lives.

Questions related to reproduction are usually the reason for wanting a carrier test. There is a desire to know whether there is a risk that future offspring will inherit the possibly serious condition in the family, for example INCL leading to intellectual and motor disability and premature death (Genetics Home Reference 2013), as was the case in the interview study. However, it is very unlikely that both of the spouses are carriers of the disease, and since it is inherited recessively, the child has no risk of developing the condition. The interviewee with a family history of INCL had received a positive test result, but since her husband had not, no decisions based on this genetic information needed to be made.

Genetic information has been described as representing well the increasing risk information based on which choices are ever more made. In Beck's (1992) risk society, individual choices and dealing with risks are obligatory. Individuals are expected and encouraged to act responsibly, govern themselves, make rational decisions, take preventive actions and to improve their lives based on the available information (Petersen 1999, 2010, Rose 2001). Rose (2001) argues that genetic information differs from other health information in the way that people can reinterpret it and make choices about their own genetic futures. There are cases in which people do exercise life politics based on genetic information. They make reproductive choices, choices about starting a family, choices about profession and choices about where to live. An interviewee who had had a predictive genetic test for Huntington's disease explained that she had dared to think about starting a family only after the negative test result near the age of 40 and hoped that it was not too late. An interviewee who had had a positive predictive test for hereditary cancer said that the concrete effect of the test on her life was the decision to live in Finland due to the regular follow-up, although she had earlier planned to move abroad. These are big decisions in individuals' lives, and they interestingly describe how life politics can be exercised based on genetic information. Giddens (1991) refers to life politics as politics

dealing with identity, lifestyle and decisions concerning one's life. In addition to big and unique choices, life politics can also be exercised on the everyday level. The interviewee who had found out about her risk of cancer mentioned lifestyle habits concerning eating and smoking as preventive actions, although there was no certainty about whether she would become ill or not. Her everyday habits represent well the responsible actions of individuals who are expected to govern themselves on the basis of their personal risk information.

As genetic information sometimes forces one to make difficult decisions, but sometimes no decisions at all, or decisions that do not seem to affect one's life, there is an obvious need for individually tailored genetic counselling. There is no single genetic counselling protocol to be applied in all counselling situations, and some of the counsellees need and expect more profound service than others. Such greater flexibility of genetic counselling protocols due to counsellees' expectations and psychological needs has been demanded before (e.g. Guimarães et al. 2013). There are situations in which medical conclusions need to be made based on genetic information, medication and surveillance for instance, and other situations in which the decisions to be made are non-medical and more personal, such as having children. These different situations may require different expertise from the healthcare professional. The practices of genetic counselling should vary as the contexts of genetic information are diverse. Everyone needs the basic elements of information and some support in genetic counselling, but more profound psychological elements and detailed information could be limited to situations in which they are needed. The guidelines that form a rather uniform vision of ideal genetic counselling could be in need of improvement in the future in this respect.

6.4 Study limitations

In this study, the expectations directed at genetic counselling have been examined, and the frames through which genetic information and genetic counselling are produced have been sought in two data sets. The practices of genetic counselling were requested from national experts and identified in the background review of previous studies concerning the counsellees' perspective. Hence, the data studied has been varied. It has therefore been difficult to pose the same research questions to all data sets. Although all these perspectives have been combined here to provide answers to the questions asked, it has to be noted that the purpose and the nature of different data sets differ, which makes comparing the answers between them complex.

The purpose of this study has been to examine the general expectations, frames and practices of genetic counselling. This has not allowed recognition of the differences among particular testing situations, which may highlight the need for more particular and tailored counselling in the different contexts of genetic testing. Since the study has focused on general expectations and practices, such particularities might not have been found in the data even if they existed. For example, the topics most frequently found in the guidelines can be interpreted as being the most important for genetic counselling in general, but it has to be noted that these may not always be the most important in particular situations. Thus, studying the expectations and practices of genetic counselling related to particular diseases or life situations could provide different answers to the questions asked. However, the purpose of the study was to map out the field of counselling in the context of genetic testing, not to concentrate on any specific situation. The limitation of this perspective has to be taken into account in the interpretation of the results.

It also has to be noted that the questions asked related to expectations and practices. Some important aspects related to the goals of genetic counselling, such as coping and the empowerment of the counsellees, were not introduced in this study because the goals of the counselling were not the focus of the study. This would also be an important theme for future research, although the goals of genetic counselling are difficult to examine due to subjective expectations and the sensitive social and ethical discussion around counselling attached to genetic testing.

In the study of the guidelines, the expectations presented regarding genetic counselling were based on their quantitative appearance in the data. Thus, they may not be the most important factors of genetic counselling in every situation, but those most applicable in all situations. For example, non-directiveness, which was not among the key topics in the guidelines analysed, may have been occupied an extremely important position in the guidelines covering prenatal genetic testing and counselling. As the guidelines reviewed had different purposes, the detail in which they addressed genetic counselling was extremely variable. They differed from one other by purpose and form, and dealt with different topics: some regulated a specific topic, such as a child-testing policy, while others discussed issues related to genetics on a more general level. Since there were very few guidelines that were specifically written to address genetic counselling, it was necessary to include all the guidelines that mentioned it in the analysis. While it could be asked whether all the guidelines analysed were suitable for providing data, these considerations were taken into account while performing the study. The guidelines were reviewed critically and their differences recognised during the analysis.

In the survey sent to national experts on genetic counselling, the respondents were asked to describe the regulations and practices of genetic counselling considering the situation in their country. The answers varied in their accuracy and according to the selected respondent. Some of the respondents may not have known about every guideline in their country. There may be legislation on different levels, and some of the respondents may have included more general health-care legislation, whereas others focused only on genetics. Some of the respondents answered more generally about genetic testing and did not focus on counselling. Therefore, the answers were not completely comparable. Some descriptions may have also reflected the opinions of only one person, although the request was that the respondents would describe the situation from a more general perspective. It is also possible that some NSHGs may not have been fully informed about all the practices of genetic counselling in their country. Considering these shortcomings, the survey however provided important information about the regulations and practices of genetic counselling in European countries.

In the background review of the studies performed on counsellees' experiences, the studies were difficult to compare because of the variety of the data, methods and research schemes used. The review brought forth only topics most often mentioned in the studies. The quantitative summary of the views of the counsellees from different studies generalised their experiences and may have missed some important, although less frequently discussed, issues. Because of these limitations in the review, it was decided that it would be used only to serve as background information for the interview study and not be published as such.

Although the interviews conducted among counsellees having visited a genetic clinic in Finland provided good data for frame analysis, it has to be noted that this data presented the opinions of only 10 counsellees from one genetic clinic. Thus, no universal conclusions can be made on the basis of these single experiences of the counsellees, although they brought forth interesting aspects. The group of interviewees also affected the frames in which genetic counselling was constructed. For example, the experiences of the interviewees who had discussed having a predictive test varied: one had been more certain about what she wanted and what sort of counselling she needed than the other two. Those who had had a diagnostic or a carrier test did not regard the process as exceptional, but this might have been different in the case of different conditions. It also has to be noted that prenatal genetic testing was excluded from the study because it was considered to involve specific ethical issues, the examination of which was not desired in this study. Involvement of these contexts probably would have emphasised the frame of emotions and the topics relating to psychological support and diminished the role of the

frame of ignorance and distancing. However, this frame would have remained, although less weight could have been put on it. Despite the limitations of the study, the interviews allowed the profound discussion of genetic counselling, the analysis of its framing and consideration of its future development from the counsellees' perspective.

The limitations of the different parts of the study have been carefully considered in the collection and analysis of the data. Despite these, the study has provided important and applicable information on the conceptions of the expected genetic counselling and how this is met in the current practices. In the future research on genetic counselling, a survey sent to a larger group of the professionals of genetic counselling would reduce the shortcomings in the study directed at national experts. Newer guidelines would also deserve further analysis of the development of the expectations towards genetic counselling. The perspective of the counsellees is ever more important as an object of study as genetic testing increases and the tests are used for more diverse purposes.

6.5 What is the future of genetic counselling?

Due to the increase in genetic testing and in the variation of situations in which genetic information is given, the ideals and practices of genetic counselling need to be reconsidered. The discussion on the public economy and on the changes in healthcare services affects this practice along with all other healthcare services. Over the decades, many professional shifts have taken place, and the realm of genetic counselling has expanded constantly (Walker 2010). Summarising simply, the prevalent models of counselling have changed from the eugenic to the non-directive, and from the non-directive to the psychological model, although these have also co-existed. What will be the next era of genetic counselling? It has been argued that an important shift will take place in tandem with genome-wide testing, personalised medicine and the increase in direct-to-consumer genetic testing (Harris et al. 2013, Walker 2010, Guttmacher and Collins 2003).

Many professional guidelines created for counselling in the context of genetic testing regard it as exceptional compared to other healthcare services. Counsellees, on the other hand, often conceptualise genetic tests as any other tests and counselling as any other information on the test, and they do not always have specific expectations or needs. The exceptionalism of the guidelines and the non-exceptionalism of many counsellees arise from different perspectives. The counsellee is usually referred for only one type of genetic testing. The expectations for counselling depend on the reason of the test and on the

information the counsellee may have gathered beforehand. Counsellees always articulate their personal experiences, while professionals need to have a more general view on the counselling. These two approaches are naturally very different, and this needs to be recognised in the analysis of the expected genetic counselling.

Genetic counselling professionals cannot base the counselling offered only on the wishes of the counsellees, since they need to guarantee the appropriate provision of information and support that the counsellees may not understand to expect. Genetic counselling needs to be tailored individually, but professional skills must be utilised in order that the quality of genetic counselling in each situation is ensured. While the counsellees' perspective is based on their individual experiences, the guidelines created by professionals often have an "on the safe side" approach so that any hint of eugenics or psychosocial harm will definitely be avoided. It may be that, along with the new genetic tests made possible by next generation sequencing, this perspective will even strengthen. Ever more perfect counselling attached to genetic testing may be required, although the counsellees may simply want information about the capabilities of the tests available.

The professionals may also wish for more guidance on genetic couselling. In the survey to the national experts, they hoped for more guidelines and standardised practices. Their lack was regarded as an obstacle for the development of the field, although one of the respondents noticed that there is no need for more guidelines since without them the practices are more flexible. The ideals of the existing guidelines are not very clear and useful for practical work in genetic counselling since they cover all the frames analysed and compromise between them. The fact that the guidelines provide a set of somewhat contradictory recommendations, if examined as a whole, restricts their capability to actually guide practical genetic counselling very strongly. If the frames are employed separately, their ideals may provide assistance to genetic counsellors in defining the desired outcomes of the different counselling situations (Rantanen et al. 2009). Wang et al. (2004) consider that, as the use of genetic information in healthcare grows, the outcomes that are expected from genetic services will be all the more important to determine. Their divisions between educating and informing, providing support and helping cope, and facilitating informed decision-making was also apparent in the use of frames in the guideline documents, although, in addition, the element of threat was also expressed. This division supports the conclusion that the frames may be used as a tool to define the ideal genetic counselling in each specific case. The use of frames may increase genetic counsellors' truthfulness towards themselves and transparency of the goals of genetic counselling (Rantanen et al. 2009). Despite this analytical division,

genetic counsellors may wish for more regulation or guidance in specific issues of genetic counselling to safeguard the appropriate practice.

On the other hand, the ideals and the practices of genetic counselling also seem to be becoming more fragmented. According to this study, the basic elements of genetic counselling, information and support, remain, but their emphases vary, and the uniform ideals are being replaced by more individualised and situational ones. The related bioethical discussion is also becoming more diverse and the emphasis of this discussion is beginning to be more on the enabling instead of repressive tones. The non-exceptionalist trend of genetics is more dominant, and alongside the increase in genetic information genetic counselling is beginning to be regarded more as an integral part of the healthcare services. As Rose (2001) has suggested, in the post-genomic era, whether we are already living in it or simply approaching it, thoughts about the gene are becoming more complex and fragmented.

Jallinoja (2002) stated in her dissertation that the ethics of genetic testing needs to be negotiated situationally, since no universal rules can be applied to all counselling situations. The same pattern seems relevant also in the case of the ideals and expectations directed at genetic counselling. The differences between the counselling situations should be taken into account when creating new recommendations and dividing resources in genetic clinics. If the majority of numerous genetic testing situations deal with diagnostic and carrier gene tests, in the contexts of which counsellees do not usually expect profound discussions and psychosocial support but simply to receive a test result clarifying their diagnosis or carrier status, it may not be appropriate to apply the general ideal of genetic counselling, safeguarding all the aspects ever assessed to be involved in genetic testing.

While the practices of genetic counselling need to be flexible due to the differences in genetic testing situations, and the recommendations could be tailored for each counselling situation, the ethical aspects and guidance need to considered at the same time. There should be balance between these two aspects. Even, and especially, in the era of individually tailored counselling, there is a need for guidelines that prevent eugenic practices and maintain the social and ethical discussion. These guidelines simply need to be considered from the clinical perspective when applied. There also needs to be a balance between the appropriate level of informational, psychosocial and bioethical training for professionals providing genetic counselling. As the number of genetic tests is set to increase, counselling may be provided more and more by other healthcare professionals who do not have all the training. This may be sensible in the case of many genetic tests, but at the same time adequate genetics training has to be secured for all

healthcare professionals. There are also counselling situations that need to be offered by professionals with profound training in genetic counselling. The balance between "light genetic counselling" and the assurance of the recognition of ethical and psychosocial aspects needs to be sought in every aspect of genetic counselling.

As the entire process of genetic testing is changing as a result of the increasing number of tests, susceptibility testing, next generation sequencing, personalised medicine and direct-to-consumer testing (Harris et al. 2013, Pagon 2002), the future roles of the genetic counsellor in different counselling situations should also be considered. Intraprofessional conflicts have always belonged to medical professionalism (Annandale 1998), and there are already differences between genetic counselling, provided in the genetic clinics, and information provided in other clinics about genetic conditions. This should be more carefully considered in the future. It has been argued that genetic exceptionalism has served to maintain a privileged position for genetic information in the clinic, requiring safeguards in its communication to patients (Ross 2001, Will et al. 2010). At the same time, professional boundaries have been built, and clinical geneticists and genetic counsellors constructed as the only suitable professionals to provide this exceptional genetic information. In the guidelines analysed, as well as in the answers of the national professional societies, the professional boundaries were built and the lack of expertise disapproved. This professionalism may have to be reconsidered if the genetic information to be delivered increases as predicted, and if its contexts become ever more diverse. The future professional roles of genetic counsellors have been examined (e.g. Finucane 2012, Harris et al. 2013, Guttmacher and Collins 2003), but the topic requires further discussion.

Although it may be necessary that the ideals of genetic counselling become more diverse according to the differing needs of the counsellees, there may also be interest in gaining a common sense of the basic issues to be covered in the counselling. As samples increasingly cross borders, particularly in the case of genetic tests for rare diseases, many molecular genetics laboratories would like to be sure that there are ideals that are accepted and followed in different countries (Rantanen et al. 2008a). If there is a desire to have a common view of what ideal genetic counselling in its basic form is, the discussion needs to focus particularly on the differences among the guidelines and on the reported practices.

The issues that were somewhat contradictory in the guidelines were: who may request a genetic test and provide genetic information in different testing situations; how much training in the psychological and ethical aspects of genetics counsellors should

receive; should information be objective or adapted to counsellee's situation; who should pass the information on to at-risk relatives; should autonomous decision-making be guaranteed, even if the patient does not want it; and should genetic counselling include the perspectives of those who live with a genetic disease or their representative associations (Rantanen et al. 2008a). In the survey on the regulations and practices of genetic counselling in European countries, the issues that were least often regulated or reported as having generally applied practices were: the duty to recontact the patient; breaching confidentiality; counselling persons from ethnic minorities; and counselling in the context of susceptibility testing for multifactorial diseases. These, however, may have growing importance in the future (Rantanen et al. 2008b). The duty to recontact the patient has inspired wide discussion (Letendre and Godard 2004, Hunter et al. 2001), and it can be seen as becoming increasingly important as some of the information received in genetic counselling may become outdated. Breaching confidentiality when contacting at-risk relatives is also a subject of wide discussion (Aktan-Collan et al. 2011, Gaff et al. 2007, Koch and Nordahl Svendsen 2005, Claes et al. 2003). Large-scale immigration in European countries may place new demands on genetic services as there will be new genetic conditions and socio-cultural issues surrounding counselling (Ibarreta et al. 2003). Susceptibility testing for multifactorial diseases has been estimated as creating potentially the biggest future demand for genetic testing services (Ibarreta et al. 2003, Yang et al. 2003, Evans et al. 2001).

Thus, in the future, new trends in genetic testing are to be considered. The issues raised by the survey, as well as some other issues estimated to have a growing role, such as pharmacogenetics and tests offered directly to consumers, may require more attention. They were covered only in a few guidelines. The guidelines generally did not go into such detail, but the lack of these topics may also indicate that they are still emerging and the time of consensus on them will follow later. It may also be that generally applied practices are not relevant in cases such as these, and that there will be a need to consider the appropriate practices separately in each case.

All in all, the future genetic counselling will occur in a more diverse environment, with counsellees requiring individualised counselling according to their needs. This demands a more flexible practice. At the same time, greater regulation and guidance is expected to make the basic practice of genetic counselling more uniform in different countries and clinics. As genetic testing improves and new issues relating to complex diseases, medication and commercialisation, for instance, need to be considered in the counselling practice, and, at the same time, the discussion about the public economy and the development of healthcare systems in general takes place, the development of future

genetic counselling practice will need to find a balance between the different demands, expectations and ideals. The two directions, standardising the practice with regulation and guidelines on the one hand and considering varying situations more individually and flexibly on the other, may occur simultaneously. It may also be that the development varies among the countries. In the survey conducted in European countries, many of the respondents who were satisfied with the existing national regulation and with the organisation of genetic counselling, came from the older EU member states, whereas the majority of the respondents who wanted more regulation came from the newer and candidate EU member states, or from European countries not involved in the EU. It may be that in these countries the standardisation of practices of genetic counselling will take place, whereas in those countries where this has already happened, the practices will be reconsidered and the recommendations rewritten due to the need for more flexible practice. The history of genetic counselling, as well as the considerations for the future presented here, reflect Western thinking. The ideals and practices of genetic counselling outside of this may differ from what has been described here.

The significance of this study lies in summarising what is expected from genetic counselling, how these expectations are met and how this affects the changing practice of genetic testing and associated counselling. It has concluded that, in the future, guidelines and the diversity of testing situations should be considered: not all genetic testing situations require the same certain counselling protocol. Some new topics have been raised that may require further attention in future, such as recontacting counsellees in the event of new information, or counselling in the contexts of multifactorial diseases. Better communication between the professional perspective and the perspectives of single counsellees could provide ideas in the development of the practices and in the division of resources in the counselling clinics. Given the future increase in genetic tests, this will be ever more important to consider.

7. Conclusions

In this study, three different perspectives were researched to examine the expectations addressed to genetic counselling and the conceptions of genetic information behind those expectations, and to evaluate how the current practices of genetic counselling encounter the expectations. International guidelines were analysed to discover the main content of the counselling ideals and the frames in which genetic information is discussed. The National Societies of Human Genetics or other experts were asked about the existing regulations and practices of genetic counselling in 38 European countries. Ten counsellees having visited a genetic counselling clinic were interviewed about their expectations and experiences. A background review of the previous studies concerning the counsellees' perspective was conducted in order to recognise the main topics identified before.

On the basis of the analysis of all these three perspectives, the ideal genetic counselling seems to consist of information about the test, the condition, the risks and their management; and of support in adjusting to this information and in decision-making concerning the test and its result. The guidelines constitute a rather consistent vision of the expected genetic counselling, but the reality experienced by the professionals and the counsellees is more diverse. The basic information is required in all counselling situations, but otherwise the ideals vary according to the test type and situation in question. These differences are also taken into account in the practices of genetic counselling, as reported by the national experts. Genetic counselling services receive all in all very good grades from the counsellees, which means that they are given time and attention more than in the healthcare services in general. Some counsellees feel that they have received surprisingly good service. Since some of the reasons for visiting a genetic counselling clinic are regarded by the counsellees as rather small and insignificant, it can be asked whether receiving exceptional service is justified simply on the grounds that there will be discussion of issues related to genes.

Genetic information is increasingly regarded as non-exceptional compared to other health information, especially by counsellees. The professional bodies may wish to maintain the boundaries of expertise that have been created to stress the importance of the clinical specialty of medical genetics. They may also wish to make a clear distinction between today and the eugenic era of genetic counselling. The non-exceptional view, the increase in the number of genetic tests and the diversity of contexts in which these tests are used and genetic information applied force us to ask whether genetic tests should be regarded as an entity at all. The concept of the gene has caused all these situations to

be treated with special safeguards, but this study supports the emerging trend that the individual situation and the desired outcomes of genetic testing should define the ideals and practices of genetic counselling more than the fact that the test result is gained via genes.

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Appendixes

Appendix 1: Guidelines concerning genetic counselling

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Appendix 2: Survey to NSHG's and other experts

Dear Genetics Colleague, EuroGentest

This survey is part of the European project EuroGentest "Genetic testing in Europe – Network for test development harmonization, validation and standardization of services" (http://www.eurogentest.org). The aim of this survey is to gather information about the practices, guidelines, recommendations and general principles **related to genetic counselling and ethical issues concerning genetic testing and counselling**. Your responses are very important for our aims to improve the quality of genetic counselling services in connection with genetic testing in Europe.

This questionnaire is sent to all national societies of human genetics in Europe. The board, the chairperson of the board or someone nominated by the board is asked to fill in the questionnaire **on behalf of the society**. The results of the survey will be disseminated through the project website and through other professional forums.

We kindly ask you to answer to the questionnaire provided in the following link preferably by November 18th, 2005. [LINK] If you have any questions, please do not hesitate to contact us.

Sincerely,
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Genetic counselling defined by the European Society of Human Genetics (http://www.eshg.org/ESHGgeneticservicesbckgrnd.pdf):

Genetic counselling is a communication process, which deals with the occurrence, or risk of occurrence, of a genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to 1) understand the medical facts of the disorder; 2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives; 3) understand the options of dealing with the disorder; 4) choose the course of action which seems appropriate to them in the view of their risk and their family goals and act in accordance with that decision; and 5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

In addition, genetic information may be given by a range of healthcare practitioners in the context of genetic testing or screening.

In the following survey we ask about the existence of national legislation, guidelines/recommendations or generally applied practices related to counselling in different types of genetic testing situations and related to ethical issues concerning genetic counselling and genetic testing.

Background information:

Country:
Person/Group who gave this information:
Email-address to be used in case of need for clarification:

In general, do the following exist in your country in the context of genetic counselling?

Legislation	YES / NO
Professional written guidelines	YES / NO
Generally applied practices[1*]	YES / NO

Explain if applicable to your situation:

I. Legislation and guidelines

1. Are there national legislation or guidelines that specially mention the following clinical situations? Please answer yes or no.

Clinical situation	**Legislation or other legally binding regulations**	**Written professional guidelines**	**Please give addresses of websites or references to the literature, or briefly describe the guidelines and the bodies that have produced them.**
Diagnostic testing			
Carrier testing			
Presymptomatic testing (e.g. HD)			
Predisposition testing (e.g. BRCA)			
Predisposition testing for multifactorial diseases (e.g. Alzheimer disease)			
Prenatal diagnosis			
Preimplantation diagnosis			
Testing of children and adolescents			

1 ** Practices that are generally recognisable in your country in the view of a geneticist

2. Are there national laws or guidelines that specially mention the following topics? Please answer yes or no.

	Legislation or other legally binding regulations	**Written professional guidelines**	**Please give addresses of websites or references to the literature, or briefly describe the guidelines and the bodies that have produced them.**
By whom / where can genetic counselling be performed			
Consent of the patient			
Non-directiveness of genetic counselling			
Counselling persons from minority ethnic groups			
Counselling minors or persons with diminished mental capacity			
Psychological support during the process of genetic testing			
Informing at-risk relatives			
Confidentiality			
Duty to recontact (Recalling counsellees if there are developments in testing, diagnosis or treatment)			

3. How do you think that the existence/lack of such legislation and guidelines affect the practical work?

4. Are there national laws or guidelines that specially mention the following topics? Please answer yes or no.

	Legislation	**Written professional guidelines**	**Please give addresses of websites or references to the literature, or briefly describe the guidelines and the bodies that have produced them.**
Direct testing (genetic tests supplied directly to the public)			
Genetic testing in employment situations			
Genetic testing and insurance			
Paternity testing			
Sex selection			
Intellectual property rights and patenting			
Genetic databases			

II. Generally applied practices

1. Are there practices that are generally applied in your country in the context of the following genetic counselling situations? (E.g. a practice to counsel before and after a carrier test)

Clinical situation	No	Yes, please describe
Diagnostic testing		
Carrier testing		
Presymptomatic testing (e.g. HD)		
Predisposition testing (e.g. BRCA)		
Predisposition testing for multifactorial diseases (e.g. Alzheimer disease)		
Prenatal diagnosis		
Preimplantation diagnosis		

2. Do you have a generally applied practice of informed consent in the context of genetic testing? If so, for which types of testing it is used and is it written or verbal?

	Written	Verbal	No practice of consent	Explain if you wish
Diagnostic testing				
Carrier testing				
Presymptomatic testing (e.g. HD)				
Predisposition testing (e.g. BRCA)				
Predisposition testing for multifactorial diseases				
Prenatal diagnosis				
Preimplantation diagnosis				
Testing of children and adolescents				

3. Are there practices that are generally applied in your country related to the following topics?

	No	Yes, please describe
By whom / where can genetic counselling be performed		
Non-directiveness of genetic counselling		
Counselling persons from minority ethnic groups		
Counselling minors or persons with diminished mental capacity		
Psychological support during the process of genetic testing		
Informing at-risk relatives		
Decision to breach confidentiality		
Duty to recontact (Recalling counsellees if there are developments in testing, diagnosis or treatment)		

4. Do you think that genetic counselling is well organised in your country? Please explain.

5. If the counselling situation is not optimal in your country, what factors affect this?

a. Lack of trained professionals
b. Prioritization of healthcare resources
c. Lack of general genetic knowledge among healthcare professionals
d. Lack of legislation and guidelines
e. Unequal access to counselling
f. Language and cultural problems
g. Geographical distances
h. Other, what:

6. What is your prediction about what changes will happen within genetic counselling practices in your country in the near future?

7. Other comments:

Thank you for your responses!

Appendix 3: Request for an interview

Hyvä TYKS:n perinnöllisyyspoliklinikan asiakas,

Pyydämme Teitä osallistumaan perinnöllisyysneuvontaa koskevaan haastattelututkimukseemme *"Perinnöllisyysneuvonta ja geneettinen tieto potilasnäkökulmasta"*. Osallistuminen on täysin vapaaehtoista, eikä kieltäytyminen vaikuta mitenkään mahdolliseen tulevaan hoitoonne tai tuleviin perinnöllisyysneuvontoihin.

Geenitestien yhteydessä annettavaa perinnöllisyysneuvontaa pyritään kehittämään koko ajan, jotta se vastaisi mahdollisimman hyvin sitä saavien ihmisten tarpeisiin ja auttaisi kyseessä olevaa sairautta ja geenitestiä koskevan tiedon saamisessa ja geenitestausta koskevien päätösten tekemisessä. Turun yliopiston lääketieteellisen genetiikan osastolla toteutetaan viisivuotista EU:n rahoittamaa EuroGentest-tutkimushanketta (www.eurogentest.org), jonka yhtenä päämääränä on kehittää perinnöllisyysneuvontaa Euroopassa.

Tutkimuksessa on jo tarkasteltu lainsäädännön, genetiikan yhdistysten ja perinnöllisyysneuvontaa antavien ammattilaisten näkökulmaa neuvontaan. Tutkimukselle on kuitenkin erittäin tärkeää saada kattava ja syvällinen kuva myös siitä, mitä asioita perinnöllisyysneuvontaa saavat asiakkaat pitävät tärkeinä neuvonnassa. TYKS:n perinnöllisyyspoliklinikka osallistuu tutkimukseen, koska yhtenä osana tätä työtä tutkijat toivovat voivansa haastatella 10-15 TYKS:n perinnöllisyyspoliklinikan asiakasta. Siksi otamme yhteyttä myös Teihin. Tutkimukselle olisi erittäin hyödyllistä, jos haluaisitte osallistua siihen ja näin auttaa perinnöllisyysneuvonnan kehittämistyötä.

Tutkimus toteutetaan haastatteluna, joka nauhoitetaan, jos haastateltava on tähän suostuvainen. Haastateltava voi keskeyttää haastattelun missä vaiheessa tahansa. Haastattelussa käsitelty tieto on luottamuksellista. Haastateltavan nimeä ei kirjata tutkimuksen tiedostoihin eikä se tule ilmi mistään tutkimuksen raportista. Haastattelussa käsitellään saamaanne perinnöllisyysneuvontaa, sen vaikutusta mahdollista geenitestiä koskevassa päätöksenteossanne sekä perinnöllisyyden merkitystä kyseessä olevassa sairaudessa ja siihen liittyvässä neuvonnassa. Haastattelijana toimii väitöskirjatutkija Elina Rantanen, joka ei itse anna perinnöllisyysneuvontaa. Voitte valita itsellenne sopivan haastattelupaikan ja -ajan. Haastattelu voidaan toteuttaa TYKS:n perinnöllisyyspoliklinikalla, Turun yliopiston lääketieteellisen tiedekunnan rakennuksessa, kotonanne tai muussa valitsemassanne paikassa. Jos Teille koituu osallistumisestanne matkakustannuksia, ne voidaan korvata Teille jälkikäteen. Mikäli olette suostuvainen osallistumaan tutkimukseen, pyydämme Teitä miettimään etukäteen itsellenne parhaiten sopivaa haastattelupaikkaa ja -ajankohtaa välillä 1.2.-1.3.2008. Haastattelu kestää noin 1-1,5 tuntia.

Haastattelut analysoidaan, ja analyysin tulokset yhdistetään aiempien lääketieteellisissä julkaisuissa kuvattujen perinnöllisyysneuvonnan potilasnäkökulmaa käsittelevien tutkimusten analyysiin. Nämä tulokset on yhdessä tarkoitus julkaista kansainvälisessä yhteiskunnallis-lääketieteellisessä julkaisussa. Julkaisussa ei paljasteta haastateltavien henkilöllisyyttä. Julkaisusta tulee osa väitöskirjatutkija Elina Rantasen väitöskirjaa. Sen tuloksia käytetään hyväksi myös EuroGentest-hankkeen kehittämistyössä.

Tutkimuksen onnistumiselle on erittäin olennaista saada haastateltavia TYKS:n perinnöllisyyspoliklinikalla neuvontaa saaneista asiakkaista. Jos olette suostuvainen osallistumaan haastattelututkimukseen, pyydämme Teitä ystävällisesti täyttämään oheisen suostumuslomakkeen ja postittamaan sen oheisessa kirjekuoressa **1.2.2008** mennessä. Merkitkää suostumuslomakkeeseen, haluatteko, että Teihin otetaan yhteyttä puhelimitse tai sähköpostitse haastatteluajankohdan sopimiseksi, vai haluatteko itse ottaa yhteyttä tutkimusryhmään. Voitte vetäytyä pois haastattelututkimuksesta vielä suostumuslomakkeen lähettämisen jälkeen.

Voitte kysyä lisätietoa tutkimuksesta milloin tahansa tutkimuksen perinnöllisyyspoliklinikan yhteyshenkilöltä, erikoislääkäri Marja Hietalalta (puh. 02 333 7453, sähköposti marja.hietala@utu.fi) tai väitöskirjatutkija Elina Rantaselta (puh. 02 333 7250, sähköposti elina.rantanen@utu.fi).

Toivomme, että koette tutkimuksen mielenkiintoiseksi ja tärkeäksi ja haluatte olla siinä mukana. Kiitämme jo etukäteen vaivannäöstänne!

Ystävällisin terveisin

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Appendix 4: Interview framework

Perinnöllisyysneuvonta ja geneettinen tieto potilasnäkökulmasta

Haastattelurunko

1. Taustaa:
- Minkä vuoksi haastateltava on ollut perinnöllisyysneuvonnassa, sairauden/tilanteen kuvailua?
- Kuinka kauan haastateltava on tiennyt sairaudesta/riskistä?
- Onko sairautta ollut perheessä aikaisemmin/muilla perheenjäsenillä?
- Kuinka paljon perheessä on ollut tietoa sairaudesta?

2. Neuvontaan hakeutuminen ja odotukset:
- Miten haastateltava/perhe on hakeutunut/päätynyt perinnöllisyysneuvontaan?
- Onko neuvontaan hakeutumista pitänyt miettiä pitkään?
- Miksi haastateltava on halunnut neuvontaa? / Onko halunnut?
- (Täsmennystä motiiveihin: perhesuunnittelu, tulevaisuuden suunnittelu, huoli perheenjäsenistä, sairauden ehkäisy/hoito…)
- Mitä odotuksia haastateltavalla oli neuvonnasta? Entä ennakkokäsityksiä?
- (Täsmennyksiä odotuksiin: tietoa sairaudesta, tietoa omasta riskistä, tietoa perheenjäsenten riskistä, tietoa hoidosta ja ehkäisystä, tietoa geenitestistä, geenitestin tuloksen kuuleminen, vahvistus omalle tiedolle, tietoa mahdollisista toimenpiteistä, psykososiaalinen tuki)
- Kuinka paljon tietoa sairaudesta ja riskistä haastateltavalla oli ennen neuvontaa? Mistä se oli peräisin?

3. Testaus- ja neuvontaprosessi:
- Keitä kaikkia terveydenhuollon ammattilaisia haastateltava on tavannut ko. sairauden takia?
- Mitä kaikkea geenitestiprosessiin on kuulunut: kuinka monta neuvontaa, testi vai ei testiä?
- Kuinka nopeasti haastateltava pääsi neuvontaan?/Miten kauan prosessi on ajallisesti kestänyt?

4. Neuvonta ennen geenitestiä / testiä harkittaessa:
- Mitä kaikkea neuvonnassa käytiin läpi?
- (Täsmennys: käytiinkö läpi seuraavia asioita:
  - lääketieteelliset faktat taudista
  - hoitomahdollisuudet
  - periytyvyys
  - testin luotettavuus
  - positiivisen testituloksen todennäköisyys
  - testituloksen psykososiaaliset vaikutukset
  - vaikutukset perheenjäseniin
  - luottamuksellisuus & vakuutus/työ
  - lastensaantiin liittyvät kysymykset)
- Käytettiinkö neuvonnassa kuvia tms. apuvälineitä tai saiko haastateltava kirjallista materiaalia? Mikä näiden merkitys oli?
- Mikä oli tärkeintä tässä neuvonnassa?
- Oliko neuvojaa helppo ymmärtää? Missä asioissa oli vaikeuksia?

- Mitä muita asioita kuin tiedon saamista neuvonta sisälsi?
- Pitikö ensimmäisen neuvonnan yhteydessä tehdä päätös testistä?
- Auttoiko neuvonta päätöksenteossa? Miten?
- Kuinka kauan neuvonta kesti? Oliko se riittävän pitkä?

5. Neuvonta testin jälkeen:
- Miten haastateltava sai testin tulokset?
- Mitä asioita jälkineuvonnassa käytiin läpi?
- Mikä oli tässä neuvonnassa tärkeintä?
- Pitikö tämän neuvonnan yhteydessä tehdä päätöksiä tulevasta?
- Saiko neuvonnasta riittävästi tukea?
- Keitä terveydenhuollon ammattilaisia haastateltava on tavannut testituloksen jälkeen?
- Olisiko haastateltava tarvinnut lisää tukea jälkeenpäin?
- Saiko haastateltava tietoa lisätuesta ja tukiryhmistä? Olisiko tarvinnut?
- Olisiko haastateltava kaivannut lisätietoa jostain?

6. Neuvonnan arviointia:
- Täyttyivätkö ennakko-odotukset neuvonnassa?
- Mikä jäi neuvonnasta päällimmäisenä mieleen?
- Jäikö haastateltava kaipaamaan jotain neuvonnasta?
- Miten haastateltava yleisesti arvioisi saamaansa perinnöllisyysneuvontaa?
- Saiko haastateltava tukea päätöksentekoon? Olisiko kaivannut enemmän tukea?
- Ohjailiko neuvoja haastateltavan päätöksentekoa?
- Olisiko haastateltava halunnut enemmän itsenäisyyttä vai apua päätöksentekoon?
- Minkälainen suhde haastateltavalla oli neuvojaan? Erosiko se jotenkin suhteista muihin terveydenhuollon ammattilaisiin?
- Mitkä olivat tärkeitä asioita neuvonnan antajassa? Erosivatko ne jotenkin asioista, joita haastateltava yleensä pitää tärkeänä terveydenhuollon ammattilaisissa?
- Mitkä tekijät neuvojassa vaikuttivat luottamuksen syntymiseen? Taustatiedot esim. koulutuksesta?

7. Geenitesteistä ja neuvonnasta yleensä:
- Näkeekö haastateltava, että geenitestien ja muiden lääketieteellisten testien välillä on jotain eroa? Jos, niin mitä?
- Onko haastateltava miettinyt geenitesteihin liittyviä eettisiä kysymyksiä? Jos, niin mitä?
- Liittyikö omaan tilanteeseen joitain tällaisia kysymyksiä?
- Onko haastateltavalla jotain pelkoja geenitesteihin liittyen? Esim. vakuutusten menettäminen, leimautuminen, syrjintä?
- Onko haastateltavalle tärkeää, että geenitestin tulokset ja neuvonta ovat luottamuksellisia? Tärkeämpää kuin muiden testien yhteydessä?

8. Perhe ja suku:
- Minkälainen rooli perheellä ja suvulla on ollut neuvontaan hakeutumisessa ja sen jälkeen? Onko tämä ollut erilainen sairauden perinnöllisyyden vuoksi?
- Onko tietoa pitänyt levittää perheessä? Onko tämä ollut vaikeaa?
- Miten muu perhe on suhtautunut?
- Onko haastateltavan suhde perheeseen ja sukulaisiin muuttunut neuvonnan/geenitestin jälkeen?

9. Oma tilanne nyt & tulevaisuus:

- Miten neuvonta ja geenitesti ovat vaikuttaneet haastateltavan elämään ja tulevaisuuden suunnitelmiin?
- Miten sairaus/riski vaikuttaa haastateltavan elämään arkipäivässä?
- Elääkö haastateltava varmuudessa vai epävarmuudessa sairauden suhteen?
- Minkälaista on / on ollut elää ”riskissä”?
- Minkälaisena haastateltava näkee ko. sairauden suhteessa muihin sairauksiin?
- Onko haastateltava kokenut geenitestistä/neuvonnasta seuranneen jotain negatiivista? Entä positiivista?
- Tuleeko haastateltava vielä olemaan yhteydessä perinnöllisyysneuvojan kanssa? Missä yhteydessä?
- Toivooko haastateltava yhteydenottoa perinnöllisyyspoliklinikalta? Missä tapauksessa?

- Onko jotain yleisiä terveisiä, palautetta tms.?