Improving Access and Quality of Genetic Counselling in Clinical Care in Sweden

- The Value of eHealth Solutions and a Validated Outcome Measure

Rebecka A. Pestoff
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Gene
tic counselling is increasingly important for investigations into 
hereditary diseases in the field of clinical genetics. The increase in 
demand is due to the discovery of more genetically caused diseases, 
increased complexity and awareness of genetic testing. However, access 
to genetic counselling is limited worldwide, as in Sweden, and not always 
offered as needed, because there is a lack of trained professionals, such 
as genetic counsellors and geneticists. Additionally, genetic counselling 
is difficult to evaluate as there is no validated quality measure for genetic 
counselling in Swedish. This work investigates important factors for 
improving access to and quality of genetic counselling in Sweden.

The studied factors include such as the genetic counsellors, the use and 
implementation of eHealth technology, and the possibility to evaluate 
these areas using a valid outcome measure in Swedish. This dissertation 
consists of four studies. The first is a questionnaire study that found that 
ge genetic counsellors in Sweden play an integral role in patient care and 
access, and provide quality patient support throughout the clinical 
encounter. However, it also found that there was a lack of trained genetic 
counsellors and that they were overly burdened with administrative work, 
such as sample handling, billing and making appointments. This reduced 
the genetic counsellors’ time spent directly with patients, thus hampering 
patient access. The second study investigated healthcare professionals’ 
pre-pandemic perceptions of using a specific eHealth technology 
providing genetic counselling via video or telephone, termed telegenetic 
counselling (TGC) throughout this dissertation. Findings showed that 
TGC was considered appropriate, believed to increase patient access and 
autonomy, and improve patient care. Yet, the healthcare professionals 
expressed some reluctance and identified many barriers to using TGC, 
such as the lack of evidence, and anticipated issues with technology and 
resources. Nevertheless, taking place during the COVID-19 pandemic, 
the third study investigated the feasibility of rapid implementation of 
TGC in a real, clinical context. Both healthcare professionals and patients
found TGC acceptable, useful and satisfactory, and TGC also improved access to genetic counselling during the pandemic. The implementation of TGC proved effective in regards to the overall goal of genetic counselling: increased patient empowerment after genetic counselling. This was measured by the newly adapted patient-reported outcome measure in genetic counselling in Swedish, the GCOS-24swe. The fourth study performed a psychometric evaluation of the GCOS-24swe and showed validity, reliability, and responsiveness of the outcome measure. Therefore, the GCOS-24swe provides a useful clinical quality measure to inform developments in genetic counselling practice, individualised patient care, and evaluation of implementation efforts in Sweden. Finally, a synthesis of these research findings results in a suggested implementation strategy for TGC in the clinical context. In summary, this dissertation identifies ways to improve the access to and measure the quality of genetic counselling in Sweden.

Keywords: Genetic counselling, empowerment, outcome measures, eHealth, implementation
Klinisk genetik utreder personer för olika ärftliga sjukdomar. Genetisk vägledning är en viktig del i denna process. Genetisk vägledning innebär information och stöd till personer under utredning, för att hjälpa dem förstå och fatta beslut om olika saker, t.ex. genetisk testning. Genetisk vägledning sker främst genom samtal som ska vara anpassade till personens behov och målet är att stärka personen i sin egen situation.


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Glossary and Abbreviations

This list provides an overview of central terms and abbreviations used in this dissertation.

**Behaviour Change Wheel (BCW):** A tool to help guide and design behaviour change necessary for the implementation of innovations.

**Capability-Opportunity-Motivation Behaviour model (COM-B):**
Used to analyse behaviour change necessary for individuals to apply a desired behaviour for implementation to occur.

**Electronic Health (eHealth):** the cost-effective and secure use of information and communication technologies in support of health and health-related fields, as defined by WHO.

**European Board of Medical Genetics (EBMG):** A non-profit educational and professional association, that aims to establish professional standards of education, training and practice in human and medical genetics and genetic counselling in Europe.

**Evidence-based medicine (EBM):** Tested evidence provides a reliable knowledge base for use in healthcare.

**Genetic and genomic counselling:** a patient-centred process involving a healthcare provider giving support to a patient in making informed decisions regarding a genetic condition and its implications. Is provided by a specifically trained healthcare professional, i.e., a genetic counsellor, nurse or medical doctor.

**Genetic counsellor (GC):** a specifically trained professional providing genetic counselling.


**Healthcare professional (HCP):** health professionals maintain health in humans through the application of the principles and procedures of evidence-based medicine and caring.
**Implementation**: the process of putting an innovation to use within a specific setting.

**Implementation determinants**: factors that act as facilitators or barriers, affecting the outcome of implementation efforts.

**In-person genetic counselling**: to provide genetic counselling in a physical meeting, face-to-face.

**Patient-reported outcome (PRO)**: PROs are patients’ subjective experiences of health, treatment, quality of life and satisfaction regarding the patient’s health condition.

**Patient-reported outcome measure (PROM)**: a measure of PRO, usually a self-administered questionnaire.

**Service delivery mode (SDM)**: different ways of offering genetic counselling, i.e., TGC or in-person.

**Telegenetic counselling (TGC)**: to provide genetic counselling consultations using telephone or video technology.

**Telehealth Usability Questionnaire in Swedish (TUQswe)**: a PROM to measure the usability of distance genetic counselling provided by telephone or video. Based on the English version of TUQ.

**Visiba Care (VC)**: a virtual care platform using digital working methods. CE-marked as a medical technology product, ISO-certified and GDPR-adapted.
LIST OF PAPERS

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IV. Rebecka Pestoff, Henrik Danielsson, Peter Johansson, Marion McAllister, Cecilia Gunnarsson (2022) Translation and cross-cultural adaptation, and validation of a patient-reported outcome measure for genetic counselling: the Swedish Genetic Counselling Outcome scale (GCOS-24swe) *Manuscript*
INTRODUCTION

This research stems from different academic research fields, genetic counselling, eHealth and implementation science, with different terminology and important aspects. This background provides a general overview that describes these aspects and links them to describe the more specific context in which this research took place and the aims of the research. A short scientific background is provided for each specific area. Challenges and knowledge gaps in the fields are identified, as well as relevant factors for implementation, and how they are connected to publication in this dissertation.

Genetic counselling

Genetic investigations are used in mainstream and specialised healthcare to find a genetic contribution to common and rare diseases. Newly discovered genes and mutations are frequently added to the list of causes of hereditary disease. More than 72% of the over 6000 known rare diseases have a genetic origin (Nguengang Wakap et al., 2020). In addition, combined susceptibility variants in the genome, together with degrees of environmental effects, can result in common multifactorial and polygenic diseases, due to additive negative effects on the proteome (Kierczak et al., 2022). This knowledge is used to provide individualised precision medicine, including genetic and genomic counselling. For the affected patient, family or relative, it is important to understand how genetics can contribute to the disease. This is what happens through the process of genetic, also including genomic, counselling. Genetic counselling is provided by specifically trained healthcare professionals (HCPs), usually a medical doctor, a trained nurse or a genetic counsellor (GC). The latest census of genetic counsellors was carried out by the Transnational Alliance of Genetic Counselling in 2018 (Abacan et al., 2019) and estimated that there were almost 7000 GCs in 28 different countries (Abacan et al., 2019). The genetic counsellors usually have
heterogeneous academic backgrounds and are commonly trained in multidisciplinary fields, including molecular biology, communication, medical ethics and psychology. The training aims to provide a highly skilled professional with appropriate competencies to provide genetic counselling in accordance with the definition below.

**Definition**

An early definition of genetic counselling was provided by Fraser in 1974 (Fraser, 1974). In 2006, the National Society of Genetic Counselors (NSGC) Taskforce in the US updated the definition:

> 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)

Genetic and genomic counselling enables individuals to gain information on complex genetic and genomic information and to understand available choices, in a patient-centred manner (McCarthy Veach; Resta et al., 2006; Skirton et al., 2015). Information and understanding are equally important for individuals to reach informed decisions and for appropriate individual adaptations to the current condition. The decisions concern different choices, regarding genetic testing; prenatal testing; prophylactic operations or regular surveillance; how to spread information to relatives at risk; or participating in research, to name a few. The overall aim of genetic counselling is to improve the patients’ empowerment.
Empowerment in genetic counselling

Patient evaluations of empowerment are being prioritised by health policies around the world (McAllister et al., 2012) Empowerment can occur through for example patient-centred interactions with a healthcare provider, engaging in patient organisations, and patient self-education. However, there seems to be a lack of conceptual consensus regarding the meaning of empowerment as the term is used in a variety of ways (Risling et al., 2017). Therefore, the definition of empowerment in this research is based on the construct according to patients and HCPs in clinical genetics that was identified by McAllister and colleagues (McAllister et al., 2012) and includes the following five dimensions:

- Cognitive control (sense-making - understanding the condition, why it happened, what help & support is available - “knowledge is power”)
- Decisional control (having some options for managing the condition/risk and being able to make informed decisions between options),
- Behavioural control (able to do something to reduce harm or improve life for self/children/at-risk relatives/descendants)
- Emotional regulation (reflecting effective coping and adjustment)
- Hope for the future (for self/relatives/future descendants).

Challenges in genetic counselling

The number of available genetic tests and the increase in genetic investigations leads to higher demand for genetic counselling. Originally, genetic counselling was provided by a medical doctor. However, medical doctors in the speciality are scarce, and since a world-wide expansion in the 1990s, genetic counselling has transitioned towards specifically trained, non-medical healthcare professionals called genetic counsellors. Genetic counsellors around the globe have been found to have similar roles, but they come from various professional and academic backgrounds and training is not globally harmonised. Differences in the role appear depending on a variety of medical systems and legislation, affecting GCs’ model of practice, as well as the level of professional recognition. In some countries (i.e., the US, Canada, UK and France), the
process of becoming a genetic counsellor is regulated, meaning that the profession is recognised as a healthcare profession by the respective government, requiring specific educational and quality standards and certification (Abacan et al., 2019). However, in most parts of the world, the profession is still unregulated (Abacan et al., 2019; Cordier et al., 2016; Middleton et al., 2017) including in Sweden (Pestoff et al., 2020). Something that is common around the world, however, is the lack of professionals trained to be genetic counsellors, which hampers the provision of adequate and timely genetic counselling (Abacan et al., 2019). An estimated need for GCs was 6-12 GCs/million inhabitants (Clinical Genetics Committee of the Royal College of Physicians, 1991) in 1991, a number that is expected to have risen since. Nevertheless, in Sweden, with 10 million inhabitants, this amounts to approximately 100 GCs. However, in 2022, there were only 35-40 professionals working as genetic counsellors in Sweden (Swedish Association of Genetic Counsellors, 2023). Limited access to GCs is one challenge. Another crucial challenge is how to measure the main outcome goal in genetic counselling, i.e., patient empowerment, in clinical genetics (Payne et al., 2008), which will be further discussed in section 1.4.

**Progression of the genetic counsellor role**

In recent years, advancements regarding the professional GC role have flourished, some of which will be discussed in this dissertation. Genetic counsellors are increasingly being used in clinics, both in Sweden and around the globe, but there is still a lack of specifically trained non-medical professionals (Abacan et al., 2019; Stoll et al., 2018). These advancements have been achieved thanks to national and international collaborations. Several non-governmental organisations have supported these improvements, such as the European Board of Medical Genetics (EBMG) and the Transnational Alliance for Genetic Counseling among others. These organisations facilitate international collaboration, research and education in the field of genetic counselling. In Europe, the EBMG sets common quality standards for genetic counselling education, by providing accreditation of MSc programmes and recognition of...
professional competence, including individual certification. In 2022, the first Swedish MSc in Genetic Counselling programme was awarded EBMG accreditation (Linköping University).

However, additional ways are still needed to increase access to genetic counselling, which leads us to consider the role of eHealth. Evidence regarding how eHealth-solutions, such as video consultations, can be sustained in healthcare is still lacking (James et al., 2021).

**eHealth and genetic counselling**

When the research in this dissertation was initially conceived in 2014, only 308 publications from studies on general eHealth were found in PubMed. However, the amount has steadily increased to over 1000 publications containing *eHealth* recently, with many more expected to come.

**Definition**

Many terms are used interchangeably, such as digital health, telemedicine and E-health, however, the following definition by the World Health Organization is the one used in this dissertation:

… the cost-effective and secure use of information and communication technologies (ICT) in support of health and health-related fields. It encompasses multiple interventions, including telehealth, telemedicine, mobile health, electronic medical or health records, big data, wearables, and even artificial intelligence. The role of eHealth has been recognized as pivotal in attaining overarching health priorities such as universal health coverage (UHC) and the Sustainable Development Goals (World Health Organization)

Telemedicine, including video or telephone consultations, is now a common alternative to in-person healthcare. Thus, telemedicine and eHealth could provide a feasible way to improve access to genetic counselling in Sweden, by narrowing the identified demand-provider gap
in clinical genetics services, apparent world-wide (Abacan et al., 2019). The rationale for this is partially based on the Swedish context, a highly tech-oriented country, and partially based on the nature of the genetic counselling communication process. Genetic counselling rarely requires a physical exam, rather verbal and non-verbal communication is used, which builds on mutual agreement and relationship building with the patient (Harper, 2010). It includes gaining the patient’s trust and establishing a working alliance (Erby et al., 2021) to support the patient to make informed decisions. Commonly, emotions surface and subtle cues of body language and facial expressions are used by the genetic counsellor to assess how to proceed in the sessions to achieve the goals (Veach et al., 2007). During a video consultation, the genetic counsellor should be able to make visual assessments and build rapport, to reach the goal.

Swedes are highly adept at using the world wide web, and most do so daily, for various reasons. The Swedish Internet Foundation, a non-profit organisation, reported in 2022 that 94% of the population over 18 years of age used the internet and that 83% were able to use an eHealth application. Among the retired part of the population (over 65 years) the corresponding numbers were 80% and 54%. One quarter of the population over 18 years had a digital healthcare consultation in 2022, and 83% reported using an eHealthcare service in that year (The Swedish Internet Foundation, 2022).

**Is telegenetic counselling a solution?**

Due to a lack of professional genetic counsellors globally (Abacan et al., 2019), increased use of eHealth solutions in healthcare (Greenhalgh et al., 2020; James et al., 2021), and COVID-19-related restrictions, research on alternative service delivery models in genetic counselling has flourished (Brown et al., 2021; Danylchuk et al., 2021; Green et al., 2022). Providing genetic counselling, this way is referred to as telegenetic counselling (TGC) from here on. Previous research has shown high levels of satisfaction, acceptability and feasibility among both patients and
providers (Gorrie et al., 2021; James et al., 2021; Platten et al., 2012). The COVID-19 pandemic resulted in a huge advancement in research on telemedicine (James et al., 2021), also regarding TGC. It showed that both patients and providers were satisfied with video and telephone consultations, compared to in-person ones (Brown et al., 2021; Danyłchuk et al., 2021; Green et al., 2022). However, to our knowledge, there were no studies on the implementation and use of TGC service delivery models during the pandemic in Sweden.

Implementation science

Implementation refers to ideas and plans that are realised in actions. Implementation is described as “a planned process and systematic introduction of innovations and/or changes of value” (Grol & Wensing, 2005). Implementation science, therefore, can help in the understanding of behaviours required to enable the use of TGC in a real-world setting. Implementation results can be strengthened when involving stakeholders in the process, for example through co-creation and co-design. (Greenhalgh et al., 2016; Vargas et al., 2022).

Implementing innovations in healthcare

Historically, implementation science has its origins in evidence-based medicine (EBM), where evidence is tested and provides reliable knowledge for use in healthcare. This requirement for evidence was also presented by HCPs as a finding in Study II. The original vision of EBM was to improve healthcare by making decisions based on rigorous research evidence. However, it became apparent that evidence alone could not change outcomes in healthcare, and implementation science grew from the need to explain and influence determinants important for the successful implementation of healthcare innovations. (Nilsen, 2010). When New Public Management was introduced, new methods and decisions were required to be based on research that proved their effectiveness and financial benefits. The Swedish government created the institution “Statens beredning för medicinsk utvärdering” (SBU, The
Swedish Agency for Health Technology Assessment and Assessment of Social Services) that still to this day gathers and provides the current research and economic evidence base for decisions to be made regarding healthcare in Sweden. To facilitate the spread and use of evidence-based practice and knowledge, the government and the healthcare regions in Sweden have now agreed to work according to the concept of “Kunskapsstyrning” (i.e. knowledge management). (Nilsen, 2010, pp. 36-39).

Implementation research studies the how and the what, that affect the process of adoption of an innovation. Implementation outcomes can be explained by studying the complex interactions between different factors on different levels.

Factors affecting implementation of telegenetic counselling

Factors influencing the implementation of innovations are called determinants and work like facilitators and barriers. Complex interactions between these determinants affect the implementation process and outcome. To better understand, show and predict the results of an implementation effort, these determinants can be systematically investigated using a framework. (Nilsen, 2010; 2015, pp. 71-86). One model, combined by Nilsen (2020) (Nilsen, 2020) from previously published research in the field, visualises relevant determinants involved in an implementation process (Figure 1). The suggested determinants are explored in this dissertation concerning the implementation of telegenetic counselling in Sweden. The following determinants are discussed in more detail: the Context in which the research is conducted; the Adopters, in this case the genetic counsellors and medical geneticists; the Outcomes, for measuring implementation and quality of TGC; and the End-users, meaning the patients’ perspective. The Implementation Object in this research is the telegenetic counselling platform (Visiba Care). Finally, following the synthesis in the Discussion chapter, some potential Implementation strategies are suggested, although not specified in the aims of any included study.
The implementation object

Our implementation object is tele-genetic counselling using an instrument called Visiba Care (Visiba Care), which, however, was not the focus of this research, since developing and changing the principles of the platform was beyond the scope of this research. That said, initial mapping of the field, supplemented with findings from Study II did identify the necessary prerequisites for the platform to be used. It must:

1. Provide both visual and audio communication
2. Be easily accessible to both healthcare providers and patients
3. Provide secure identification, to ensure patient safety

These requirements were fulfilled by the Visiba Care (VC) healthcare platform. VC was granted to the Department of Clinical Genetics approved by Region Östergötland, as part of a larger, regional trial.
Visiba Care was developed in Sweden in 2014 and initial demonstrations indicated that our prerequisites were met: the ability to host secure video meetings with multiple participants; providing a chat function and the ability to share visual information; easy accessibility and a user-friendly interface. Security is achieved by using the Swedish BankID, which is equivalent to showing a passport. BankID is used by government authorities, certified according to the Swedish e-ID quality mark and approved by the EU framework for e-identification, the eIDAS Regulation (BankID; Visiba Care).

The implementation object, Visiba Care, is here considered a healthcare innovation, which according to Rogers (2003) is an object, idea or praxis that is considered new by a person or adopter (Rogers, 2003). Providing genetic counselling through Visiba Care was new to the healthcare professionals at the Department of Clinical Genetics in 2020, and due to its innovative nature, there was quite naturally a lack of evidence. However, in implementation, it is important to integrate practical knowledge with research knowledge, and evaluations of outcomes. This evidence can reduce barriers to successful implementation, such as innovation resistance. Outcome evaluations can be considered an important step to achieve this evidence (Nilsen, 2010, pp. 109-125) thus defining a knowledge gap regarding factors affecting the use and implementation of TGC in Sweden.

**Outcome measures in telegenetic counselling**

As noted in Figure 1, it is important to be able to measure the outcomes of an implemented innovation. One way to improve the evidence-based approach is to measure and evaluate the quality of genetic counselling by using a patient experience measure (Higgs et al., 2022). Patient-reported outcomes (PROs) can be used to measure the effectiveness of healthcare interventions or specific clinical outcomes and help create an evidence base in genetic counselling. PROs are patients’ subjective experiences of health, treatment, quality of life and satisfaction regarding their health condition (Acquadro, 2001). The PROs are operationalised in patient-
reported outcome measures (PROMs) usually by self-administered, short questionnaires. It is possible to measure the underlying dimensions of a particular construct, by using a PROM, containing direct and indirect statements regarding the patients’ subjective experiences. The patients’ responses provide results that are valid and comparable across samples, studies, and participants. This is in line with patient-centred care, as the objective value of care is based on the patient’s preferences and evaluation of outcomes, which can lead to higher-quality and more patient-centred care (Santana, 2018; Tseng & Hicks, 2016).

However, a PROM is only useful if specific aspects of the instrument are assured. These aspects are: it measures the correct outcome or overall goal of the intervention (i.e. valid); it is easy to administer and use; it is easy to understand for the respondent; it is easy to analyse for the provider; and it gives the same results when measured many times (i.e. reliable) (Terwee et al., 2018). Reliability and validity are scrutinised through a psychometric evaluation.

There is a need for a valid and reliable PROM for clinical genetic practice and genetic counselling was lacking in Sweden. Previous research showed that it was difficult to identify appropriate outcome measures for genetic counselling. Traditional validated measures in healthcare were not deemed appropriate to show effectiveness in clinical genetics (Clarke, 1997; Macleod, 2003). Even though quality underpins the ethos of genetic counselling professional practice, evidence-based quality measures for this means were surprisingly rare (Higgs et al., 2022), indicating how difficult it can be to find (or create) the right measures. In our research group, we discussed the possibility of creating a PROM from scratch, including conducting a psychometric evaluation. However, this was deemed too time-consuming for the scope of this PhD. Instead, we chose one of the only existing measures specifically designed to capture the empowerment outcome from genetic counselling: The genetic counselling outcome scale-24 (GCOS-24) (McAllister, Wood, et al., 2011).
Evaluating genetic counselling

The GCOS-24 was selected as an appropriate outcome measure because it was originally developed together with patients and has been evaluated and shown validity and reliability for clinical use in English. The GCOS-24 was created in 2011 and is based on a review of existing validated outcome measures for clinical genetic services (Payne et al., 2008) together with research findings on key patient outcome goals in genetic counselling (McAllister, Wood, et al., 2011).

The GCOS-24 instrument captures the key outcome goal of genetic counselling: patient empowerment (McAllister, Wood, et al., 2011). Patient empowerment uses educational objectives to reinforce or develop general psychosocial skills. It should be based on a mutual patient-provider relationship and should be patient-centred and not disease-specific (Aujoulat et al., 2007). The empowerment measured using GCOS-24 is described as a patient’s belief system that allows them to feel in, or able to take control of, their lives, and have responsibility and autonomy over decisions and choices regarding their genetic condition. This includes patient adaptation and understanding on medical, psychological and familial levels, and includes their ability to make informed choices. The empowerment construct aligns closely with the genetic counselling definition presented earlier (McAllister & Dearing, 2015; McAllister, Dunn, et al., 2011; McAllister et al., 2008; Resta et al., 2006).

Psychometric evaluations of the original GCOS-24 in English show that it is valid, reliable and sensitive to change over time with medium-to-large effect size. It contains 24 items, on a 7-point Likert scale (total score range 24-168). It is suggested that the scale be treated as one-dimensional, specific to the context of clinical genetic services. GCOS-24 captures the following patient aspects: Decisional control; Cognitive control; Emotional control; Behavioral regulation and Hope (McAllister, Wood, et al., 2011). Over the past 10 years, GCOS-24 has become a well-established measure, used in clinical genetics services worldwide. It has been translated, adapted, and occasionally validated in
several other cultural contexts (Denmark, Spain, the Netherlands, Brazil, Singapore, Norway, and Japan (Diness et al., 2017; Munoz-Cabello et al., 2018; Segundo-Ribeiro et al., 2020; Voorwinden et al., 2019; Yuen et al., 2020; Lleuger-Pujol et al., 2022; Løvik et al., 2022; Mochiki et al., 2022). This may allow for future international comparisons between different genetic counselling settings, languages and service delivery models, based on the same measure.

Evaluating telehealth consultations

It is also valuable to capture the patient-related evaluation of using eHealth technologies. The Telehealth Usability Questionnaire combined previously published measures to cover all usability factors of eHealth technologies. It captures patients’ and HCPs’ perceptions of how user-friendly and accessible a certain telehealth technology is in a consultation. Usability is defined as “the extent to which a product can be used by specified users to achieve specified goals with effectiveness, efficiency and satisfaction...” (Parmanto et al., 2016). In our case, it measures the perceived usability of TGC (telephone or video) according to the patients only. Psychometric evaluation of the TUQ has shown good to excellent reliability (Cronbach’s Alpha: 0.81–0.93) and construct validity in previous studies (Parmanto et al., 2016). The measure contains 21 items regarding different aspects of usability including ease of use; usefulness; satisfaction; effectiveness; and reliability. Patients score usability on a Likert scale, by assigning 1–7 points for each item, ranging from Strongly disagree (1) to Strongly agree (7). A higher score corresponds to a higher perceived level of usability, within the range of 21–147 points. The TUQ was translated and cross-culturally adapted to Swedish (Appendix 4) following a modified protocol by Beaton (Beaton et al., 2000), as described under Methods.
RATIONALE

While some advancements have been made recently regarding the genetic counsellor profession and the use of eHealth solutions, access to and quality of genetic counselling are still uncertain. There is still a growing demand for genetic testing and an increased need for genetic counsellors and the provision of genetic counselling, especially when the complexity and availability of genetic tests are expected to increase over time. How to increase access to, and quality of genetic counselling is important to understand better, for example by using eHealth. However, very little is known about this area in the Swedish healthcare context. Research is needed to investigate the utilisation of genetic counsellors, the implementation of TGC, and ways to measure the outcome of these, to better understand how access and quality of genetic counselling in Sweden can be improved, as seen in Figure 2.

Figure 2. A knowledge gap was identified in the intersection of three different scientific areas in which the studies have their theoretical anchoring, i.e., genetic counselling, eHealth and implementation science.
AIMS

The overall aim of this research was to investigate how to improve access to and quality of genetic counselling, through the implementation of telegenetic counselling (TGC) in Sweden. This was done by mapping important determinants for the implementation process, testing the feasibility and providing a quality outcome measure of telegenetic counselling, as well as suggesting an underlying implementation strategy.

Specific aims in the included studies are described below:

Study I: to describe the current practice of genetic counsellors and to explore the role and added value of genetic counsellors in the clinical setting in Sweden

Study II: to identify the factors that influence the implementation and use of TGC in clinical practice, according to HCPs in Sweden

Study III: to investigate the preliminary feasibility of TGC following the rapid implementation process in the pandemic context

Study IV: conduct translation, cross-cultural adaptation, and preliminary validation of a GCOS-24 for measuring outcomes from (tele)genetic counselling in Sweden
METHODS

This chapter describes the different methods used, the study settings, participants, recruitment, data collection and analysis. The chapter ends with ethical considerations.

Research framework

To better understand which aspects of implementation are relevant to the implementation process in this study, a theoretical framework was used to guide the research in reaching its aims in the included studies. The implementation framework of determinants, compiled by Nilsen (2020) was used to conceptualise the determinants of importance regarding the implementation process for TGC (Nilsen, 2020). The different determinants are covered in different studies and the dissertation discussion as illustrated in Figure 3 and explained in more detail in Table 1.
Each determinant is described in more detail in the Background chapter, as shown in Table 1, and Table 2 shows an overview of the research approaches used in each study.
Table 1. Overview of each implementation determinant and where it is studied.

<table>
<thead>
<tr>
<th>Determinant</th>
<th>Who or what</th>
<th>How</th>
<th>Where</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Context</strong></td>
<td>Genetic counsellors working in the Swedish Healthcare system</td>
<td>Investigation of the professional context</td>
<td>Study I</td>
</tr>
<tr>
<td><strong>Adopters</strong></td>
<td>Healthcare professionals providing GC in Sweden</td>
<td>Identifying facilitators and barriers to TGC</td>
<td>Study II</td>
</tr>
<tr>
<td><strong>Outcomes</strong></td>
<td>GCOS-24swe (Patient Reported Outcome Measure)</td>
<td>Providing a quality outcome measure</td>
<td>Study IV</td>
</tr>
<tr>
<td><strong>End-users</strong></td>
<td>Patients referred for GC</td>
<td>Investigating usability and demand</td>
<td>Studies III+ IV</td>
</tr>
<tr>
<td><strong>Implementation strategies</strong></td>
<td>Processes that occurred in each study</td>
<td>Analysis of behaviour change in HCPs</td>
<td>Dissertation discussion</td>
</tr>
<tr>
<td><strong>Implementation object</strong></td>
<td>TGC using the Visiba Care Healthcare Platform</td>
<td>not studied</td>
<td>not studied</td>
</tr>
<tr>
<td>Study</td>
<td>Year</td>
<td>Design</td>
<td>Setting</td>
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<tr>
<td>I</td>
<td>2014</td>
<td>Mixed-methods</td>
<td>National healthcare system</td>
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<td></td>
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<tr>
<td>II</td>
<td>2017-2018</td>
<td>Qualitative</td>
<td>Regional healthcare services - Southeast Sweden</td>
</tr>
<tr>
<td></td>
<td>2019*</td>
<td></td>
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<tr>
<td>III</td>
<td>2020</td>
<td>Mixed-methods</td>
<td>Regional healthcare services - Southeast</td>
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<td>Quantitative</td>
<td>Two regional healthcare services</td>
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<tr>
<td>IV</td>
<td>2020-2021</td>
<td>Qualitative</td>
<td>Regional healthcare services - Southeast</td>
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<td>Quantitative</td>
<td>Two regional healthcare services</td>
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</table>

* The line indicates the start of COVID-19 restrictions

** Patient participants also included in Study IV patient cohort.
Design

The studies included in this dissertation have both qualitative, quantitative, and mixed-methods designs. The choice of design was based on individual study aims and access to relevant participants. This chapter covers relevant aspects of each study’s design: setting, participants, recruitment, data collection and analysis, as seen in Table 2.

Qualitative methods

Qualitative methods are often used when exploring concepts or phenomena with little or no previous research and were applied in Studies I, II and III. The findings were not intended to be generalised to the entire population, but rather give ideal examples of the phenomenon studied (MacFarlane et al., 2014). These methods commonly involve inductive, theory-building efforts, and are rather labour intensive. Typically, a smaller number of participants are included compared to quantitative methods, making it a suitable design when research participants may be scarce or difficult to reach, such as HCPs in Studies I and II, and patient representatives in Study III. The qualitative approaches require awareness of the researchers’ preconceived notions and expectations and emphasises the importance of having an open mind to unexpected findings from the data, which can lead to new areas of research (MacFarlane et al., 2014). This awareness allows for objectivity in the analysis phase and needs revisiting throughout the qualitative process, to reduce the risk of subjective influences and increase trustworthiness in the qualitative findings (Shenton, 2004). The recruitment methods were mainly purposeful, convenience sampling. This sampling technique was deemed appropriate as the targeted participants were somewhat difficult to reach and identify, and because a broad spectrum of participants was optimal, providing a wide range of opinions and experiences (Patton, 2015).
Quantitative methods
Quantitative methods are commonly chosen to confirm a specific hypothesis and to draw conclusions about a population, using statistical analysis, and were applied in Studies I, III and IV. These methods usually require appropriate statistical power and were feasible as a larger sample was available. Large data collection using surveys allows for comparisons between subgroups within a population to be made. Additionally, to ensure the clinical utility of the instrument used to gather such data from patients, a psychometric evaluation was conducted. (MacFarlane et al., 2014).

Mixed-methods
A mixed-methods design was also applied in Studies I and III. This research approach combines qualitative and quantitative strategies in the same study. This approach is becoming more common in scientific research and is beneficial when neither qualitative nor quantitative methods alone can provide a thorough enough understanding of the phenomenon studied. Through triangulation of findings from a combination of approaches, the results can enhance validity and utility to become mutually informative (Bryman, 2012; MacFarlane et al., 2014). Different reasons are used to motivate mixed-methods research designs. For example, the combination of methods allowed us to draw on the strengths and offset the weaknesses of each design, while allowing for improved credibility and utility of findings. It also had to do with the specific sampling and context (Bryman, 2012), for example collecting both open- and closed item responses.

Settings
All four studies took place within the Swedish healthcare system. Sweden has decentralised and universal health, medical and dental care, which is divided into public and private sectors, separately managed by either the municipality or the region. For Swedes, this separation results in varying
availability of healthcare services despite national goals of equal access to care for all inhabitants. Healthcare is mainly financed through regional and municipal taxes, although most medical visits and treatments also have a small fee, up to a certain annual limit. Specialised care, including genetic services, is mainly public and located at the six university hospitals distributed across the country. (Swedish Association of Local Authorities and Regions, 2023; The National Board of Health and Welfare, 2023). However, expansion is to be expected with an increase in both public and private providers of genetic services in the near future.

Studies I and II
Study I was conducted nationwide in Sweden during 2014, while Study II took place in 2017-2018 in the Southeast Healthcare Region.

Studies III and IV
Studies III and IV were conducted during the outbreak of the COVID-19 pandemic. Study III included participants from May 2020 until December 2020, and Study IV included the same participants and those collected until December 2021. They took place at the Departments of Clinical Genetics in both the Southeast Healthcare Region and in the Stockholm and Gotland Healthcare Region. These clinics are responsible for genetic queries and provide genetic services and counselling to approximately 1.1 million respective 2.5 million inhabitants (Statistics Sweden). TGC in both these studies was provided using a specific eHealth innovation; Visiba Care (VC). VC is a healthcare platform provided by the Region Östergötland. The Department of Clinical Genetics was invited to take part in this pilot in 2019 and use the platform for video consultations. VC allows for secure video consultations, capable of sharing screens and multiple users simultaneously, which fulfilled many of the requirements identified as important by HCPs in Study II. Factors such as visibility and the possibility to share visual aids; patient safety; ease of use and access to the technology had been considered important.
Participants, recruitment and data collection

The participants included in all studies were either healthcare professionals, patients or patient representatives and engaged in genetic counselling, in some way or other. Below follows a more comprehensive overview of each study.

Study I

Study I gathered experiences and perceptions of genetic counsellors practising in genetics clinics in Sweden. Because there was no national registry of professional genetic counsellors at the time (still not available today), recruitment was done using the listserve of the SFGV (Swedish Association of Genetic Counsellors). SFGV is a non-profit professional interest organisation that provides a network for individuals interested in the genetic counselling profession. Membership is entirely voluntary and is payable by an annual fee. At the time of recruitment, it was estimated that there were nearly 30 genetic counsellors who practised clinically and eligible to enrol in the study. The original questionnaire was developed in Google Forms and contained both closed and open-ended questions on GCs work situations and opinions on different tasks relating to genetic counselling, and who should perform them (see Appendix 1). The questionnaire was developed by researchers experienced in genetic counselling. Items were mainly based on findings from a previous Delphi study on genetic counsellor tasks (Skirton et al., 2013) and piloted on two academic colleagues. Convenience sampling was applied using the snowballing technique, and the study consent information and a web link to the questionnaire were sent to the entire e-mail list of current (2014) SFGV members, totalling 54 individuals. Two reminders were sent and responses were anonymous. We could not control how many individuals were reached by the invitation.

Study II

Study II took place in 2018, and was an interview study with healthcare professionals practising in the Southeast Healthcare Region. Recruitment
was done using purposeful, strategic sampling, and triangulation of participants was performed to attain a broad variety of participant backgrounds to improve the trustworthiness of results (Shenton, 2004). Potential participants expected to have relevant knowledge of genetic counselling were invited to participate by research collaborators. Interested participants from a variety of backgrounds were then contacted by me with more information about the study plan. The interview was conducted by phone or in person, according to the participants’ preference. To ensure a systematic and comprehensive process, a semi-structured, open-ended interview guide was used (see Appendix 2) and interviews were recorded and transcribed verbatim (Patton, 2015).

Studies III and IV

Studies III and IV in part used the same patient participants for questionnaire responses. Participants were recruited using strategic and convenience sampling from the Department of Clinical Genetics, either at Linköping University Hospital or Karolinska University Hospital, in Stockholm, from May 2020 until December 2021. Invitations were sent, together with the patient’s appointment, via post and included the following: a study information sheet, a decline form and three questionnaires. The questionnaires were the translated GCOS-24swe (before and after appointment) (see Appendix 3) and Telehealth Usability Questionnaire (TUQswe) (see Appendix 4) to capture the patient experience of using the specific service delivery model (video or telephone). The questionnaires were to be filled out at two different time points: before (GCOS-24swe) and after genetic counselling (GCOS-24swe and TUQswe). Additional items regarding preferences before the pandemic were included to capture potential COVID-related effects (see Appendix 4 – at the start of TUQ). Filling out each questionnaire was expected to take approximately 10-20 minutes. Reminders were sent out at 2 and 4 weeks post appointment, only to those who had consented to participate. Appointments were exclusively via video or telephone, due to COVID-19 restrictions. Inclusion was broad, inviting all patients with a planned appointment for genetic counselling to participate, with some
exceptions. For example, the need for an interpreter and very complex cases (such as syndromology consults) were excluded. If a participant did not fulfil the technical requirements for a video appointment (as listed in their invitation letter) or did not return a valid email address (to receive the web link for a meeting in Visiba Care) they were automatically switched to a telephone appointment instead, as telephone numbers are readily available using the Swedish national telephone directory. Consent was given by returning a filled-out questionnaire.

Study III included the first 49 consenting respondents, recruited from May 2020 until December 2021. Meanwhile, Study IV included consenting respondents from the entire recruitment period (n=374). Studies III and IV applied descriptive statistics using R-studio to show the demographic data of included participants. All statistical analyses in Studies III and IV were conducted with the software R in the interface software RStudio (RCor Team, 2022; Schreiber, 2006) and RStudio (RStudio Team, 2020).

Moreover, Study III was initiated at the beginning of the pandemic and was obviously affected by changed circumstances around the world. Therefore, it was agreed that the TGC feasibility trial was to be initiated rapidly. A group of six clinical staff were selected from the Department of Clinical Genetics in Linköping. Recruitment was strategically carried out to recruit a variety of professionals including administrative staff (n=2), medical doctors (n=3) and genetic counsellors (n=1), comprising a variation in age and experience. First, they trialled Visiba Care for the provision of TGC and filled out short evaluations directly after each session they conducted (in-house questionnaire with 5 items to capture experiences – see Appendix 5). Additionally, they participated in regular, recurring discussion meetings. Participant involvement was central, with open discussions regarding design, planning, execution and problem-solving based on participants’ clinical experiences of TGC. This was to inform the next step of implementation: introducing TGC broadly to all staff in the clinic. The process was similar to co-design as described by Vargas and colleagues and included compiling researchers’ notes from
the group discussion and survey data from Visiba Care (Vargas et al., 2022).

In Study IV, additional participants were recruited using strategic sampling to form an expert committee. The committee included five researchers with relevant expertise in the fields of genetic counselling and qualitative research (CG, RP, MM), eHealth and nursing (PJ) and implementation science (PN). The expert committee consulted and performed the initial GCOS-24 questionnaire adaptation and supported the qualified translators with translation, into the Swedish language and context. Adjustments were proposed and agreement was reached on changes necessary in regular group discussions during the whole questionnaire adaptation process (Details of the process shown in Figure 5).

Additionally, in Study IV, six patient representatives were recruited, using convenience sampling with the help of a patient support organisation collaborating with the Centre for Rare Diseases in the Southeast Healthcare Region (Centre for Rare Diseases Southeast). Recruitment aimed at finding participants with a broad range of experiences, to participate in in-person, semi-structured cognitive interviews using an interview guide and applying the probing technique (Beatty, 2007; G.B. Willis, 2006). The purpose was to capture participants’ understanding and cognitive processing of each item in the translated version of GCOS-24. The data collected comprised written report compiled for each item of the GCOS-24swe questionnaire from each interview.

**Qualitative analysis**

This chapter describes the qualitative analysis that took place in the different studies.
Study I

Thematic qualitative analysis was used to answer a specific question (Braun & Clark, 2006), in this case, “What do the genetic counsellors contribute in the clinical investigations and encounters in Sweden?” The involved researchers (RP, CI, HS) all had relevant expertise and competencies, working in, and teaching genetic counselling. Also, CI and HS had substantial experience of qualitative research and design in the field. This expertise, together with findings from the literature, was used to design the questionnaire. The analysis meant that the researchers processed the collected data in the following way: Familiarising themselves with the data; Coding data and selecting codes relevant to the research question; Grouping of codes; Finding patterns of meaning; Organising key themes; Reviewing findings with all involved researchers. (Braun & Clark, 2006; Polit & Tatano Beck, 2011). Awareness of researchers’ preconceived notions was important to allow for more objective data analysis. This was done by revisiting the different codes, patterns and themes identified, in the light of new findings in the material – to assure consistency and objectivity of findings. The review of findings together with all involved researchers (triangulation) was also applied important to increase trustworthiness.

Study II

Phenomenographic analysis is inherently exploratory and appropriate to generating new theories in a previously unexplored area. Phenomenography uses a second-order perspective to describe an individual’s perspectives of a specific phenomenon, in this case, the HCPs’ prospective use of telegenetic counselling (Marton, 1981). To assure trustworthiness, the semi-structured interview guide was developed and piloted with two colleagues. The guide allowed the interview process to become more comprehensive and systematic (Patton, 2015). The following steps were included in the analysis process, based on the transcribed interview data: Familiarisation and compilation; Identifying core components; Grouping into sub-categories; Comparing
categories; Common understanding between researchers; Contrastive comparison in an overarching, hierarchical outcome space (Marton, 1981; Sjöström & Dahlgren, 2002). To improve trustworthiness, we aimed to include a broad range of participants, with different frames of reference, from different backgrounds and locations. This allowed for a variation of experiences to be illuminated. Furthermore, triangulation of findings was also applied in this analysis process, allowing all involved researchers (RP, CG, PN, PJ) to revisit findings from previous steps in the process, raise questions regarding these and reach an agreement on the final findings in the analysis, in an iterative way.

**Study III**

*Phase 1:* The first phase of Study III used co-design and engaged the six HCP participants in the development and planning of rapid implementation of TGC in the clinical setting, which directly would impact the work of the participants involved. Using consensus among the HCPs, they were engaged to create plans for the use and implementation of TGC and to suggest and try solutions to problems that arose when trialling TGC. The process was re-iterative and repetitive and allowed HCPs to suggest solutions throughout the implementation process of TGC. Agreed-upon changes were tested by the HCPs themselves and then discussed in the next group meeting. Each discussion could result in new suggested changes, again incorporated and tested, repeatedly in a cyclical fashion, as shown in Figure 4. The process was repeated until no more changes were considered necessary by the HCPs, and TGC was deemed ready for broader implementation to all the staff in the clinic. Analysis occurred simultaneously and repeatedly throughout the process, through HCP discussions of experiences and reaching agreement. The initial design also incorporated findings from Study II, i.e. required provision of technical equipment and support; learning and testing opportunities; and instructions and guidelines. HCP discussions were initially held every two weeks but were later changed to monthly meetings once all personnel were introduced by mandatory training. The process model used in the first phase of Study III, involving stakeholders.
in the design and plan of implementation in healthcare, corresponds to the co-creation in the healthcare model, called co-design. It is defined by Vargas (2022) as “engaging diverse stakeholders in the process of understanding complex problems, and designing and evaluating contextually relevant solutions” (Vargas et al., 2022). The Kalamazoo Consensus (Participants in the Bayer-Fetzer Conference on Physician-Patient Communication in Medical Education, 2001) proposes six collaborative steps to facilitate co-productive relationships that are relevant for co-creating health (Realpe & Wallace, 2010). The HCP co-design process in this research facilitates some of the essential components of professional collaboration identified by the Kalamazoo Consensus. Using a co-design model for the development and planning of the implementation of healthcare innovations was deemed appropriate as it considers the users (HCPs) to be experts on their circumstances, and adds value to the outcomes of how to use TGC, potentially also improving implementation (Vargas et al., 2022).

Figure 4. Illustration of the iterative process of co-design with HCP participants in design and planning activities for implementing TGC in the clinical setting.
Phase 2: The second phase of Study III concerned the broad implementation of TGC and included all the staff in the clinic. This was a mixed-methods approach to collect data for triangulation, based on different data sources that included collected qualitative and quantitative data (MacFarlane et al., 2014). The data sources used were: the researcher’s notes and results from the HCP co-design process (from phase 1), specific organisational information, and survey data collected from both patients and HCPs. The following surveys were included: TUQswe, GCOS-24swe, in-house questionnaire for HCPs (Appendix 3-5) and Visiba Care post-visit question (Asking: “Where you satisfied with the digital consultation?”). The feasibility of implementing and using TGC in the clinical setting was analysed, using the compiled results from each data set. This approach used both qualitative and quantitative data, which allowed for a mixed-methods approach to analyse feasibility from the following aspects: acceptability, demand, efficacy and implementation of TGC. This allowed for analysis using triangulation, showing the data from the various sources that were consistent and overlapping and mutually verified (Bryman, 2012; MacFarlane et al., 2014).

Study IV
Translation and cross-cultural adaptation of TUQ and GCOS-24 were carried out (prior to Study III in time) to create semantic, idiomatic, experiential and conceptual equivalence in the Swedish language and context, see Appendices 3 and 4 for final versions in Swedish). The process was adapted from Beaton (2000) and involved the above equivalence aspects, which were defined as follows: Semantic – the meanings of the words are transferred across translation. Idiomatic – changing idioms for equivalent expressions in Swedish. Experiential – statements seeking to capture experiences of daily life in Sweden. Conceptual – focus on the conceptual differences between meanings of words in different cultures (Beaton et al., 2000). The process involved
professional translators and the expert committee that met regularly to discuss the translation and adaptation of the GCOS-24. The process included the steps shown in Figure 5 (Beaton et al., 2000; Beatty, 2007).
**Figure 5. Cross-cultural adaptation process.**

- **Forward translation:** Translated 1 (T1)
- **Synthesis:** The report committee agreed on a combined translated version (T1-2) based on conceptual and cultural equivalence.
- **Back-translation:** A native speaker translated the T2 back to English to highlight any linguistic imperfections.
- **Consolidation:** The report committee, including the originator, Metin Makitalo, resolved any discrepancies and agreed on a consolidated translated version.
- **Cognitive interviewing:** Participants from the target population (at risk of or affected by a genetic condition) were interviewed to test the readability and understandability, using a semi-structured cognitive interview technique. Probing was employed to elicit further explanations of anything unclear to the interviewer.
- **Agreement:** The report committee agreed on necessary modifications based on the written reports from all previous steps in the process. The final version was ready for psychometric validation.

By independent, professional translators, without prior knowledge of the questionnaire.

Forward translation; Synthesis into Swedish; Back-translation to English; Consolidation in Swedish; Cognitive interviewing; Agreement.
An interview guide and the probing technique (Appendix 6) were used for the cognitive interviews in step 5 (Figure 5). This was an important step to assure face validity and cognitive understanding of items in the target population, i.e., patients who receive genetic counselling. Interview data was documented using researchers’ notes from the interviews with each respondent on each item (Beatty, 2007; Willis, 2006). All compiled data from translations and cognitive interviews were discussed in the expert committee, regarding semantic, idiomatic, experiential and conceptual equivalence, aiming to stay true to the original, and any adjustments were only made following group consensus. Thus, face validity was established by the expert committee using data from the cognitive interviews with patient participants (Bolarinwa, 2015).

**Quantitative analysis**

This chapter describes the quantitative analysis that took place in the different studies.

**Study I**

SPSS Version 22.0 (IBM Corp., 2013) was used for descriptive statistics based on the fixed-choice item responses, and Chi-2 comparisons to identify statistically significant differences between groups, using Bonferroni adjustments to correct for potential bias.

**Study III**

In Study III, preliminary efficacy evaluation was performed. This analysis involved using Welch t-tests (allowing for unequal variances) to measure differences in scores from GCOS-24swe before and after. As this was a pilot study, statistical power was not determined, because of the smaller sample size.

**Study IV**

In Study IV, psychometric evaluation of the GCOS-24swe was performed, analysing reliability, validity and responsiveness. Reliability,
also referred to as internal consistency, indicates whether the GCOS-24swe questionnaire had good internal consistency and was measured using Cronbach’s alpha (Bolarinwa, 2015). Responsiveness is used to see whether the questionnaire can identify changes over time. This was important as GCOS-24swe is meant to capture changes in patients’ empowerment before compared to after genetic counselling, and was analysed using effect size of the mean difference, or Cohen’s d according to the following cut-offs: 0.2 = small; 0.5 = medium and 0.8 = large effect (Cohen, 1992). The validity of the theoretical construct for GCOS-24swe was analysed using confirmatory factor analysis (CFA). It compared to previously published GCOS-24 versions that provided psychometric results in English, Dutch and Chinese (McAllister, Wood, et al., 2011; Voorwinden et al., 2019; Yuen et al., 2020). The evaluation was based on a selection of fit indices representing many different properties of a model. (RCore Team, 2022; Schreiber, 2006; RStudio Team, 2020).

The sample size was important to calculate, as to achieve statistical power relevant to detect an important effect in the model. The value of 0.8 was considered sufficient power in Study IV. The sample size was calculated to be a minimum of 240 participants, matched to 10 times the number of items in the GCOS-24swe (Muthén & Muthén, 2002).

**Missing data**

Missing data occurred in the gathered questionnaire data in Studies I, III and IV. In Study I, missing item responses ranged between 0 to 31% and were not included in the analysis of the specific response. In Studies III and IV, missing data were imputed with previous answers for missing data less than 5%, and were treated as missing at random, to create a complete data set. Questionnaires with more than a 50% dropout rate were excluded entirely (van Buuren, 2011).
ETHICAL CONSIDERATIONS

In research, there are always ethical considerations to make, and therefore international agreements are in place to protect the research subjects (Declaration of Helsinki. Ethical Principles for Medical Research Involving Human Subjects, 1964). Throughout these research projects, careful consideration and compliance with these ethical guidelines have been pursued. Study I was considered exempt from needing ethical approval after querying the Swedish Ethical Review Authority. Ethical approval from the same was obtained for the last three studies, Dnr 2019-01051 and Dnr 2020-05243. Appropriate participant information has been provided, including rights of withdrawal at any given time, without reason or any adverse effects on the care provided. In regards to genetic counselling, it is always necessary to observe ethical considerations, as it is a process that inherently raises questions of right and wrong, moral and justice, both on personal and family levels for patients, healthcare professionals, and society as a whole. The ethical considerations of research and genetic counselling align closely – and it is a natural part of the ethos to practice flexibility and patient/participant-centredness throughout both processes, be it in consultation with a patient or in an interview with a research participant. Particular focus is also placed on adapting information to the individual’s needs, emphasising voluntariness and anonymity, and obtaining informed consent throughout the entire processes.
FINDINGS FROM THE STUDIES

This chapter presents the main finding from each study as shown in Table 3. A compilation and synthesis of these results can be found in the Discussion chapter.

Table 3. Overview of the main finding from each study.

<table>
<thead>
<tr>
<th>Study</th>
<th>Main findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Genetic counsellors add quality and accessibility for patients</td>
</tr>
<tr>
<td>II</td>
<td>TGC increases patient access and autonomy when specific requirements are fulfilled</td>
</tr>
<tr>
<td>III</td>
<td>TGC is feasible regarding acceptability, demand, efficacy and implementation, and increases access to genetic counselling</td>
</tr>
<tr>
<td>IV</td>
<td>GCOS-24swe is a valid and reliable outcome measure that can assess the effectiveness and overall quality of genetic counselling and evaluate the implementation process</td>
</tr>
</tbody>
</table>

Study I

The first study aimed to explore the use of the GC profession in the Swedish context. It found that in 2014, genetic counsellors were infrequently used, the role was mostly undefined, and there was no common educational pathway. However, the findings also showed that genetic counsellor professionals practising in clinical settings in Sweden added quality and improved access for patients. Furthermore, results showed that most genetic counsellors were responsible for similar tasks before, during and after clinic appointments. Examples of these common tasks were collecting relevant information from the patient and drawing a family tree, participating in consultations, and providing follow-up contacts with patients. More than half of the respondents performed
additional tasks, such as research, teaching and professional development. Results also highlighted the heterogeneous backgrounds of the genetic counsellors, and the variation in roles, responsibilities and tasks that they had. Nevertheless, many genetic counsellors were overly burdened by administrative tasks and instead desired more direct patient time.

Four main themes were identified from the qualitative data:

1) The genetic counsellor acts as the “spider in the web” preparing each case
2) The genetic counsellor's capacity allowed for a holistic, ethical and psychological view of the patient and family
3) The genetic counsellor could build relationships and provide continuous support to the patients, which facilitates an increase in patient-centredness
4) The genetic counsellor was more accessible compared to medical geneticists

**Study II**

The second study aimed to explore important determinants for the implementation of TGC. It found that healthcare professionals (with relevant experience in genetic counselling) believed that TGC could lead to patient benefits, mainly through increased autonomy and access to genetic counselling. Improved access could be achieved regardless of long travel distances and other time constraints, and by simultaneously including several family members, even in different locations. However, the implementation success of TGC depended on specific requirements, which included evidence of effectiveness, i.e. an appropriate outcome measure, and specific resources, as shown in Figure 6. The resources concerned both staff and technical aspects (Level 1). The participants also showed concerns regarding anticipated changes to their work environment and patient interactions. They also worried that TGC might disadvantage individuals without adequate technical knowledge or support (Level 2) But, despite some hesitancy among healthcare
professionals, the study identified that Sweden, as a country, also had several favourable conditions for the implementation of TGC, such as very high technological access and acceptance among inhabitants already, and that ultimately there were several patient benefits (Level 3). The main findings are ordered hierarchically in the outcome space, as shown in Figure 6.

Figure 6. The main findings from the phenomenographic analysis as shown in the outcome space. (Printed with permission, R.Pestoff et al., 2019)

**Study III**

The third study aimed to explore the feasibility of implementing TGC during the pandemic and started right at the beginning of COVID-19. The context changed dramatically, and it was decided that Study III would serve best as a feasibility study of rapid implementation of TGC, likely to benefit both patients and staff. This allowed genetic counselling consultations to continue, despite physical appointments being
prohibited. The co-design process uncovered important requirements from HCP participants for the implementation of TGC in the clinical setting. They are shown as Examples in Figure 4 and are discussed in more detail below.

HCP participants required a specifically adapted checklist for conducting genetic counselling using TGC. This checklist to facilitate the use of TGC is provided in Figure 7. Additionally, HCPs requested the development of guidelines and clinical routines preceding the broad clinical implementation of TGC. Other important aspects were to allow voluntary participation among staff, and that all staff undergo a mandatory introduction and training sessions. This included a lecture on the background, current (research) findings, other participating clinics, and a hands-on tutorial to try the system. These educational sessions, on-demand support and regular Q&A sessions were made available to all staff to facilitate their use of TGC. Resistance to using TGC among staff was low, whilst acceptance and demand were high. However, the context had suddenly become more favourable due to the pandemic, as there were not many other options at the time. Thus TGC allowed staff to carry on their day-to-day work and continue to provide genetic counselling to patients. The study showed preliminary efficacy, shown by a significant improvement in empowerment as measured by GCOS-24swe, comparing before TGC to after TGC (mean improvement +13.9, \( p>0.001 \), 95\% SD=14.9), as shown in Figure 8. As can be seen, there was slightly more improvement in GCOS-24swe scores for patients receiving TGC via video, when stratified for video and telephone TGC, as shown in Figure 9, however, this was non-significant (n=49 (\( p=0.224 \))). The TUQ surveys showed a mean rating of 5.4 for usability (mean 113, max 140, SD=19.6), where an item mean score over 5 is considered a good rating of usability. This was the rating for both telephone (M=107, SD=19.1) and video (M=117, SD=19.2) genetic counselling. Also here, there was a non-significantly higher rating for video than for telephone appointments (n=49 (\( p=0.081 \))). The study concluded that TGC was feasible and successfully implemented in our clinic.
Figure 7. The checklist provided to guide HCPs in using TGC in a clinical setting

Checklista för genetisk vägledning via video:

1. Identifiera dig och patienten (ta personnummer på närvarande patienter)
2. Förfara premisserna: Video-besök i första hand – om det ej funkar:
   a. Telefonbesök Vilket telefonnummer till patienten?
   b. Mottagningsbesök.
3. Gällingen inkommande remiss och frågeställning.
4. (Fråga om skickat in enkät för utvärdering när lämpligt)
5. Förfara syfte och upplägget för besöket (välj lämpiga punkter):
   a. Ta medicinsk anamnes.
   b. Genetisk bakgrund (visa bilder från powerpoint för genetisk vägledning)
   c. Förfara uppkomst av mutationer och dess betydelse för sjukdomar
   d. Ta släktanamnes eller visa befintligt släktträd (genom att dela skärml)
   e. Förfara relevant nedärvningsgång och relatera till patientens fall (visa bilder)
   f. Förfara bakgrunden till den kliniska misstanken
   g. Förfara genetisk testningsförfarandet (tex blodprov, remiss, väntetid, hur och när ny kontakt)
   h. Diskutera eventuella farhågor/fördelar/nackdelar ur patientens perspektiv
   i. Penetra psykologisk anamnes och social situation (när tillämpligt)
6. Stäm av frågor och funderingar hos patienten/anhöriga/gäster?
7. Gör en tydlig planering för den fortsatta utredningen, inklusive ny kontakt (video/telefon/besök)
8. Be patienten summera informationen från besöket i 2-3 korta punkter, alternativt sommar själv besöket
9. Avsluta besöket
10. Meddela administratör på respektive enhet att besöket genomförts (för att faktura ska skickas och anteckning läggas på rätt vårdsnivå i Cosmic)
11. Ankomstregistrera patienten efter besöket: Välj Besökslista i Cosmic; därefter ändra till Anlänt.
12. Diktera på Besök över internet
Figure 8. The difference in GCOS-24swe scores before TGC (M = 106), as compared to after TGC (M = 120). There is a significant improvement of +13.9 (p<0.001) n=49 (Reproduced with permission, R. Pestoff et al., 2020)

Figure 9. The GCOS-24swe improvement scores before TGC as compared to after TGC, stratified on the mode of delivery: Telephone (M= 10) vs Video (M=15) (p=0.224) n=49 (Reproduced with permission, R.Pestoff et al., 2019)
Study IV

The final study aimed to evaluate the adaptation and validation of GCOS-24swe for use as a clinical outcome measure. It included translation and cross-cultural adaptation of the GCOS-24 instrument, which were considered rather unproblematic by the expert committee. This indicated a sufficient translation and adaption process, whilst retaining the equivalences to GCOS-24. Retaining equivalence was prioritised by the expert committee, to provide comparable outcomes to other studies. The process did, however, lead to some minor changes in the wording of different items, to achieve conceptual equivalence. Both the expert committee and cognitive interviews indicated that the word “condition” (included in items #2, 3, 4, 6, 7, 9, 11, 12, 13, 16, 17, 18, 21, 22, and 24), that was initially translated into sjukdom or diagnos, should rather be translated into tillstånd, to achieve better semantic equivalence. Similarly, it was indicated that negatively worded items ought to be reversed, as they were difficult to understand. They were therefore reworded into a positive mode. Essentially, this means that understanding improved and that the scoring of these items (items # 10, 12, 17, 18) no longer needs to be reversed.

Due to the rigorous process of translation and adaptation, the results from Study IV suggest that GCOS-24swe showed face validity, whilst maintaining semantic, idiomatic and conceptual equivalence. The statistical analysis showed good reliability (Cronbach’s alpha 0.86), and responsiveness, with Cohens d= 0.65 showing medium effect size ($p<0.001$) and a mean empowerment improvement of 12.87 (95% CI[10.74, 14.99]) on the group level. Results from CFA revealed that data derived from GCOS-24swe had a good enough fit to a one-factor model, which indicates construct validity with factor loadings on one dimension. Therefore, the GCOS-24swe offers the first validated outcome measure appropriate for use in clinical genetics in Sweden. The final version of GCOS-24swe is shown in Figure 10.
**Frågor om genetisk vägledning och ärlig diagnos**


<table>
<thead>
<tr>
<th>Svar</th>
<th>Instämmer inte alls</th>
<th>Instämmer något</th>
<th>Instämmer lite</th>
<th>Varken instämmer eller inte</th>
<th>Instämmer lite</th>
<th>Instämmer något</th>
<th>Instämmer inte alls</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Jag förstår varför jag besöker den genetiska mettagningen</td>
<td></td>
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<tr>
<td>2</td>
<td>Jag kan förklara vad det medicinska tillståndet innebär för de femte medlemmarna som kan behöva veta</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>Jag är medveten om vilken påverkan det medicinska tillståndet kan ha på mitt/mina (eventuellt framtida) barn</td>
<td></td>
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<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Jag menar att jag är en bra person på det medicinska tillståndet min familj</td>
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<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Jag vet att jag ska hämta mig för att få genomföra detta som jag och/eller mitt familj behöver (t.ex. försörjningslagar, mediciners, behandling, kontroller)</td>
<td></td>
<td></td>
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<td></td>
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</tr>
<tr>
<td>6</td>
<td>Jag menar att det medvetandes del av min familj har kunnat se något positivt</td>
<td></td>
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<td></td>
<td></td>
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<tr>
<td>7</td>
<td>Jag upplever att jag har kontrollerat hur det medicinska tillståndet påverkar min familj</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>Jag känner mig positiv inför framtid</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>Jag känner att jag kan hantera att ha det medicinska tillståndet i min familj</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>Jag vet att jag har för nytta av de alternativ som finns tillgängliga för mig (t.ex. genetisk testning, delta i kontrollprogram, fosterdiagnostik)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>Att ha det medicinska tillståndet i min familj gör mig onödig</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>Jag vet att det medicinska tillståndet kan påverka mina egen och min familjs näringstruktur på något sätt (t.ex. psykiskt, fysiskt/funktionell, foster/montering, kruskiga)</td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>
Figure 10. The final version of GCOS-24swe was used in Studies III and IV. (Reproduced with permission from publisher Wiley &Sons)
DISCUSSION

The dissertation found that is possible to increase access to and measure the quality of genetic counselling in Sweden, despite the lack of professionals to provide genetic counselling. The first study finds that genetic counsellors improve access to, increase the quality of, and provide added value to patients in the clinical setting, but that the GCs are underutilised. Subsequent findings show that TGC is a feasible way of expanding access to genetic counselling and is acceptable and effective in the right circumstances. Furthermore, this research provides a valid and reliable PROM for measuring quality and effectiveness regarding genetic counselling, both in-person and when using TGC. All in all, the results provide a knowledge base for improving access to genetic counselling by utilising genetic counsellors and TGC more extensively, and they also provide a tool for evaluating outcomes of genetic counselling.

Compiling a dissertation leads you to consider what knowledge has been contributed to the field. Three general parts are identified below and discussed individually, followed by a synthesis of findings. It is important to point out that Studies I and II took place before the pandemic, while Studies III and IV were carried out during it. The onset of COVID-19 brought both challenges and opportunities from a research perspective, meaning that the contexts were impacted differently by COVID-19.

Genetic counselling today and in the future

Developments and issues regarding the genetic counselling profession in Sweden were identified in 2014, nearly a decade ago, when the first study took place. The findings showed that genetic counsellors improved access, added value and quality to patient and healthcare professionals’ encounters, and also provided a more holistic perspective. The holistic perspective involved the professional attending to the needs of the whole individual and their family, including psychological and ethical support. This indicated a maturing professional identity similar to the one
described in North America (Baty, 2018). The findings showed that despite their diverse backgrounds, mostly as nurses or biologists, genetic counsellors were highly skilled learning by doing in their respective clinics. Naturally, this resulted in heterogeneous knowledge and responsibilities, which indicate a need to harmonise genetic counsellor education and create a united knowledge base. It also indicates that genetic counselling services could improve with better utilisation of genetic counsellors’ skills. Since then, the need for genetic counselling has increased further and the provider-demand gap has widened considerably (Abacan et al., 2019; Stoll et al., 2018). In terms of addressing the identified issues, 2021 marked the start of the first MSc GC programme in Sweden. Now, the education and role of genetic counsellors can transition from a non-regulated, in-house, individual training into a mutual, quality-assured educational pathway. This can result in common core competencies for genetic counsellor professionals harmonised at the Swedish and European levels, via the EBMG, assuring quality and patient safety.

Worldwide, multiple different initiatives have addressed the issue of access to genetic counselling, while maintaining quality. For a comprehensive summary see Stoll et al., 2018 (Stoll et al., 2018). Examples include employing assistant or extended GCs to unburden GCs of many administrative tasks (Pirzadeh-Miller et al., 2017); use of technical solutions on the web to answer simpler patient queries, such as chatbots or asynchronous messaging (Biesecker, 2018; Koerner et al., 2023; Siglen et al., 2022); genetic counselling group sessions for patients concerned about being at risk of the same genetic condition (Otten et al., 2015); mainstreaming genetic testing to other clinics (Baty, 2018; Patch & Middleton, 2019; Quinn & Mazur, 2022); providing only post-test genetic counselling for identified mutation carriers in specialised healthcare (Fonda Allen et al., 2016); integrating GCs into primary care (Slomp et al., 2022); use of different supporting decision aids (Adam et al., 2018; Freed et al., 2021; Hilgart et al., 2012); as well as interest in
providing telegenetic counselling (even before the pandemic) (Elliott et al., 2012; Gardner et al., 2015; Zilliacus et al., 2009).

**Prepared for telegenetic counselling**

TGC is generally considered well-suited for genetic counselling consultations due to its conversational nature, and the initial studies showed promising results, i.e. regarding knowledge retention, satisfaction, acceptability, convenience and ability to build trust and rapport between patient and provider (Gardner et al., 2015; Orlando et al., 2019; Otten, Birnie, Ranchor, et al., 2016; Stoll et al., 2018; Zilliacus et al., 2010). However, many barriers are also identified, for example, the inability to provide appropriate psychosocial care; reduced relational aspects; technical problems; and less compliance with follow-up (Danylchuk et al., 2021; Gorrie et al., 2021; Green et al., 2022; Vrecar et al., 2017; Bradbury et al., 2011; Terry et al., 2019). It is clear that TGC has been readily available for quite some time, but infrequently implemented in clinical settings before COVID-19 (James et al., 2021; Otten, Birnie, Lucassen, et al., 2016). The question was why uptake of TGC had been low, despite the availability of the technology and results showing positive findings in several settings, and what could be done about it.

**Implementation factors**

Studies II and III explored the barriers to and facilitators of the implementation, and use of TGC in Sweden. Implementation of innovations in healthcare is known to be challenging and involves complex changes that disrupt long-established routines and processes (Greenhalgh et al., 2020; Grol & Wensing, 2005). This can explain some of the hesitancy that HCPs showed regarding the use of TGC, before COVID. The two studies identified several important factors: The Swedish context demonstrated a high technological acceptance which, paired with improved utilisation of genetic counsellors (findings from Studies I and II), could increase patient access to genetic counselling.
However, the adopters (HCPs) required additional resources, such as support, evidence and education to use TGC. Both end-users (patients) and adopters found that TGC was acceptable and in demand, similar to findings reported by Greenhalgh and colleagues. (Greenhalgh et al., 2020).

Despite the identified barriers and hesitancy by HCPs, implementation of TGC was deemed successful in the Swedish clinical context in Region Östergötland. Since the pandemic, similar findings have been reported from other contexts, for example, in genetic cancer, prenatal, metabolic, and psychiatric settings (Costanzo et al., 2022; Douglas et al., 2023; Salman et al., 2022; Shannon et al., 2021; Shur et al., 2021), and in different locations, for example in the USA, Canada, the Philippines, South Africa, India and Italy to mention just a few (Choi et al., 2022; Norman et al., 2022; Pagliazzi et al., 2020; Rao et al., 2021; Shannon et al., 2021; Tumulak et al., 2021; Wessels et al., 2021). Also taking the end-user perspective, Dantas and colleagues recently showed that TGC improved access, especially during the pandemic, and that it also reduced cost and time spent, according to patients in Portugal (Dantas et al., 2023).

The importance of an outcome measure

Outcome measures to improve quality in the field of genetic counselling, and to evaluate research on the implementation process, are necessary to evaluate quality and implementation in healthcare, as previously identified (Higgs et al., 2022; Danylchuk et al., 2021). Likewise, our HCPs required an appropriate outcome measure to create evidence and measure the effects of TGC. However, Higgs and colleagues found that appropriate outcome measures were still often lacking for genetic counselling in many regards (Higgs et al., 2022). The relevant and useful validated GCOS-24swe could therefore serve as a suitable outcome measure for this purpose. GCOS-24swe could also be useful for future clinical evaluations and comparisons, both between countries, and also between different service delivery models, i.e. in-person genetic
counselling compared to TGC. Furthermore, GCOS-24swe could be used for tailoring genetic counselling sessions to the patient’s individual needs and for measuring the baseline empowerment levels before a consultation, which has also been indicated by other studies (Mochiki et al., 2022; Salman et al., 2022). This could help tailor each session individually, improve patient-centredness and the perceived quality of genetic counselling even further.

The missing implementation factors

Readers will hopefully have noticed that two implementation determinants from the theoretical implementation framework by Nilsen (2015) have not been given adequate consideration in this research. The implementation object, TGC, uses the Visiba Care platform, which was a pre-determined factor provided by Region Östergötland, and beyond the scope of this research. However, the final determinant: the implementation strategy, is of great importance for providing a complete understanding of the implementation process that took place. Nevertheless, the strategy was not specifically considered in either of the individual studies and therefore remained “the unknown factor” throughout the project. Moreover, work on this dissertation led to reflections on its contributions to the field of implementation research. Key findings on implementation activities are synthesised, and identified as a potential implementation strategy to be used in future research and guide future implementation of similar innovations in similar healthcare contexts. This synthesis is entirely retrospective and has not been presented elsewhere.

A synthesis of findings in this dissertation

As described in implementation research, new guidelines and recommendations in healthcare are infrequently followed or fully integrated by healthcare professionals (Greenhalgh et al., 2017). Implementing a new healthcare innovation, such as TGC, can therefore be challenging for HCPs (Greenhalgh et al., 2020) since it requires
behaviour change both among adopters (i.e. healthcare professionals) and end-users (i.e. patients).

**Using a model for behaviour change**

A well-known model for behaviour change, the Behaviour Change Wheel (BCW), is based on a systematic review of 19 existing frameworks for behaviour change interventions (Michie et al., 2011). The model contains three different analysis steps: 1) Behaviour analysis 2) Identifying implementation activities 3) Identifying policy (facilitators or barriers), as shown in Figure 11, to guide necessary steps for successful implementation.

![Figure 11. The Behaviour change wheel: A method of characterising and designing behaviour change interventions. CCBY Copyright 2011, Michie et al.: license BioMed Central Ltd.](image)

The first step (in the centre of the BCW) is to identify capabilities, opportunities and motivations that can lead to changed HCP behaviours.
(COM-B). The second step is to identify effective implementation activities, and the third step is to identify types of policy that support the specific implementation. This deductive analysis resulted in a preliminary, suggested Implementation strategy.

1. The first analysis step (Sources of behaviour in Figure II) identified several HCP behaviours that needed to change, to allow for the right capability, opportunity and motivation to achieve the desired behaviour (i.e. to use TGC - based mostly on findings from Study II):

   **Capability** (physical and psychological)
   Healthcare professionals may not have the knowledge or skills to use TGC.

   **Opportunity** (social)
   Staff do not know about other HCPs’ use of the video system, i.e. they needed role models.

   **Motivation** (reflective and automatic)
   There was no current evidence in the literature.
   There was a need to establish routines and habits.
2. The second analysis step (Intervention functions in Figure 11) identified implementation activities which were used to change the HCPs’ behaviour. These findings are specific implementation activities in Study III, as shown in Table 4.

<table>
<thead>
<tr>
<th>Activity</th>
<th>Definition (from COM-B)</th>
<th>Activities that took place during TGC implementation</th>
<th>Addresses the following component (from COM-B)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Education</td>
<td>Increasing knowledge and understanding</td>
<td>Using co-design with HCPs to plan and design implementation in the clinical setting Providing mandatory introduction and voluntary “drop-in” sessions offered regularly with spokesperson</td>
<td>Psychological capability Reflective motivation</td>
</tr>
<tr>
<td>Persuasion</td>
<td>Using communications to stimulate actions</td>
<td>Providing regular feedback on use, and good examples from the Region and clinics (using each other as examples)</td>
<td>Automatic motivation Reflective motivation</td>
</tr>
<tr>
<td>Training</td>
<td>Imparting skills</td>
<td>Providing opportunities for all HCP to practice in a “mock” environment with a spokesperson</td>
<td>Physical capability Psychological capability Physical opportunity Automatic motivation</td>
</tr>
<tr>
<td>Restriction</td>
<td>Using rules to increase the target behaviour</td>
<td>COVID-19-related restrictions prohibiting all in-person consultations Providing instructions for use of TGC</td>
<td>Physical opportunity Social opportunity</td>
</tr>
<tr>
<td>Environmental restructuring</td>
<td>Changing the physical and social context</td>
<td>Providing space, laptops, microphones, cameras and access to Visiba Care technology</td>
<td>Physical opportunity Social opportunity Automatic motivation</td>
</tr>
<tr>
<td>Modelling</td>
<td>Providing an example for people to imitate</td>
<td>Through co-design. Spokespersons show use and benefits. Giving examples of other clinics and HCPs using the system</td>
<td>Social opportunity Automatic motivation</td>
</tr>
<tr>
<td>Enablement</td>
<td>Facilitating capability and reducing barriers</td>
<td>Providing guidelines, simple checklists, relevant image library for conducting TGC and access to immediate technical support</td>
<td>Physical capability Psychological capability Physical opportunity Social opportunity Automatic motivation</td>
</tr>
</tbody>
</table>
3. The third analysis step (*Policy categories in Figure 11*) identified policy thought to have facilitated the desired behaviour change in HCPs. The identified policies belong to different categories, as listed here:

**Communication**
The co-design process with select HCPs, followed by regular information sessions and updates during the whole project phase. Q&A allowing all HCPs to be involved from an early stage.

**Guidelines**
Checklists created for TGC. Clinical guidelines and recommendations developed for practice.

**Regulation**
National restrictions in place regarding in-person consultations (COVID-19). Encouraged use of TGC from regional and clinical management.

**Environmental and social planning**
Staff were provided with technical equipment in the clinic. Using co-design to plan and design the environment.

**Service provision**
Providing TGC access, training and support to all staff.

**Results from the BCW analysis**
This retrospective analysis and evaluation of the implementation process may not be the traditional way of using the COM-B and BCW models. However, it highlights different components, behaviours and activities of relevance to the successful implementation of TGC, and important activities taking place before, during and after the launch of TGC. This can be recognised as an implementation strategy. These activities likely
affected HCPs’ behaviour change, which facilitated the implementation of TGC in this project. However, the list is by no means exhaustive and requires further investigation and research.

**Before**

- Use a qualified team member to manage the implementation process
- Involve stakeholders in planning, design and implementation
- Prepare the environment to enable use: create guidelines, routines, check-lists, tips and supporting materials, space, laptops, microphones, cameras and access

**During**

- Do not be forceful – let early and late adopters take their own time
- Hold regular meetings with different purposes: education and training; Q&A; inspiration and modelling; and in-person technical support
- Meetings should cater to people with different needs (both mandatory and non-mandatory)

**After**

- Evaluate the efficacy using validated measures of outcome
- Provide feedback to stakeholders
- Use validated outcome measures to create research evidence

This list can be considered part of an implementation strategy and aligns with a call for more research regarding the clinical, technical, organisational and policy questions that arose during the natural experiment of rapid implementation that occurred during COVID-19 (Greenhalgh et al., 2020). Greenhalgh suggests several aspects likely to improve the wide adoption of video consultations in clinical settings: in-person support, training of staff, providing guidelines, and allocating clinical resources (including staff time), also identified by this synthesis.
External context

Naturally, there are many implementation factors not included in these studies, due to time constraints. Yet, the external context cannot be ignored, since it involves uncontrollable factors such as a worldwide pandemic and political decisions. These factors cannot easily be influenced by HCPs’ behavioural change. The regional barriers identified within the Swedish healthcare context are worth mentioning. Despite the goal of eHealth overcoming geographical barriers to access, and improving equality, some invisible structural barriers remain. In Sweden, each healthcare region makes its own economic and strategic decisions, which means that technological systems are often incompatible between clinics and regions. Efficient collaboration and mutual development are sometimes inhibited, thus hampering improved access, quality and development of healthcare provision in general. More detailed evaluations regarding this are necessary for future studies, as discussed below in the chapter on Research implications.

Methodological consideration

It is crucial to select appropriate methods to answer each specific research aim and question. It is not always possible to select, or even know about, the most perfect fit at the time of a study. Despite the careful consideration given to selecting the most suitable methods known at the time of each study, there are specific benefits, pitfalls and drawbacks in every choice made. Below, some of the most relevant considerations are highlighted.

Qualitative methods

Qualitative methods allow explorations of a sample to provide new theories and ideas and create a new foundation from which to conceive new research. However, this approach needs specific actions to avoid pitfalls. Triangulation, transparency, consistency in interviews, and the use of clear protocols and guides are examples of actions that increase
trustworthiness (Shenton, 2004). A qualitative approach is usually very time-consuming and does not provide generalisable results.

**Quantitative methods**

The development of psychometrically sound instruments is important but requires a lot of time and participants. Therefore, this was only done for one of the measures, specifically the GCOS-24swe in Study IV. The psychometric evaluation also involved complex statistical analysis, which requires a high level of expertise. This was addressed by involving a statistical expert in Studies III and IV.

Missing values pose a threat to instrument validity. Study I had up to 31% missing data for certain items, which were excluded to correct for family-wise errors. Bonferroni adjustment was applied as it provides a conservative correction to the data analysis. Study IV had only 0.34% missing data per item that was treated as missing at random, and multivariate imputation was applied to create a complete dataset.

Attrition, i.e. participants dropping out of a study, commonly occurs during data collection (MacFarlane et al., 2014). This happened mainly in Study IV, as there were two different collection points. Response rates dropped between the first and second data collection point from 374 to 254. This equals a 32% drop-out rate, and a drop-out analysis would have been valuable to identify potentially confounding variables.

Research taking place in a real, live setting is complex and there can be many confounding variables. Two important aspects are 1) *Practice effects* – which can occur both in patient participants (regarding any previous experiences of receiving genetic counselling) and HCP participants (regarding their ways of providing the same genetic counselling) and 2) *Procedural inconsistency* - that patient participants are treated differently. Such differences may have occurred in Study 4, for example as patients are selected to receive TGC by video or telephone, included at one of the two participating centres, and when booked to see a GC or a medical doctor. (MacFarlane et al., 2014)
Recruitment

Recruitment issues are very common and important to acknowledge. Systematic selection bias and inclusion/exclusion criteria (for example excluding patients with interpretation needs in all studies) introduce a bias in the sample of selected participants so that they do not represent the general population. This could be addressed by applying a randomised controlled trial method.

All the qualitative studies had rather small sample sizes. However, in Study I this was considered a sound coverage of the practising genetic counsellors in Sweden in 2014. For Study II, with a qualitative inductive approach, recruitment aimed at reaching a broad spectrum of participants instead, purposefully including a wide variety of backgrounds and characteristics using the channels available at the time. Study III was a pilot feasibility study, and the number of participants was deemed sufficient, as was the number of included participants for cognitive interviews (MacFarlane et al., 2014)

Retrospective applications

This research was mostly explorative and additional approaches were identified and incorporated at the time of writing this dissertation, adding to the learning experience. These retrospective applications include the Co-design model in Study III, and the COM-B and BCW models used to compile results in the Discussion. Ideally, these models would have been applied at the time of design, planning and execution of these studies, however, this was not the case. Perhaps if these models had been identified and applied earlier in the research process, this would have guided and structured the research and implementation processes more stringently.
**IMPLICATIONS**

The findings from this research have among other things contributed to a deeper understanding of the field, added to the evidence base regarding the use and implementation of TGC in clinical practice, suggested an implementation strategy, and identified remaining gaps requiring further research.

**Clinical implications**

This dissertation shows that it is possible to improve access to genetic counselling for patients in Sweden. TGC is a complement to in-person GC and can be used to improve access, flexibility and autonomy for patients, since it has proven feasible as well as acceptable to patients and HCPs. Video consultations have shown slightly better outcomes compared to telephone consultations, regarding usability and increased empowerment in patients, but this still requires more research. By providing alternative service delivery models for genetic counselling, clinics can improve patient- centredness and be better prepared for a continued increase in demand, and even for future pandemics.

To facilitate the implementation process, and potentially reduce HCP reluctance, useful activities have been suggested to facilitate the implementation of TGC, while allowing necessary adaptations to the current context. Several suggested activities directly involve HCPs, for example in the co-design process, using iterative development, offering recurrent training and Q&A sessions, and allocating adequate in-person support and resources for HCPs. These activities probably enabled the desired behaviour of using TGC, at least in our clinical setting.

A measure for evidence and evaluation of genetic counselling and TGC has long been needed in Sweden. Now, for the first time, there is a validated and reliable PROM for genetic counselling in Swedish. The GCOS-24swe is appropriate to inform quality evaluations and clinical improvements in the clinical setting to further improve patient care.
Furthermore, GCOS-24swe can be used to inform the genetic counselling sessions, based on the individual’s scores before genetic counselling, to improve patient-centredness. Similar steps have been taken in neighbouring countries Norway and Denmark (Diness et al., 2017; Løvik et al., 2022), potentially encouraging and facilitating more Scandinavian collaborations in the future.

To further improve access to and quality of genetic counselling for patients in Sweden, professional genetic counsellors can be better utilised. This can be done by allowing GCs more time for direct patient care, and by reducing the burden of many administrative tasks. It also involves strengthening the professional GC role as a healthcare professional and providing accredited training and continuing professional education, to assure quality and patient safety.

TGC can be used as a complement to improve access and patient-centredness while maintaining quality in clinical genetics services. Nevertheless, TGC should be based on patient preferences, allowing patients to choose their preferred service delivery mode (video, telephone or in-person consultation) to avoid negative consequences whenever TGC is not a suitable option for the patient, for various reasons.

**Research implications**

More research is needed in the combined field of genetic counselling, implementation science and eHealth. However, some suggested future research studies have already been realised. For example, the lack of higher-level academic education for genetic counselling professionals was addressed in several previous publications (Pestoff et al., 2019; Pestoff et al., 2018; Pestoff et al., 2020). Subsequently, in 2021 a national Master of Science programme for genetic counsellors was launched in Sweden. (Linköping University). It complies with the Professional and Educational Standards of the European Board of Medical Genetics (EBMG) (European Board of Medical Genetics) and was granted EBMG accreditation in 2022, as one of nine accredited MSc programmes in Europe. Thus, European-level quality standards for the GC profession
can be achieved and EBMG professional certification can further improve the quality of and access to professional genetic counselling for all patients in Sweden.

Many different stakeholders have been involved in the included studies, i.e., genetic counsellors, medical doctors, patients, patient representatives, and the management of the Clinical Genetics Department. But, Study I identified the need to gather the experience and opinions of medical doctors in clinical genetics regarding the use of genetic counsellors in the clinical setting. This was also addressed in a qualitative, European-wide study (Paneque et al., 2017), to which I also contributed. The study concluded that access to, and the quality of genetic counselling improved for patients, throughout Europe, including Sweden, when genetic counsellors were utilised in multidisciplinary teams in clinical genetics.

It is appropriate to address the lack of HCPs in general, and genetic counsellors specifically. Relevant research questions related to improving the access to and quality of genetic services to patients are to investigate if and how can tasks be shifted between genetic counsellors and other HCPs (so-called task transfer)? To evaluate whether harmonisation of the genetic counsellor role, quality standards and education improve quality and access for patients, research should involve the key stakeholders including genetic counsellors, medical doctors, patients and clinical management. In addition, it is necessary to consider the role of politicians, as they can ultimately determine the status of HCPs through legislation.

Furthermore, the GCOS-24swe instrument can benefit from further psychometric evaluations, such as establishing test-retest reliability and clinical utility. Establishing the Minimal Clinical Important Difference (MCID) in Sweden, similar to research carried out in the UK, would allow for improved clinical utility (Thomas & McAllister, 2019). As genetic testing is becoming mainstream, evaluation of these healthcare services providing genetic testing, but taking place outside of the
departments of clinical genetics, will become necessary. Future studies can work on an adaptation of the GCOS-24swe into a smaller and simpler format, like the Genomic Outcome Scale (GOS) for non-clinical genetics departments, as done in the UK (Grant et al., 2019; Ting et al., 2021).

Of course, the GCOS-24swe can be used to inform future developments in the field of genetic counselling. Examples of future studies are to evaluate the outcomes of quality and effects of genetic counselling in different countries or patient groups, i.e. hereditary cancer vs other hereditary conditions; in group vs individual genetic counselling consultations; in TGC vs in-person GC, and to evaluate the effects of new eHealth solutions, such as chatbots or web-based counselling aids, to give some examples.
CONCLUSIONS

Although the need for genetic counselling and professionals with adequate training is pronounced, access to and quality of genetic counselling for patients in Sweden remains uncertain. Access was uncertain due to a lack of trained GCs in patient-centred roles, and quality was uncertain because there were no valid outcome measures for this purpose. Nevertheless, we have suggested ways of addressing these uncertainties. Access can be improved by providing genetic counselling from a distance using TGC, and quality can be measured by using valid and reliable outcome measures for genetic counselling. Both patients and HCPs perceive TGC as acceptable and satisfactory in the clinical setting. Also, the implementation of TGC is feasible, showing good levels of demand and effectiveness, when measured using the GCOS-24swe in the Swedish context.

The clinical implications of this research are several and can be used to improve access to genetic counselling, whilst maintaining efficacy and quality. Implementation of TGC in the clinical setting was feasible, but affected by specific implementation activities and (uncontrollable) external circumstances.

Future studies should also include in-depth research regarding the most effective implementation strategy. It is also of interest to measure and evaluate outcomes from genetic counselling improvements in Sweden, by using the GCOS-24swe. Nevertheless, as long as the professional and regional healthcare barriers remain intact, providing equal access and quality genetic counselling to all patients in Sweden by using TGC may prove difficult.
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APPENDICES

Appendix 1: Genetic counsellor web questionnaire (Study I)
Appendix 2: Guide for HCP interviews (Study II)
Appendix 3: GCOS-24swe questionnaire (Study III+IV)
Appendix 4: TUQswe questionnaire (Study III)
Appendix 5: Descriptive HCP questionnaire (Study III)
Appendix 6: Guide for cognitive interviews (Study III)
Appendix 1: Genetic counsellor web-questionnaire (Swedish) used in Study I

Enkätundersökning om genetisk vägledning (anonym)
(förvaltsfrågor om inget annat anges)

Bakgrundsinformation

1. Födelseår
2. Kön
3. Arbetsplats (sjukhus/klinik)
4. Yrketitl
5. År inom yrket
6. Ger du genetisk vägledning till patienter? Ja/Nej
   a. Om Ja hur länge har du gett genetisk vägledning?

För genetiska vägledare

1. Arbetar du som genetisk vägledare? JA/Nej
   a. Hur stor del av din arbetstid ____% 
   b. Om mindre än 100% Vad gör du på din övriga tid? __________________
2. Vilken grundutbildning har du (öppen fråga)

3. Har du någon formell vidareutbildning inom genetisk vägledning
   a. Program (master/magister)
   b. Relevanta kurser (ange vilka)
   c. Uppfård på min arbetsplats
4. Vad är din yrkestitel (öppen fråga):

Dina arbetsuppgifter som genetisk vägledare

Vilka av följande arbetsuppgifter stämmer med vad du gör som genetisk vägledare:

5. Före mottagningsbesöket, gör du något av följande:
   a. Tar fram information inför besöket (admin/ingen patientkontakt)
   b. Journalverifiering
   c. Ritar pedigrees
   d. Gör en riskbedömning
   e. Telefonkontakt med patienten
      i. för att informera om utredningen
      ii. För att samla in information till utredningen
   f. Brevkонтакт/mejl med patienten
      i. för att informera om utredningen
      ii. För att samla in information till utredningen

6. Under mottagningsbesöket, gör du något av följande:
   a. Ger du genetisk vägledning?
      i. Om ja
Appendix 1: Genetic counsellor web-questionnaire (Swedish) used in Study I

1. På egen hand, vid besöket
2. tillsammans med läkare, vid besöket
3. tillsammans med senior GV, vid besöket
4. Annat
   ii. Om nej
      1. Ger inte vägledning, men är med på patientbesök
      2. År inte med på mottagningsbesök

b. Provtagning, tex blodprover

7. Efter mottagningsbesöket, gör du något av följande:
   a. Provhantering, tex registrera och skickar prover
   b. Skriver du remisser för
      i. genetiska analyser
      ii. Annan provtagning
      iii. Kliniska underökningar i samband med genetisk utredning
      iv. Kliniska undersökningar till följd av genetisk utredning (uppföljning)
      v. annat
   c. Har du kontakt med patienter efter mottagningsbesöket
      i. Telefonkontakt med patienter
      ii. Brevkontakt/mejl
      iii. Uppföljande samtal

8. Ingår följande i dina övriga arbetsuppgifter:
   a. Forskning
   b. Utbildning/undervisning av
      i. Patienter
      ii. Medarbetare
      iii. Annan vårdpersonal
      iv. Allmänheten
      v. andra
   c. Fungera som kontaktperson/vårdkoordinator åt patienten
   d. Annat

9. Öppen fråga: Vad är största skillnaden mellan vad du gör som genetisk vägledare och vad du anser att en genetisk vägledare bör göra?
Appendix 1: Genetic counsellor web-questionnaire (Swedish) used in Study I

Den genetiska vågledningssituationen

10. Vem har huvudansvaret att utföra följande moment i genetisk vågledning, enligt dig själv?
   DU kan bara ange ett svar för varje moment. (Välj en: Klinisk genetiker/dr/GV/båda (lika ansvar)/annan (tex barnmorska/ssk)

   • Ta en sjukdomshistoria i släkten
   • Göra en riskbedömning av sjukdomshistorian i släkten
   • Ge patienten information om grundläggande genetik och nedärvningsmönster
   • Ge patienten information om nedärvda sjukdomen och riskbedömning
   • Förmédla genetiska analysresultat
   • Ställa en medicinsk diagnos
   • Diskutera olika möjligheter för genetisk analys och behandlingar
   • Diskutera olika möjligheter för behandlingar
   • Göra medicinska undersökningar
   • Diskutera etiska aspekter av genetisk sjukdom och genetisk diagnostik
   • Göra medicinskbedömningar och överväganden utifrån individ och familj
   • Ge svåra besked på ett empatiskt sätt
   • Diskutera psykosociala aspekter av att leva med ärftlig sjukdom/risk för sjukdom
   • Informera om psykologiska reaktioner såsom sorg och skuld
   • Identifiera och bemöta människor i kris
   • Hjälpa patienten att anpassa sig att leva med sjukdom eller ökad risk för sjukdom
   • Vägleda patienter till att fatta beslut baserat på sin genetiska information, utifrån sin situation
   • Skriva remisser till andra specialistinstanser för vidare analys, tex röntgen
   • Ge icke-direktiv vågledning, på ett neutralt sätt
   • Möjliggöra och uppmuntra patienten att uttrycka sin egen oro och sina egna frågeställningar.
   • Reflektera och värdera sina egna reaktioner i vågledningssituationen
   • Diskutera familjebildning
   • Ge psykologiskt stöd
   • Förskrivaa medicinering vid behov
   • Ha huvudsaklig kontakt med andra medicinska specialiteter
   • Medvetandegöra hur genetisk analys kan påverka familjrelationer
   • Informera om behandling, uppföljnings- och kontrollprogram
   • Medvetandegöra och diskutera känslor och relationer relaterade till genetisk sjukdom
   • Vem bör ge vågledning i följande situationer:
     a. Presymtomatisk testning
     b. Diagnostisk testning
     c. Prenatal diagnostik
     d. Oklara diagnoser
     e. Oklara fynd
Appendix 1: Genetic counsellor web-questionnaire (Swedish) used in Study I

Skillnader mellan en genetisk vägledare och en klinisk genetikers arbetsuppgifter (öppen fråga):

11. Vad är den största skillnaden mellan vad en genetisk vägledare gör och en klinisk genetiker gör i den genetiska vägledningssituationen?
12. Vad anser du att en genetisk vägledare tillför den genetiska vägledningssituationen?
13. Vad anser du att en genetisk vägledare tillför det klinisk genetiska teamet?
Appendix 2: Interview guide for interviews with healthcare professionals about TGC (Swedish) used in Study II

Interviewguide: Vårdpersonal om Genetisk vägledning on-line (genetisk vägledning on-line) 2017-02-27

Introduktionsvinjett (Läses högt för alla deltagare):
"Föreställ dig att en patient som är remitterad för genetisk vägledning kan ha sitt ”besök” med vårdgivaren via videolänk på sin dator/surfplatta/mobiltelefon. Patienten och vårdgivaren kommer kunna se och höra varandra, samt visa dokument och bilder, medan de befinner sig på olika geografiska platser. I denna studie vill vi undersöka vårdgivares och patienters inställningar till Genetisk vägledning on-line – med fokus på acceptans, lämplighet och genomförbarhet av Genetisk vägledning on-line."

Namn: 
Ålder: 
Arbetsplats: 

1. Beskriv din erfarenhet av genetisk vägledning?
2. Beskriv din erfarenhet av web-möten på nätet?
3. Vad är din uppfattning av genetisk vägledning on-line (dvs video-baserad patientkonsultation)?
   a. Kan du beskriva vad som skulle kunna vara barriärer för genetisk vägledning on-line?
   b. Tror du det finns några negativa aspekter? Beskriv dessa:
   c. Tror du det finns några vinster? Beskriv dessa:
   d. Kan du beskriva vad som skulle kunna underlätta för genetisk vägledning on-line?
   e. Kan du beskriva viktiga faktorer för att genetisk vägledning on-line ska fungera bra?
4. Något mer att lägga till?
Frågor om genetisk vägledning och ärfilig diagnos


All information kommer avidentifieras.

Instruktioner:
1. Vänligen läs igenom deltagarinformationen (se separat blad)
2. Fyll i information om dig själv nedan på denna sida
3. Besvara enkäten om din upplevelse av den ärfliga diagnosen i släkten och av den genetiska vägledningen

Om mig:

Kön
☐ Kvinna ☐ Man ☐ Annan

Ålder
______________ år

Vilket medicinskt tillstånd söker du Klinisk genetik för?
☐ __________________________
☐ Vet ej

Har du själv detta tillstånd? ☐ Ja ☐ Nej ☐ Vet ej

Har någon i din släkt detta tillstånd? ☐ Ja ☐ Nej ☐ Vet ej

Har du tidigare fått genetisk vägledning? ☐ Ja ☐ Nej ☐ Vet ej

Har du barn? ☐ Ja ☐ Nej ☐ Vill ej svara

Fortsätt till enkäten på nästa sida
Frågor om genetisk vägledning och ärfilig diagnos


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<td>1 Jag förstår varför jag besöker den genetiska mottagningen</td>
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<td>2 Jag kan förklara vad det medicinska tillståndet innebär för de familjemedlemmar som kan behöva veta</td>
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<td>3 Jag är medveten om vilken påverkan det medicinska tillståndet kan ha på mitt/mina (eventuellt framtida) barn</td>
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<td>4 Jag blir bekymrad när jag tänker på det medicinska tillståndet i min familj</td>
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<td>5 Jag vet var jag ska vänta mig för att få det medicinska stöd som jag och/eller min familj behöver (tex förbyggande åtgärder, mediciner, behandling, kontroller)</td>
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<td>6 Jag tycker att det medicinska tillståndet i min familj har lett till något positivt</td>
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<td>7 Jag upplever att jag har kontroll över hur det medicinska tillståndet påverkar min familj</td>
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<td>8 Jag känner mig positiv inför framtiden</td>
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<td>9 Jag känner att jag kan hantera att ha det medicinska tillståndet i min familj</td>
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<td>10 Jag vet vad jag har för nytta av de alternativ som finns tillgängliga för mig (tex genetisk testning, delta i kontrollprogram, fosterdiagnostik)</td>
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<td>11 Att ha det medicinska tillståndet i min familj gör mig orolig</td>
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<td>12 Jag vet hur det medicinska tillståndet kan påverka mina övriga släktingar på något sätt (tex syskon, farbröder/morbröder, farstrar/mostrar, kusiner)</td>
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<td>Mina beslut rörande det medicinska tillståndet kan påverka framtiden för mitt/mina (eventuellt framtid) barn</td>
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<td>14</td>
<td>Jag förstår varför jag har en remiss till genetiska mottagningen</td>
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<tr>
<td>15</td>
<td>Jag vet hur jag kan få övrigt stöd jag och/eller min familj kan behöva (t.ex. från kurator, Försäkringskassan, kommunen, ekonomiskt stöd, socialt stöd)</td>
<td>☐</td>
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<tr>
<td>16</td>
<td>Jag kan förklara vad det medicinska tillståndet innebär för personer utanför familjen som kan behöva veta (t.ex. vänner, skola, socialtjänst, arbetsplats, habilitering)</td>
<td>☐</td>
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<tr>
<td>17</td>
<td>Jag vet vad jag kan göra för att förändra hur det medicinska tillståndet påverkar mig eller mitt/mina (eventuellt framtid) barn</td>
<td>☐</td>
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<tr>
<td>18</td>
<td>Jag vet vilka i familjen som riskerar att utveckla det medicinska tillståndet</td>
<td>☐</td>
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<tr>
<td>19</td>
<td>Jag tror att mitt/mina (eventuellt framtid) barn kan få ett så gott liv som möjligt</td>
<td>☐</td>
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<tr>
<td>20</td>
<td>Jag kan planera för framtiden</td>
<td>☐</td>
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<tr>
<td>21</td>
<td>Jag har dåligt samvete för att jag eventuellt kan förda det medicinska tillståndet vidare till mitt/mina (eventuellt framtid) barn</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>22</td>
<td>Jag känner mig maktlös att påverka något angående det medicinska tillståndet i min familj</td>
<td>☐</td>
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<td>☐</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>23</td>
<td>Jag förstår varför jag har kontakt med genetiska mottagningen</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>24</td>
<td>Med kunskaper om det medicinska tillståndet kan jag fatta beslut som kan påverka framtiden för mitt/mina (eventuellt framtid) barn</td>
<td>☐</td>
<td>☐</td>
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</tr>
</tbody>
</table>

Tack för din medverkan!
Appendix 4: Telehealth Usability Questionnaire in Swedish (TUQswe) used in Study III

TUQ för distansbesök efter GV

KOD-nr ________________________________

Utvärdering efter distansbesök för genetisk vägledning


Persondata skyddas genom att vara kodade.

Instruktioner till deltagare:
1. Vänligen läs igenom deltagarinformationen (se separat blad)
2. Fyll i information om dig själv och ditt besök nedan
3. Besvara enkäten om din upplevelse av distansbesöket för genetisk vägledning

Om mig:

Kön □Kvinna □Man □Annan
Ålder ________________ år

Vilket medicinskt tillstånd söker du Klinisk genetik för? ________________ □Vet ej

Har du själv detta tillstånd? □Ja □Nej □Vet ej

Har någon i din släkt detta tillstånd? □Ja □Nej □Vet ej

Har du tidigare fått genetisk vägledning? □Ja □Nej □Vet ej

Har du barn? □Ja □Nej □Vill ej svara

Om mitt besök:

1. Vilken typ av besök hade du?
   □Telefon (dvs endast ljud)
   □Video (dvs ljud och bild)
   □Fysiskt (dvs träffas för besöket)
TUQ för distansbesök efter GV

2. Vilken yrkesroll träffade du vid ditt distansbesök?
   □ Läkare
   □ Genetisk vägledare
   □ Båda
   □ Vet ej

3. Hade du träffat samma vårdgivare tidigare?
   □ Ja
   □ Nej
   □ Vet ej

4. Var det ditt val att ha genetisk vägledning på distans?
   □ Ja
   □ Nej

6. Skulle du välja besök för genetisk vägledning via distans igen i framtiden?
   □ Ja
   □ Nej

7. Om ja – Av vilken/vilka anledning/-ar skulle du välja telefon/videomottagning?

8. Övriga kommentarer om distansbesök:

⇒ Fortsätt till TUQ-enkäten på nästa sida
Appendix 4: Telehealth Usability Questionnaire in Swedish (TUQswe) used in Study III

TUQ för distansbesök efter GV

Frågor om hur näjd Du är med Ditt distansbesök för genetisk vägledning


| Fråga | Svar Ändra
<table>
<thead>
<tr>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Distansbesök gör hälso- och sjukvården mer tillgänglig för mig</td>
</tr>
<tr>
<td>2</td>
<td>Distansbesök besparar mig restid vid hälso- och sjukvårdsbesök</td>
</tr>
<tr>
<td>3</td>
<td>Distansbesök tillgodosser mina behov av genetisk vägledning</td>
</tr>
<tr>
<td>4</td>
<td>Systemet för distansbesök var enkelt att lära mig använda</td>
</tr>
<tr>
<td>5</td>
<td>Systemet för distansbesök var enkelt att använda</td>
</tr>
<tr>
<td>6</td>
<td>Jag tror att jag snabbt skulle kunna bli duktig på att använda systemet för distansbesök</td>
</tr>
<tr>
<td>7</td>
<td>Jag gillar att använda systemet för distansbesök</td>
</tr>
<tr>
<td>8</td>
<td>Det är trevligt att använda systemet för distansbesök</td>
</tr>
<tr>
<td>9</td>
<td>Systemet för distansbesök är enkelt och lätt att förstå</td>
</tr>
<tr>
<td>10</td>
<td>Systemet distansbesök klarar allt som jag vill att det ska klara av</td>
</tr>
<tr>
<td>11</td>
<td>Det var lätt att prata med vårdpersonal genom systemet för distansbesök</td>
</tr>
<tr>
<td>12</td>
<td>Jag kunde höra vårdpersonalen tydligt vid användning av systemet för distansbesök</td>
</tr>
</tbody>
</table>
Appendix 4: Telehealth Usability Questionnaire in Swedish (TUQswe) used in Study III

TUQ för distansbesök efter GV

<table>
<thead>
<tr>
<th>Svar</th>
<th>Instämmer inte alls</th>
<th>Instämmer mycket lite</th>
<th>Instämmer lite</th>
<th>Varken instämmer eller inte</th>
<th>Instämmer</th>
<th>Instämmer mycket</th>
<th>Instämmer helt och hållet</th>
</tr>
</thead>
<tbody>
<tr>
<td>13 Jag kände att jag kunde uttrycka mig så som jag ville</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>14 Med systemet för distansbesök kunde jag se vårdpersonal lika bra som om vi träffats på riktigt (OBS! gäller endast vid videobesök)</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>15 Jag tycker att distansbesök är detsamma som att träffas på riktigt</td>
<td></td>
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<td></td>
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</tr>
<tr>
<td>16 När jag gjorde ett misstag i användningen av systemet för distansbesök kunde jag lätt och snabbt fixa det</td>
<td></td>
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<td></td>
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</tr>
<tr>
<td>17 Systemet gav felmeddelanden som tydligt talade om hur felet skulle fixas</td>
<td></td>
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<td></td>
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<tr>
<td>18 Jag känner mig bekväm med att använda systemet för att kommunicera med vårdpersonalen</td>
<td></td>
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<tr>
<td>19 Distansbesök är ett godtagbart sätt att få genetisk vägledning</td>
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</tr>
<tr>
<td>20 Jag skulle använda distansbesök igen vid behov</td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>21 Överlag är jag nöjd med detta system för distansbesök för genetisk vägledning</td>
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</tbody>
</table>

Tack för din medverkan!
Utvärdering av distansbesök för genetisk vägledning
Enkät: vårdpersonal


1. Vilken typ av besök hade du?
   ☐ Telefon (dvs endast ljud)
   ☐ Video (dvs ljud och bild)
   ☐ Fysiskt (dvs träffas för besöket)

2. Vilken typ av besök föredrar du?
   ☐ Telefon (dvs endast ljud)
   ☐ Video (dvs ljud och bild)
   ☐ Fysiskt (dvs träffas för besöket)

   Beskriv vilken/vilka anledning/-ar som ligger bakom ditt svar?

3. Var det ditt val att ha genetisk vägledning på distans?
   ☐ Ja
   ☐ Nej

4. Hur många personer träffade du vid ditt distansbesök?
   ☐ 1
   ☐ 2-3
   ☐ 4-5
   ☐ fler än 5
Personalenkät för distansbesök GV

5. Hade du träffat samma patient tidigare?
☐ Ja
☐ Nej
☐ Vet ej

6. Hur upplevde du distansbesöket?
☐ Bättre än förväntat
☐ Enligt förväntningarna
☐ Sämre än förväntat

7. Vilken typ av besök föredrar du?
☐ Telefon (dvs endast ljud)
☐ Video (dvs ljud och bild)
☐ Fysiskt (dvs träffas för besöket)

Beskriv vilken/vilka anledning/-ar som ligger bakom ditt svar:

6. Övriga kommentarer till hur besöket upplevdes (tex teknikstrul, andra problem, något särskilt bra):

__________________________________________________________

__________________________________________________________

__________________________________________________________

__________________________________________________________
Appendix 5: Descriptive HCP questionnaire (Study III)

Personalenkät för distansbesök GV

Datum

➔ Fortsätt till enkäten
på nästa sida
Appendix 5: Descriptive HCP questionnaire (Study III)

Personalenkät för distansbesök GV

Datum

Frågor om hur nöjd Du är med Ditt distansbesök för genetisk vägledning


<table>
<thead>
<tr>
<th>Svar</th>
<th>Instämmer inte alls</th>
<th>Instämmer mycket lite</th>
<th>Instämmer lite</th>
<th>Varken instämmer eller inte</th>
<th>Instämmer</th>
<th>Instämmer mycket</th>
<th>Instämmer helt och hållet</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Distansbesök gör hälso- och sjukvården mer tillgänglig för mig</td>
<td></td>
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<td></td>
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<tr>
<td>2 Distansbesök besparar mig restid vid hälso- och sjukvårdsbesök</td>
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<tr>
<td>3 Distansbesök tillgodosser mina behov av genetisk vägledning</td>
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<tr>
<td>4 Systemet för distansbesök var enkelt att lära mig använda</td>
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<td>5 Systemet för distansbesök var enkelt att använda</td>
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<tr>
<td>6 Jag tror att jag snabbt skulle kunna bli duktig på att använda</td>
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<td>systemet för distansbesök</td>
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<td>7 Jag gillar att använda systemet för distansbesök</td>
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<td>8 Det är trevligt att använda systemet för distansbesök</td>
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<tr>
<td>9 Systemet för distansbesök är enkelt och lätt att förstå</td>
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<tr>
<td>10 Systemet distansbesök klarar allt som jag vill att det ska klara av</td>
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<tr>
<td>11 Det var lätt att prata med vårdpersonal genom systemet för</td>
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<tr>
<td>distansbesök</td>
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<tr>
<td>12 Jag kunde höra vårdpersonalen tydligt vid användning av systemet</td>
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<td></td>
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<td>för distansbesök</td>
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</tbody>
</table>
## Appendix 5: Descriptive HCP questionnaire (Study III)

### Personalkö för distansbesök GV

<table>
<thead>
<tr>
<th>Datum</th>
</tr>
</thead>
<tbody>
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### Svar

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<th>Instämmer lite</th>
<th>Varken instämmer eller inte</th>
<th>Instämmer</th>
<th>Instämmer mycket</th>
<th>Instämmer helt och hållet</th>
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</thead>
<tbody>
<tr>
<td>13</td>
<td>Jag kände att jag kunde uttrycka mig så som jag ville</td>
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<tr>
<td>14</td>
<td>Med systemet för distansbesök kunde jag se vårdpersonal lika bra som om vi träffats på riktigt (OBS! gäller endast vid videobesök)</td>
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<tr>
<td>15</td>
<td>Jag tycker att distansbesök är detsamma som att träffas på riktigt</td>
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<td>16</td>
<td>När jag gjorde ett misstag i användningen av systemet för distansbesök kunde jag lätt och snabbt fixa det</td>
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<td>17</td>
<td>Systemet gav felmeddelanden som tydligt talade om hur felet skulle fixas</td>
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<td>18</td>
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<tr>
<td>19</td>
<td>Distansbesök är ett godtagbart sätt att få genetisk vägledning</td>
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<td>20</td>
<td>Jag skulle använda distansbesök igen vid behov</td>
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<tr>
<td>21</td>
<td>Överlag är jag nöjd med detta system för distansbesök för genetisk vägledning</td>
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</tbody>
</table>

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**Tack för din medverkan!**
### Intervjuguide för utvärdering av GCOS-24swe enkät

**Deltagarinformation:** Med hjälp av skalan nedan, ringa in det nummer bredvid varje påstående som visar hur mycket du instämmer med påståendet. Vänligen svara på alla frågor. För frågor som inte gäller dig välj nummer 4 (svara varken instämmer eller inte).

**Intervjuguide PROBE (endast till forskaren – deltagare ser ej detta):** används för att få fram detaljerna kring hur respondenten tänkt och förstått innebörden när de läst frågan och vad de svarat och resonemanget bakom detta.

#### Datum:____

**Intervju#:**

**Start kl:____ Slut kl:______**

<table>
<thead>
<tr>
<th></th>
<th>Instämmer inte alls</th>
<th>Instämmer mycket lite</th>
<th>Instämmer lite</th>
<th>Varken instämmer eller inte</th>
<th>Instämmer mycket</th>
<th>Instämmer helt och hållet</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Jag är medveten om varför jag besöker en klinisk genetik-mottagning</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td><strong>Intervjuguide PROBE:</strong> vad betyder Klinisk genetisk mottagning för dig? Vad tycker du ingår i klinisk genetisk mottagning?</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Jag kan förklara vad mitt medicinska tillstånd innebär för de familjemedlemmar som kan behöva veta</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td><strong>PROBE:</strong> Vad betyder ordet medicinska tillstånd för dig? Vilket tillstånd tanker du på då? Vilka menas med &quot;familjemedlemmar&quot; tycker du?</td>
<td></td>
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<td></td>
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</tr>
<tr>
<td>3</td>
<td>Jag är medveten om den påverkan mitt medicinska tillstånd har på mitt (mina) eventuella barn</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td><strong>PROBE:</strong> På vilket sätt tolkar du ordet påverka? Vad menas med eventuella barn? OM du ej har barn – vad skulle du svara?</td>
<td></td>
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<td></td>
<td></td>
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</tr>
<tr>
<td>4</td>
<td>Jag blir upprörd när jag tänker på det medicinska tillståndet i min familj</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td><strong>PROBE:</strong> Kan du beskriva vad du tror man menar med upprörd? Vilka menar du ingår i din familj?</td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Jag vet inte var jag ska vända mig för att få det medicinska stöd som jag/min familj behöver</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
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<tr>
<td></td>
<td><strong>PROBE:</strong> Kan du ge exempel på vart du ska vända dig för medicinskt stöd? Vilken sorts stöd tror du frågan avser?</td>
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<tr>
<td>6</td>
<td>Jag kan inse att det medicinska tillståndet i min familj har lett till något positivt</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td><strong>PROBE:</strong> Kan du beskriva något positivt om du instämmer i frågan?</td>
<td></td>
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</tr>
<tr>
<td>7</td>
<td>Jag kan kontrollera hur detta medicinska tillstånd påverkar min familj</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
</tbody>
</table>
**PROBE:** På vilket sätt kan du kontrollera påverkan på din familj om du instämmer i frågan?

_Hur kom du fram till detta svar?_

8 Jag känner mig positiv inför framtiden

_PROBE:_ Vill du beskriva hur du känner inför framtiden?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

9 Jag kan hantera att ha detta medicinska tillståndet i min familj

_PROBE:_ På vilket sätt hanterar du detta tillståndet i familjen?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

10 Jag vet inte vad jag skulle kunna ha för nytta av de alternativ som står till buds för mig

_PROBE:_ Vad tanker du på för ”alternativ” i denna fråga? Vad menas med ”står till buds”?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

11 Att ha detta medicinska tillstånd i min familj gör mig orolig

_PROBE:_ Hur får det här tillståndet dig att känna dig?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

12 Jag vet inte om detta medicinska tillstånd kan påverka mina övriga släktningar (syssen, farbröder/morbröder, fastrar/mostrar, kusiner)

_PROBE:_ Vad menas med ”påverka” i frågan tror du? Stämmer beskrivningen med din uppfattning om övriga släktningar?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

13 Beträffande det medicinska tillståndet i min familj, så kommer mina beslut inte att påverka framtiden för mina barn eller för de barn jag eventuellt får

_PROBE:_ Vilka beslut kan menas i denna fråga? Hur tolkar du ordet ”framtiden” i denna fråga?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

14 Jag förstår varför min läkare remitterade mig till klinisk genetikmottagning

_PROBE:_ blev du remitterad av en annan läkare? Var du införstådd i remissen till klinisk genetik?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

15 Jag vet hur man kan få det icke-medicinska stöd jag/min familj behöver (t.ex. sociala insatser, ekonomiskt stöd, socialt stöd)

_PROBE:_ vad betyder ”icke-medicinska stöd” för dig? Vad betyder ”sociala insatser” för dig? Kan du ge exempel på ekonomiskt och socialt stöd?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |

16 Jag kan förklara vad det här medicinska tillståndet innebär för personer utanför min familj som kan behöva veta (t ex lärare, socialarbetare)

_PROBE:_ Vad betyder den här frågan för dig? Kan du ge fler exempel på personer som kan behöva veta mer information?

| 1 | 2 | 3 | 4 | 5 | 6 | 7 |
Tack för din medverkan!
Papers

The papers associated with this thesis have been removed for copyright reasons. For more details about these see:

https://doi.org/10.3384/9789179295943
Improving Access and Quality of Genetic Counselling in Clinical Care in Sweden

- The Value of eHealth Solutions and a Validated Outcome Measure

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