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Adaptive Translation of the Genetic Counselling Outcome Scale – A Patient-Reported Outcome Measure for Application in Genetic Counselling in Norway

Iselin Kleppe Løvik, *Master candidate*

Elen Siglen, *Genetic Counsellor at Department of Medical Genetics, Haukeland University Hospital*

Cathrine Bjorvatn, *Associate Professor, University of Bergen*

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Contents

Abbreviations:

GCOS-24 – The Genetic Counselling Outcome Scale

GOS – Genomics Outcome Scale

CGS – Clinical genetics services

PROs – Patient-reported outcomes

PROM/PROMs – Patient-reported outcome measure(s)

PRG – Patient representative group

EP – Expert panel

Tables and figures:

Appendices:

Abstract:

Bakgrunn: The Genetic Counselling Outcome Scale (GCOS-24) er et instrument for pasientrapportert utfallsmål (PROM) opprinnelig utviklet på britisk engelsk. Dens hensikt er å måle og evaluere pasientenes opplevelse av genetisk veiledning. Det originale instrumentet av McAllister, Wood, et al. (2011) er oversatt til flere språk og tilpasset til klinisk praksis i flere land, bl.a. Danmark, Spania, Nederland og Brasil (Diness et al., 2017; Muñoz-Cabello et al., 2018; Segundo-Ribeiro et al., 2020; Voorwinden et al., 2019). En validert norsk versjon av dette spørreskjemaet er ennå ikke tilgjengelig. Denne oppgaven rapporterer prosessen med å oversette og kulturelt tilpasse Genetic Counselling Outcome Scale (GCOS-24) til norsk språk og kontekst. Metoder: Måleinstrumentet ble oversatt etter retningslinjer av Wild et al. (2005), og kulturelt tilpasset etter retningslinjer fra Beaton et al. (2000) som anbefalt av instrumentutvikler. Kognitiv debrifing gjennom intervjuer med en gruppe pasientrepresentanter ble gjennomført for å sikre at påstandene ble forstått som tiltenkt. Resultater: Pasientrepresentantene forsto de fleste påstandene slik de var ment. Tilbakemeldingene fra intervjuene ga innsikt i hvordan spørreskjemaet ble oppfattet og førte til enkelte endringer. Ved å følge retningslinjene som anbefalt, har prosjektet produsert en norsk kulturelt tilpasset oversettelse av GCOS-24 Konklusjon: Resultatene fra oversettelsen, den kulturelle tilpasningen og de kognitive intervjuene har gitt sluttproduktet GCOS-24no, nå klart for psykometrisk testing. Denne studien understreker viktigheten av å følge publiserte, anerkjente retningslinjer for spørreskjemaoversettelse. Resultatene understreker viktigheten av tverrkulturell adaptasjon av pasientrapporterte utfallsmål, selv mellom land med populasjoner og helsevesen som er sammenlignbare.

Background: The Genetic Counselling Outcome Scale (GCOS-24) is an instrument for patient-reported outcome measure (PROM) originally developed in British English. Its intended use is to measure and evaluate patient outcomes in the context of clinical genetics services (CGS). The original instrument by McAllister, Wood, et al. (2011) has been translated to multiple languages and adapted into clinics in several countries, e.g. Denmark, Spain, the Netherlands and Brazil (Diness et al., 2017; Muñoz-Cabello et al., 2018; Segundo-Ribeiro et al., 2020; Voorwinden

et al., 2019). A validated Norwegian-language version of this questionnaire is not yet available. This thesis reports the process of translating and culturally adapting the Genetic Counselling Outcome Scale to the Norwegian language and context (GCOS-24no). Methods: The measure was translated following guidelines by Wild et al. (2005), and culturally adapted following guidelines from Beaton et al. (2000) as recommended by instrument developer. Cognitive debriefing through interviews with a patient representative group were conducted to ensure the understanding of the items' intended meaning. Results: Patient representatives understood most items as intended. The feedback provided insight into how the questionnaire is perceived and led to some changes. Following the guidelines as recommended, the project has produced a final Norwegian product: GCOS-24no. Conclusion: Results of translation, adaptation, and cognitive interviews led to the final product GCOS-24no, ready for psychometric testing. The present study emphasized the importance of following published, renowned guidelines. It confirms the feasibility of cross-cultural adaptation of patient-reported outcome measures and stresses the importance of such adaptation, even between countries where populations and healthcare systems that are comparable.

Keywords:

GCOS-24, PROM, Genetic Counselling, Translation, Empowerment, Cross-Cultural Adaptation

1. Introduction

This section will present the context of the present project, including an outline of why this project was carried out and why it is of significance in the field of clinical genetics and genetic counselling in Norway.

1.1 – The importance of Patient-Reported Outcome Measures

Quantifying differences in patients' experiences with healthcare services is difficult without a proper instrument. Subjective measures aim to quantify an outcome by utilizing the patient's own assessment of healthcare practices. Patient-reported outcomes (PROs) are inherently subjective forms of measures. Payne et al. (2008) include the following in their examples of applications for PROs: patients' reports of a health condition and its treatment, health related quality of life, satisfaction with treatment, or treatment preferences. To quantify such reports, proper instruments are needed for valid and reliable results. An instrument is a valid and reliable tool or device for collecting data about study participants. In the present project, the instrument is a questionnaire that can be distributed to genetic counselling attendees to collect data. A measure is an item or, most commonly a set of items that provides an indication of the quantity or nature of the phenomenon under investigation. Every measure relies on applying underlying dimensions, which are factors or components that provide qualitative values. These are often used to categorize, segment, or provide details about the data used (American Psychological Association n.d.).

Theoretically, the question of overall patient satisfaction and outcome of treatment or health care could be asked directly as 'How satisfied are you with your medical care?' This may be measured on a 5 or 7-point Likert scale with numerical or ordinal values, ranging from "dissatisfied" to "highly satisfied". The problem with these categorical measures is the lack of uniformity in distance between numerical values of the categories (D'Orazio, 2021). In other words, it is hard to quantify. For example, if 'dissatisfied' provides a score of 1, and on the other end of the scale 'highly satisfied' provides a score of 7, the values mean little when not seen in relation to one another. Such a measure, when not universal and established with the necessary conceptual basis, cannot really compare data between studies or between subjects.

Alas, it is only useful when comparing scores at different points in time, measuring a change within the same individual. The scale will tell the researcher what subjective satisfaction levels exist in the sample, but the lack of uniformity leads to inaccurate data. This type of scale gives rise to problems with generalization to larger populations or comparisons between samples, as the perception of satisfaction will vary with individuals and populations. The lack of construct validity makes it unable to quantify and compare with other measures.

A 'Patient-Reported Outcome' (PRO) is a direct report of the patient's health, quality of life, or functional status in the face of health care or treatment. Such reports are direct from the patient and not interpreted by a clinician. They can be used in absolute terms such as pain severity ratings, or in relative terms such as change in measures of side effects following drug treatment (Weldring & Smith, 2013). To measure such reports, commonly used instruments are 'Patient-Reported Outcome Measures' (PROM). They are often self-administered questionnaires that measure the patient's health status, quality of life and symptoms, functionality, physical, mental, or social health (Santana et al., 2018; Weldring & Smith, 2013). By applying a PROM, one can measure the underlying dimensions of a specific construct through direct or indirect statements regarding the patient's subjective experience. This will in turn provide results from patients' responses that are both valid and comparable across participants, studies, and samples. The value of such measures, although subjective, should not be underestimated. According to patient-centred care, it is the patient's own evaluation of outcome that decides the objective value of the treatment or intervention. This comes as opposed to value-based care, which defines value in health care as quality of care modified by cost (Tseng & Hicks, 2016).

The expansion of PROMs used outside of clinical research has led to a wide recognition of their potential to transform health care, by both clinicians and regulatory bodies. Data from PROMs can be used to improve both safety and quality of the practices by recognizing the patient as the main decision-maker in treatment (Weldring & Smith, 2013). With this expansion, a need for guidance on development, translation and adaptation, use, measurement, and analysis of PROMs has emerged. When developing a PROM, a conceptual foundation needs to be established (Weldring & Smith, 2013). When thoroughly constructed, a validated PROM can utilize recognized constructs that have a measuring value for the treatment or health

care in question. PROs from such a questionnaire will in turn produce reliable results that are, among others, comparable with studies that have used the same measure. Thus, the subjectivity of the outcomes is framed within the given construct to utilize the measures to promote patient-centred care.

'Patient-Centred Care' was first conceptualized by Balint (1969), and has been an evolving concept in health care, focusing on understanding the patient as a unique human being. It has later been acknowledged as an essential foundation and a high priority for how to build and improve successful health practices (Santana et al., 2018). A patient-centred approach to health care must base its evaluation of outcomes on the patient itself and the outcomes that the patient values as a result of the health care they receive. Only by taking into consideration the valuable role of the patient, can the health care be truly personalized and patient-centred. The patient-centred care model encourages health care workers to collaborate with patients in designing their individualized care, which will improve the quality and efficiency of healthcare (Santana et al., 2018). In their study, Santana et al. (2018) propose PROMs as one of the outcome components in their framework for achieving patient-centred care. 'Integrating these measures into clinical practice have shown to improve outcomes as well as improve quality of care' (Santana et al., 2018, p. 435). This finding supports the notion that PROMs and patient-centred care are undeniably affiliated. One can benefit from applying PROMs in evaluating interventions, working towards a more patient focused practice, and as a result improve outcomes in genetic counselling.

1.2 – The need for a genetic counselling specific PROM in Norway

'Genetic counselling' as defined by Fraser (1974, p. 637) is a 'communication process which deals with the human problems associated by occurrence, or the risk of occurrence of a genetic disorder in a family'. The main goal of genetic counselling, also constituted by law in Norway, is to facilitate informed decision making in patients and families before, during, and if needed, after their decision about testing for genetic conditions (the Biotechnology Act, 2003). According to Fraser (1974) the process should involve an attempt by at least one appropriately trained person; a genetic counsellor, geneticist, or specialized doctor to help the individual or family in achieving five goals:

> (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. (Fraser, 1974, p. 637)

Genetic counselling has been defined by various authors, in various studies since. Most definitions are based on the work of Fraser (1974), hence this definition is chosen as the most relevant. This definition and its associated goals can be attributed to the relevant PROs to be measured in genetic counselling. It is crucial that the definition makes the foundation for the desired outcomes of genetic counselling (Yuen et al., 2020). These five goals, all contribute to the main goal of genetic counselling; empowering the patient to understand their health challenges, and to make decisions that gives hope for the future. By including the goals in the development of a genetic counselling specific PROM, the measure can be used to gather information on the patients' outcome from the intervention, which in turn can be utilized to increase the quality over time (McAllister, Wood, et al., 2011).

A systematic review performed by Payne et al. (2008) ahead of development of a genetics specific PROM, found 67 different measures spread across 1688 studies and articles worldwide that were used to evaluate outcomes in clinical genetics. Only 30 were genetics specific, and 46 out of the 67 different measures were used and reported in only one study each. This finding is indicative of the variety and disagreement on appropriate use of measures. This inconsistency supports the notion that guidelines are needed for development of conceptually founded PROMs. The study concluded that none of the 67 measures used on its own addressed all potential benefits or outcomes of using clinical genetics services. This systematic review was an attempt to structure the approach in identifying which validated outcome measures serve as realistic options for robust, evidence-based approaches to the potential benefits or deficiencies of 'Clinical Genetics Services' (CGS).

McAllister and Dearing (2015) mention such outcome evaluations to have been problematic with no clear consensus about which PROs are directly attributable to genetic counselling. As a result, more investigation was needed before establishing a PROM for use in CGS.

Clinical genetics services in this context refers to genetic testing and counselling by a genetic counsellor or geneticist in the genetic out-patient clinic. The information provided in counselling is intended to enable the patient to make well-informed decisions about their future and potential post-test support systems (e.g., cardiological surveillance, mammogram screening or MRI) tailored to the individual. There are many examples from Norway of genetic testing increasingly being performed outside of the Department of Medical Genetics and the traditional counselling session. These may include diagnostic testing for common *BRCA*-mutations in breast- and ovarian cancer patients, or prenatal diagnostics performed in women's and birth clinics. These services are often referred to as genetic information, and are different from counselling in CGS, as it is not performed by health care workers specifically trained in medical genetics. A genetics specific PROM is especially important because of the difference between CGS or other counselling-based healthcare services and treatment-based services. Treatment value in cases of genetic conditions cannot be measured by the number of people recovered from disease (McAllister & Dearing, 2015).

Studies have provided evidence that patients who attend CGS are often seeking information together with a supportive relationship. The reported benefits of CGS include relief of uncertainty and feelings of vulnerability, increased self-efficacy, and adaptation to the genetic condition in the family (Bernhardt et al., 2000; Macleod et al., 2002; McAllister et al., 2008; Payne et al., 2007; Skirton, 2001; Slomp et al., 2018). The outcome from a counselling service should rather focus on such information as well as social support, as this facilitates informed decision making, an important goal of genetic counselling. The accuracy of such information can be measured with objective measures. However, for measuring the outcome values, focusing on the patient's perspective may be a more relevant approach (McAllister & Dearing, 2015). The empowerment construct, which will be addressed in the next section has been found to have the highest correlation with patient outcomes from genetic counselling. McAllister, Dunn, et al. (2011) propose the construct as a

measure and conceptual foundation for such outcomes. In their structured review on PROs, the authors applied the theoretical framework to the measures available. Empowerment was identified as a key patient outcome goal of genetic counselling, and has been used as an overarching construct to represent many other PROs used for CGS (McAllister & Dearing, 2015).

A patient-reported measure in this regard, using empowerment as the framework for obtaining outcome data related to genetic counselling, has not yet been established in Norway. Until now, research on patient reports has been based on data from the already translated Norwegian version of the 'Satisfaction with Genetic Counselling scale' (Bjorvatn et al., 2007). This scale measures different dimensions of patient satisfaction; instrumental, affective, and procedural. Satisfaction is relevant for service evaluations, but it does not include all potential benefits from genetic counselling. When considering both empowerment and the goals of genetic counselling as listed by Fraser (1974), the Satisfaction with Genetic Counselling scale falls short. This measure is neither designed for, nor intended to contain said goals of genetic counselling, which leaves an opening for the instrument in the present project. There is unexplored value in having a Norwegian instrument available that measures the actual patient-reported outcomes and how these outcomes might empower the patients after a genetic counselling session.

The Department of Medical Genetics, Haukeland University Hospital has expressed a desire for a translation and adaptation of the 'Genetic counselling Outcome Scale' (GCOS-24) into Norwegian language and context. In light of the pandemic, there have been drastic shifts into digital alternatives to the traditional genetic counselling sessions. Such digital alternatives include video or phone-based consultation platforms, as well as an emerging field of web-based solutions such as chatbots (Siglen et al., 2021). With counselling sessions varying between patients, it might be fair to assume an increase of variation in outcomes. As the field of medical genetics is rapidly and continuously progressing, genetic counselling must dynamically adapt to both the expanding field of research as well as the individual's needs. With this variety, an instrument in this regard is even more relevant for assessing the individual needs of patients facing the various counselling services. A revision of the Biotechnology Act (2003, § 5-5 added June 19th 2020) decided that genetic counselling in Norway should increasingly adapt to individual's needs rather

than follow a strictly general informational form. This is in line with promoting patient-centred care in genetic counselling, as it tailors to the individual's needs. This also supports why the patients themselves are the most reliable source for reports. The produced empowerment score tells the clinician whether the patient has received the support that genetic counselling is meant to provide. An instrument in this regard would make way for new research that allows for comparisons across specialties, e.g., studying differences in patient outcomes when counselled for the various cancers, at-risk pregnancies, or other genetic conditions.

1.3 – Purpose, aim and delimitation

1.3.1 – Purpose of the project

This study describes the process of translating and cross-culturally adapting the Genetic Counselling Outcome Scale' (GCOS-24) to Norwegian language and context. Evaluations of clinical genetics services in Norway require a reliable and valid 'Patient Reported Outcome Measure' (PROM) that captures all relevant outcomes of genetic counselling. Empowerment, the underlying construct of the GCOS-24 measures these outcomes based on genetic counselling goals. The nature of the project requires a dynamic method that utilizes equivalence, construct retainment, cognitive interviews, and cross-cultural adaptation to ensure a translation of high quality.

1.3.2 – Aim of the study

The aim of the study is to translate the 'Genetic Counselling Outcome Scale' (GCOS-24) according to guidelines for adaptation of 'Patient-Reported Outcome Measures', utilizing ISPOR's translation and Beaton's cross-cultural adaptation methods. The process produces a final Norwegian language PROM: 'GCOS-24no'. This corresponds with the instructions provided by the instrument developer after being granted permission to translate and use the instrument in a Norwegian context.

1.3.3 – Delimitation

Reliability and validity testing to prepare for an upscaled use among patients fall outside the scope of this study, but will be initiated by Department of Medical

Genetics, Haukeland University Hospital as soon as the present study has been completed. Further, the project opens the possibility of a simultaneous validation of the Norwegian short form GOS if desired.

2. Theoretical foundation of the study

The goal of this chapter is to position the present study in its field of research, providing relevant literature for the execution of the project. The theories and dimensions that form the foundation of the GCOS-24 are here introduced. This section also discloses the psychometric properties and development process of the original instrument.

2.1 – Empowerment makes the dimensional foundation of the questionnaire

Patient empowerment has been explored thoroughly in the context of genetic counselling by McAllister and Dearing (2015). The authors define 'Empowerment' as: 'a set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future' (McAllister, Dunn, et al., 2011, p. 125). Empowerment included four dimensions:

> the beliefs that one (1) can make important life decisions in an informed way (Decision-Making), (2) has sufficient information about the condition, including risks to oneself and one's relatives, and any treatment, prevention and support available (Knowledge And Understanding), (3) can make effective use of the health and social care systems for the benefit of the whole family (Instrumentality) and (4) can look to the future with hope for a fulfilling family life, for oneself, one's family and/or one's future descendants (Future Orientation). (McAllister, Dunn, et al., 2011, p. 125)

In addition to these four components underlying the empowerment construct, further research identified the need for adding a fifth dimension when used in a genetic counselling context. Participants in the qualitative study by Yuen et al. (2020) proposed the addition of the dimension 'emotional regulation'. The participants – both patients and counsellors emphasized the importance of support and guidance on how to control and regulate one's emotions. It is instrumental in how

they perceive and evaluate the outcome of genetic counselling and is valued as a significant part of counselling. Counsellors recognized the importance of families to address and deal with the difficult emotions that may arise in facing genetic risk and disease. Another change proposed by the participants was renaming dimension 4 'Hope' which is a more widely known term, to further emphasize the feeling of hopefulness which was considered important by participants. Empowerment was found by McAllister, Dunn, et al. (2011) to share conceptual similarities with the three dimensions cognitive, decisional and behavioural control as captured by the 'Perceived Personal Control' construct. This resulted in further renaming dimensions 1-3 in the empowerment construct. McAllister and Dearing (2015) designed the current framework to capture:

> (1) Cognitive control: having sufficient knowledge and understanding about the condition, including risks to oneself and other relatives. (2) Decisional control: having options or feeling able to make informed decisions between options for managing risk. (3) Behavioural control: feeling able to use the health and social care systems effectively to reduce harm/improve life for oneself and other relatives. (4) Emotional regulation: feeling able to effectively manage emotional consequences of genetic information. (5) Hope: for a fulfilling family life for oneself, relatives and future descendants (McAllister & Dearing, 2015, p. 116).

2.2 – The Genetic Counselling Outcome Scale (GCOS-24)

In 2011, the GCOS-24 was developed by McAllister, Wood, et al. (2011) with the purpose to identify qualitatively, and measure quantitatively the empowerment construct consisting of the five dimensions of outcomes from genetic counselling. These are measured through 24 items in the GCOS-24, which in turn produces the empowerment score for each respondent (McAllister, Wood, et al., 2011). The GCOS-24 is now a well-established PROM for use in genetic counselling. In the process of developing the questionnaire, an item pool of 84 items was generated from qualitative data, items from the 'Perceived Personal Control scale' and a subscale of the 'Revised Illness Perception scale' called emotional representation. Their study sample was derived from patient support groups (N=527) of whom empowerment

levels would not likely change significantly for test-retest reliability. Their responses to the original 84-item questionnaire were analysed. From the 84 items, 3-5 items from each factor were selected by high factor loadings but excluded if similar.

The empowerment construct identified through earlier qualitative research by McAllister, Dunn, et al. (2011) contributed to directing the selection of items through providing insight into the items focused on the more troubling issues in genetic conditions. This includes items addressing communication with at-risk family members and feelings of guilt and shame among patients. In the process of testing the final 24-item questionnaire, the sample (N=241) was drawn from the actual population of patients, who were evaluated both before and after counselling. This sample gave support for the evidence of sensitivity to change with a medium-to-large effect size (Cohen's d = 0.70). Construct validity was tested with correlation with the GCOS-24 scores and measures of various other constructs, including perceived personal control, and satisfaction with life which were found to explain about 30% and 16,8% of the variance in empowerment respectively. With these results, the empowerment construct measured through the 24 items shows good convergent validity (McAllister, Wood, et al., 2011). The final product is presented in figure 1.

The Genetic Counselling Outcome Scale (GCOS-24)

Using the scale below, circle a number next to each statement to indicate how much you agree with the statement.
Please answer all the questions. For questions that are not applicable to you,
please choose option 4 (neither agree nor disagree).

1 = strongly disagree
2 = disagree
3 = slightly disagree
4 = neither disagree nor agree
5 = slightly agree
6 = agree
7 = strongly agree

		strongly disagree	disagree	slightly disagree	neither agree nor disagree	slightly agree	agree	strongly agree
1	I am clear in my own mind why I am attending the clinical genetics service.	1	2	3	4	5	6	7
2	I can explain what the condition means to people in my family who may need to know.	1	2	3	4	5	6	7
3	I understand the impact of the condition on my child(ren)/any child I may have.	1	2	3	4	5	6	7
4	When I think about the condition in my family, I get upset.	1	2	3	4	5	6	7
5	I don't know where to go to get the medical help I / my family need(s).	1	2	3	4	5	6	7
6	I can see that good things have come from having this condition in my family.	1	2	3	4	5	6	7
7	I can control how this condition affects my family.	1	2	3	4	5	6	7
8	I feel positive about the future.	1	2	3	4	5	6	7
9	I am able to cope with having this condition in my family.	1	2	3	4	5	6	7
10	I don't know what could be gained from each of the options available to me.	1	2	3	4	5	6	7
11	Having this condition in my family makes me feel anxious.	1	2	3	4	5	6	7
12	I don't know if this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	1	2	3	4	5	6	7
13	In relation to the condition in my family, nothing I decide will change the future for my children / any children I might have.	1	2	3	4	5	6	7
14	I understand the reasons why my doctor referred me to the clinical genetics service.	1	2	3	4	5	6	7
15	I know how to get the non-medical help I / my family needs (e.g. educational, financial, social support).	1	2	3	4	5	6	7
16	I can explain what the condition means to people outside my family who may need to know (e.g. teachers, social workers).	1	2	3	4	5	6	7
17	I don't know what I can do to change how this condition affects me / my children.	1	2	3	4	5	6	7
18	I don't know who else in my family might be at risk for this condition.	1	2	3	4	5	6	7
19	I am hopeful that my children can look forward to a rewarding family life.	1	2	3	4	5	6	7
20	I am able to make plans for the future.	1	2	3	4	5	6	7
21	I feel guilty because I (might have) passed this condition on to my children.	1	2	3	4	5	6	7
22	I am powerless to do anything about this condition in my family.	1	2	3	4	5	6	7
23	I understand what concerns brought me to the clinical genetics service.	1	2	3	4	5	6	7
24	I can make decisions about the condition that may change my child(ren)'s future / the future of any child(ren) I may have.	1	2	3	4	5	6	7

Figure 1 - GCOS-24 (McAllister et. al., 2011)

2.3 – The Genomics Outcome Scale (GOS)

In 2018, GCOS-24 was modified and reduced to a six-item scale in the new questionnaire 'Genomics Outcome Scale' (GOS) by Grant et al. (2019). Their primary motivation for doing so was the need for a PROM to be put into the context of not only genetic counselling, but also associated genomic testing services like in paediatrics or oncology. The nature of this questionnaire makes it more tailored to the services that provide genetic information outside of CGS in Norway. The authors also highlight the increasing use of 'main-streaming genetic testing', occurring in cancer predisposition genes and neurogenetic testing. 'This study aims to take the first step towards establishing a PROM which would be appropriate for routine use in audit and clinical evaluations of genetic services. The specific aim is to develop a short form of the GCOS-24 (using both qualitative and IRT methods), suitable for use both within and outside the context of CGS and in research, which still appropriately captures the empowerment construct' (Grant et al., 2019, p. 326). The study emphasizes the usefulness of a shorter version of the GCOS-24 as more applicable for the use of clinical genetics services outside of a traditional counselling session (i.e., non-physical counselling, prenatal diagnostics, and diagnostic testing without pre-counselling).

Regarding the length of the questionnaire, a shorter version will most likely increase responsiveness among patients through reduced completion time, making it easier to integrate into main-streaming genetic testing. A major factor in the purpose of the study was to retain the GCOS-24 empowerment construct, carrying it into GOS. The final questionnaire was found to correlate significantly with both GCOS-24, and the empowerment construct underpinning the items (r = 0.838 at 99% confidence). The ten items with the most consistent information curves, that were not redundant and with well discriminative properties, were put into Rasch Analysis. Optimal performance was proven with the combination of items 4, 16, 17, 18, 20 and 24, resulting in the final product of GOS (Figure 2) (Grant et al., 2019).

		Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
1	I can explain what the condition means to people outside my family who may need to know (e.g. teachers, social workers).	1	2	3	4	5
2	I know who else in my family might be at risk for this condition.	1	2	3	4	5
3	When I think about the condition in my family, I get upset.	1	2	3	4	5
4	I know what I can do to change how this condition affects me/my children.	1	2	3	4	5
5	I am able to make plans for the future.	1	2	3	4	5
6	I can make decisions about the condition that may change my future or my child(ren)'s future.	1	2	3	4	5

Figure 2 - Genomics Outcome Scale (Grant et. al. 2019)

2.4 – Properties of GCOS-24 and GOS

Notable differences between GCOS-24 and GOS include changing the 7-point Likert scale to a 5-point scale, removing the non-ordinal point of 'not applicable', as well as the vague 'slightly' (agree/disagree) point. This could be performed due to the removal of poor discriminative items from GCOS-24, resulting in items that can be assumed to apply to all patients involved in CGS. All reversed items worded with 'I don't' (know) were changed to affirmative answers, so that all the measured data result in positively scored values (Grant et al., 2019). Reversed items are a common solution to revealing fabricated or random answers from respondents of self-reported questionnaires, which might be the case for a 24-item scale. Since GOS includes only 6 items, the nature of the negatively worded questions may create more confusion than it is a necessity, as the length does not promote fatigue in respondents when filling out the questionnaire.

2.5 – Previous research

Studies published in different countries that utilize their translations of the GCOS-24 in their practice or research, have proven its usefulness in various contexts. Among these are service evaluations in both the UK and Canada, with consistent results showing significant improvement of empowerment scores in patients post-counselling (Costal Tirado et al., 2017; Inglis et al., 2015; McAllister, 2016). GCOS-24 has also been used for quality improvement of counselling services, assessing

different approaches of gathering pedigree information in counselling, and comparing outcome levels in a support group of adult participants with parents of child participants (Costal Tirado et al., 2017; Palmer et al., 2018; Slomp et al., 2018). Costal Tirado et al. (2017) suggest that there is value in using this measure and its outcome scores to evaluate both cost and time seen in relation to change in outcome and overall benefit. Slomp et al. (2018) found the measure useful in evaluating whether patients would benefit from the pedigree information being collected prior to the counselling session, as opposed to during. Their findings suggest that there was no difference in outcome levels between groups (p = .369). These studies demonstrated that clinical genetic counselling services can deliver measurable patient benefits. It is clear from investigating the literature that GCOS-24 needs further exploration, and that the full scope of usefulness has not yet been established.

Several countries, such as Denmark, Brazil, Spain, and the Netherlands have already translated and adapted the GCOS-24 into their language and cultural context (Diness et al., 2017; Muñoz-Cabello et al., 2018; Segundo-Ribeiro et al., 2020; Voorwinden et al., 2019). The scale is increasingly applied in genetic counselling in different countries and makes a Norwegian version even more valuable for both research and clinical purposes. Not all these countries are comparable to Norway in culture, but experiences with translating and culturally adapting the GCOS-24 can be inferred from Denmark (Diness et al., 2017). Denmark has similarities to Norway both in terms of language and culture. Therefore, it may be useful to take the Danish authors' experiences with translating the questionnaire, into account in the present study. Still, it might be useful to note the few differences in regulations of medical use of biotechnology, where Denmark has a slightly more liberal legislation than does Norway. Thus, Danish law allows double donation and doesn't explicitly prohibit surrogacy for assisted reproduction, Norwegian law does not allow either technique (Pedersen et al., 2022). The results from Diness et al. (2017) suggested a successful adaptation of the questionnaire into the target language. With 61 patients completing the adapted Danish version of the questionnaire, it obtained an internal consistency by Cronbach's alpha of $\alpha = 0.79$, with the number of missing responses kept to a degree deemed acceptable. These results can be used as a point of reference when evaluating and interpreting the results of the present study, and the later process of validity and reliability testing for upscaled use of the Norwegian GCOS-24.

3. Methods:

3.1 – Setting

The present study utilizes two converging methods in the process of translating and adapting GCOS-24. These include translation, following ISPOR's 'Principles of Good Practice' guidelines developed by Wild et al. (2005), and cross-cultural adaptation with guidelines developed by Beaton et al. (2000). The term 'Cross-Cultural Adaptation' as defined by the authors is 'a process that looks at both language (translation) and cultural adaptation issues in the process of preparing a questionnaire for use in another setting' (Beaton et al., 2000, p. 3186). The study will be conducted as illustrated in figure 3. Further, the description of each step performed in the project, are presented in tables (1-4) as proposed by Wild et al. (2005). Beaton et al. (2000) proposes such a report for full transparency of decisions made by the expert panel for achieving equivalence between the source and target instrument. The cross-cultural adaptation is further described as a thorough process designed to maximize the attainment of semantic, idiomatic, experiential, and conceptual equivalence across cultures. The expert panel meetings, and their discussions of the translations are essential for this purpose.

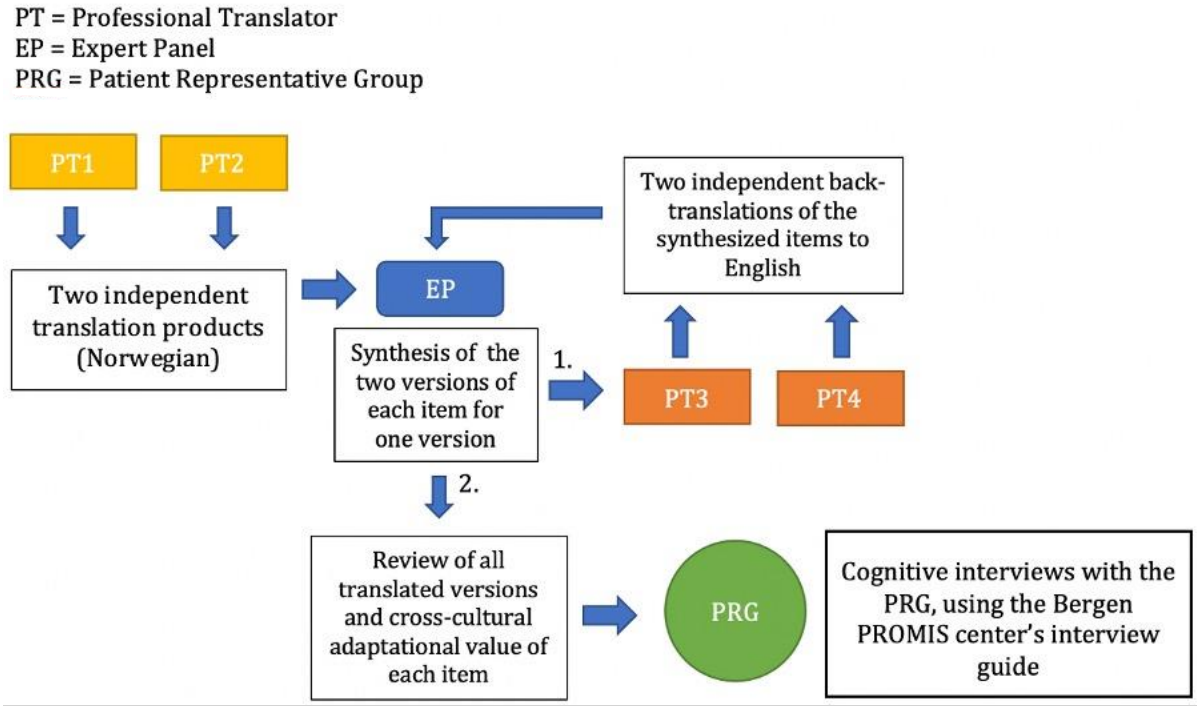

Figure 3 – Translation process in the present study, adapted from Wild et al. (2005) and Beaton et al. (2000).

Beaton et al. (2000) propose four areas to be focused through the expert panel meetings. 'Semantic equivalence' entails the transference of the meaning of the words across translation. There may be multiple meanings or grammatical differences or difficulties between languages. 'Idiomatic equivalence' refers to phrases common to, or exclusively understood by a particular population. In translation, idioms from the source country benefit from being replaced with equivalent expressions, adapted to the target country. 'Experiential equivalence' addresses the items seeking to capture experiences of daily life in a specific country or culture. To reach equivalence, statements from the source country that are not equal to experiences in the target country must be replaced. Finally, 'conceptual equivalence' addresses challenges of words with different conceptual meaning between cultures (Beaton et al., 2000).

Table 1: Step 1 – Preparations as outlined in Wild et al. (2005).

Critical components	Rationale	Execution
1. Obtaining permission to use the instrument.	Respect copyrights.	Through e-mail correspondence with Wiley journal, Haukeland University Hospital was granted permission for translation and non-profitable use of the instrument for research.
2. Consulting REK[1] about approval and ethical concerns.	Ensure privacy for participants' data, as well as safety for their mental and physical health.	The privacy representative at Haukeland University Hospital was consulted to evaluate the project. It was confirmed that privacy and safety were not compromised, and no further approvals were needed.
3. Ordering translation with the company.	Obtain a professional translation and ensure grammatical accuracy.	Haukeland University Hospital (RKAK[2]) placed an order with a professional translation company.
4. Recruiting participants for the expert panel.	Ensure that the translation is context-specific and prevent the loss of equivalence in terms through translation.	RKAK recruited specialists involved in future use and implementation of GCOS-24. This is in accordance with the cross-cultural adaptation method from Beaton et al. (2000).
5. Reporting and explaining of concepts in the instrument.	To strengthen the conceptual equivalence of the forward translations, and to avoid ambiguities.	Using the Empowerment definition from McAllister and Dearing (2015), the expert panel was well informed of the concept's five dimensions.

[1] Regional Committee for Medical Research Ethics
[2] Regional Competence Center for Hereditary Cancer

		Participants were asked to keep the empowerment construct in mind while evaluating the translations.

3.1.1 – Participants

The two main groups of participants that were identified through the literature of Wild et al. (2005) and Beaton et al. (2000) are the 'Expert Panel' (EP) that discuss the translations and 'Patient Representative Group' (PRG) that are included in the cognitive interviews. The EP was formed from an interdisciplinary team from the Department of Medical Genetics, Haukeland University Hospital, all with different backgrounds and expertise. The panel included a geneticist, two experienced genetic counsellors, a research nurse, an administrative manager without any medical background, as well as the master candidate together with the supervising genetic counsellor (n = 7). The PRG consisted of volunteer patients that had been counselled in the period of recruiting (n = 8).

The translation was conducted by four independent professional translators, none included in the EP. Some modifications were made to the originally proposed method by Wild et al. (2005) due to cross-cultural adaptation, as well as convenience. Changes were discussed by the research team and consist of excluding the translators from the expert panel and postponing the meeting with instrument developer until after the cognitive interviews. This was decided for the sake of keeping the translators independent and context unaware. Postponing the meeting with instrument developer from step 10, as suggested by (Beaton et al., 2000) to step 16 was to make sure all issues were exposed, thus ensuring full profit of the meeting.

3.2 – Forward translation

The forward translation was conducted according to the ISPOR guidelines (Wild et al., 2005), by two independent translators who produced two Norwegian forward translations of the original instrument, named FT1 and FT2. These two translations were further discussed in the first EP meeting, according to guidelines by Beaton et al. (2000) to address discrepancies and ambiguous wording. The EP discussion was recorded with permission from all participants, in order to strengthen the design's replicability and transparency of reporting. From the two translations, one reconciled forward translation, named FTX, was produced and sent to the

translation company for back translation. The execution of these steps and their rationale are presented in table 2.

Table 2: Step 2 – Forward translation and reconciliation, as suggested by Wild et al. (2005).

Critical components	Rationale	Execution
6. Comparing and evaluating the two forward translated Norwegian versions.	Detect errors and discover ambiguity in the translated versions. Reducing bias and maintaining concept validity through translation.	Always keeping the construct in mind, as well as comparing ambiguous wording to the original instrument, the most suitable wording was after thorough discussion selected by the EP.
7. Suggesting changes to the translations.	Adding clinical context to the professional translations, and correcting wording that was not sufficient in the initial translations. This ensures idiomatic equivalence, and tailors the questionnaire to the target country.	If none of the two translations achieved a sufficient standard when translating an item, the expert panel would select a third wording more suitable to the context of CGS.
8. Reconciliation.	Obtain one single Norwegian product that back translates similarly to the original English instrument.	The most suitable wording for each item was patched together into a joint Norwegian version that was sent back to the company for back translation.

3.3 – Back translation

The back translation was conducted by the same translation company as the forward translation, but with two new translators, as recommended in the ISPOR guidelines (Wild et al., 2005). From the reconciled forward translation (FTX), two back translations were produced, called BT1 and BT2. In addition to language adaptation, other aspects of geographically delimited differences between the source and target countries were addressed in this step as cross-cultural adaptation was applied. A thorough use of back translation is proven effective in translating scales from source to target language (Hagell et al., 2010; Lee et al., 2019). The EP discussion was once again recorded with permission from all participants, for strengthening the design's replicability and transparency of reporting. For successfully implementing the instrument in the Norwegian healthcare system, adaptation of the measure must consider the differences between the two countries. It is crucial that the method

considers both differences in culture, as well as the structure of the respective healthcare systems. From the two back translations, the EP discussed discrepancies and ambiguities and applied changes to the FTX. This produced a prefinal version, named FTY, ready for cognitive debriefing. The execution of these steps and their rationale are presented in table 3.

Table 3: Step 3 – Combined back translation and harmonization in accordance with Wild et al. (2005) and Beaton et al. (2000).

Critical components	Rationale	Execution
9. Back translations of the Norwegian reconciled version back into the source language.	This step is meant as a quality control, to reveal any unintentional changes to the meaning derived from each item.	Back translated versions should show similarity and absence of major changes to the original version. Any items diverging between versions suggested a need for a change of phrasing of the Norwegian instrument.
10. Review of the back translations against the original instrument.	The review is to ensure equivalence of the translation. The EP should focus on detecting the items that show experiential and conceptual inequivalence, and take appropriate action.	Discrepancies between the two back translations were seen in comparison to the original instrument and amended where needed. Conceptual and experiential equivalence, culture, and sentence structure were kept in mind when reviewing. Consistent back translations resulted in no changes.
11. Harmonization of all new translations with each other and the source version.	Harmonization is the final step of the back translation, which detects and deals with the discrepancies in translation between the versions. This is important quality control before the cognitive debriefing.	The two back translations were used as guides to edit FTX where needed. This produced a prefinal version, FTY. Any items that proved difficult, where the EP could not reach consensus on wording for conceptual equivalence, were scheduled to be discussed with the instrument developer.

3.4 – Cognitive debriefing

The patient perspective is crucial for providing insight into respondents' perception of the questionnaire. Wild et al. (2005, p. 97) suggests a step following the translation, called 'Cognitive Debriefing', which is described as 'testing the instrument on a small group of relevant patients or lay people to test alternative wording and to check understandability, interpretation, and cultural relevance of the translation'. The debriefing was conducted through telephone interviews, following

an interview guide provided by the Centre on Patient-Reported Outcomes Data at Haukeland University Hospital (see appendix 7). This step initiated the discussion of transferability and applicability in the population of CGS-users.

Table 4: Step 4 - Cognitive debriefing and finalization as proposed by Wild et al. (2005)

Critical components	Rationale	Execution
12. Preparations before recruiting cognitive interviewees.	To ensure that the criteria for recruiting are understood, and that all counsellors use the same criteria. To provide patients with the framework and tools for reading and evaluating the questionnaire.	Written information (see Appendices 7 and 8) was provided by the MSc candidate, describing: 1. [3] Aims of the project and inclusion criteria: *Aged 18 years, *Norwegian first language speakers, *Physically attending genetic counselling. 2. [4] An introduction to the scope of the project and a brief description of what participation entails. 3. The interview guide was included together with the prefinal version FTY.
13. Recruiting patients from the target population to the PRG.	To involve patients when testing how the questionnaire is perceived by respondents.	Counsellors followed the description given by the MSc candidate and recruited 12 patients between 16.12.21 and 20.01.22.
14. Cognitive debriefing of the prefinal Norwegian version, with patients drawn from the target population.	To provide an insight into how the items are perceived in the target language and to strengthen experiential equivalence. To capture possible discrepancies between source and target language, as well as improve wording. Useful for testing items that the EP did not reach a unanimous decision for.	Patients were called at the scheduled times for the interview to be conducted. Patients were first asked about what condition was relevant for them. Further, the MSc candidate went through each item together with the patient, making notes of their feedback. Uncertain items and words were asked about specifically, to avoid missing any valuable insight. The conversations were not recorded.
15. Cognitive debriefing results are reviewed.	To ensure semantic equivalence between the original and translated instrument. This step also filters the PRG feedback and	The results were first discussed in a meeting in the research team, consisting of MSc candidate and supervisors, with the intention of filtering the most crucial

[3] For counsellors
[4] For patients

	makes sure that no changes compromise the conceptual equivalence between the original English instrument and the translated Norwegian product.	information from the feedback. The information was then organized in Table 5 before a meeting with the instrument developer Marion McAllister was scheduled.
16. Meeting with the instrument developer for finalization of the translation.	To further ensure conceptual equivalence between translations. This step will fill in gaps where this project's team could not draw a conclusion.	Instrument developer was contacted to clarify original intent, where both the translators and the EP failed in producing a satisfying translation. An online meeting was scheduled between the research team and instrument developer. In the meeting, problems were discussed and resolved with McAllister providing the remaining insight needed.

3.5 – Ethical considerations

It is necessary to clarify any research ethical considerations within the project. The privacy representative at Haukeland University Hospital was first consulted about the potential need for an application to the Regional Committee for Medical Research Ethics (REK) or the Norwegian Centre for Research Data (NSD). The information needed about the participants in this project did not include any identifiable or sensitive data. Points of interest were the patients' perspective on the wording in the translation, and they were not expected to fill out the questionnaire. The patients were informed of what participation entails by their genetic counsellor (Appendix 8). After the information was given, they were asked about consent for volunteering for the project. The volunteers were allowed to deny participation at any point in time. For contact with the patients, the project made use of the hospital journal system DIPS for accessing their phone number. This decision was made to avoid storing any personal, or identifiable information on the patients outside of the protected hospital system. It was confirmed that the project indeed does not compromise ethical considerations, such as the privacy or safety of the patients.

4. Results

This section utilizes the guidelines proposed by Beaton et al. (2000), for full transparency in the reporting of decisions made by the EP for achieving equivalence between the source and target instrument. The authors propose four areas to be focused through the EP meetings; semantic, idiomatic, conceptual, and experiential equivalence, as explained in the methods section, and will be reported in the current section.

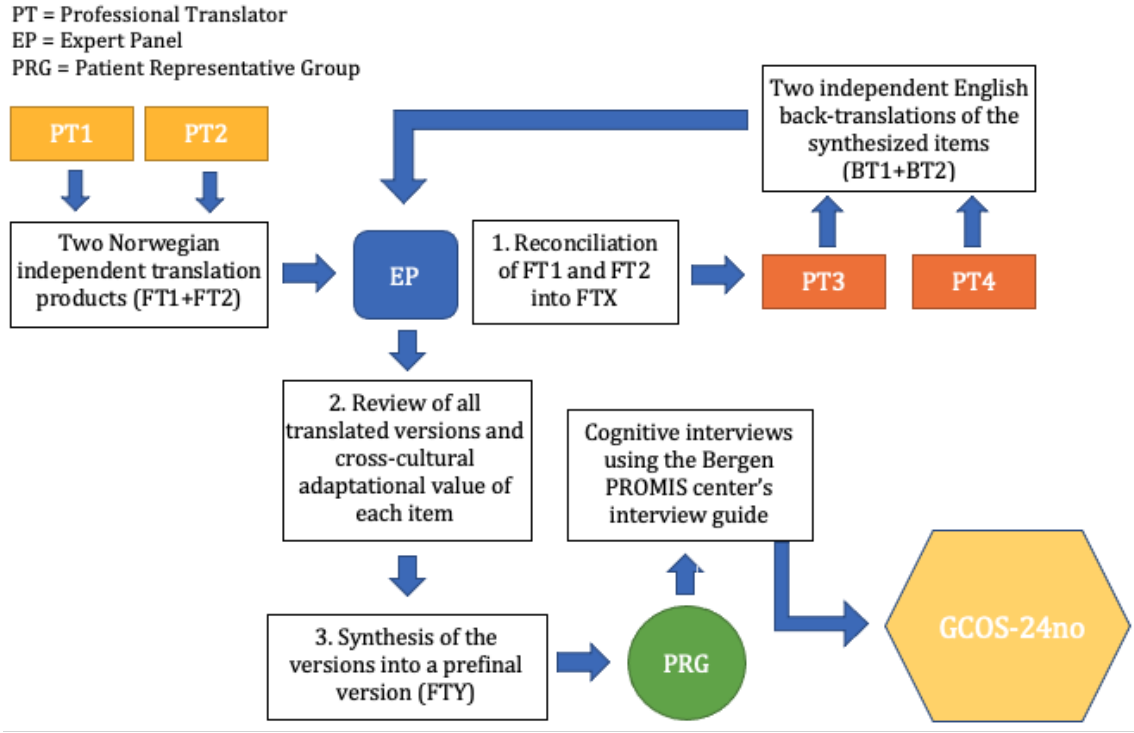

Figure 4 – Extended translation process in the present study, with version names included. Adapted from Wild et al. (2005) and Beaton et al. (2000).

4.1 - Forward translation results

The two forward translations FT1 and FT2 are included in appendix 1 and 2. Items that forward translated particularly well (needed little discussion by the EP) were: 1, 2, 3, 4, 5, 6, 10, 11, 12, 13, 16, 17, 19, 21 and 24. The results of the EP discussion of the remaining items are presented in table 5.

Table 5: Issues discovered and amended in step 2 - forward translation and reconciliation.

Item	Issues	Solution
7. 'I can control how this condition affects my family.'	Two different words for 'control' were used in the two different translations: 'Styre' (FT1) and 'kontrollere' (FT2).	The EP decided to not use either FT1/2 ('kan kontrollere'/'kan styre'). To be able to control, and to control are two different intentions. A literal translation would not capture the experience of control compared to the original item. Taking the patient perspective, the EP hypothesized that both items suggested by the translators would create some confusion in patients. An alternate phrasing; 'jeg har kontroll' was deemed the best suggestion.
8. 'I feel positive about the future.'	FT1 and FT2 produced two quite different item wordings, which needed to be discussed thoroughly.	Alternative phrases were discussed by the EP, and FT1 was selected to better fit the general Norwegian language. 'Jeg ser lyst på fremtiden' is a more common way of saying that you feel positive about your future.
9. 'I am able to cope with having this condition in my family.'	Neither FT1 nor FT2 provided a suitable translation of 'cope'.	Lacking a better suggestion for the translation, the EP deemed FT2 sufficient, in hopes that the back translation of this item would perform satisfactorily.
14. 'I understand the reasons why my doctor referred me to the clinical genetics service.'	'My doctor', used by both FT1 and FT2 does not include the full scope of how Norwegians are referred to CGS.	Wording was changed after forward-translation due to organizational differences, to ensure experiential equivalence. In Norway, it is not only your general practitioner or hospital specialist that refers you to genetic counselling. You can also refer yourself if your family is already assigned a pedigree ID number, or a relative has tested positive for a genetic variant. 'Doctor' was removed to increase relevance for a broader group of patients. It was replaced with a more neutral statement asking if the patient understands why they have been referred.
15. 'I know how to get the non-medical help I / my family needs (i.e., educational, financial, social support).'	'Non-medical help' was translated literally in both FT1/2, but was deemed unclear, and caused discussion in the EP. 'Educational' was not satisfactorily translated by either FT1/2.	'Ikke-medisinsk' was changed to 'andre former for hjelp', which is better understood together with the examples. This was supported by the Danish translation. The Norwegian word 'utdanning' refers to a degree or education. The word had to be changed to 'undervisning/kurs' which better represents the realistic non-medical help available to the patients.

18. 'I don't know who else in my family might be at risk for this condition.'	Conceptually inequivalent translations due to lack of contextual awareness from translators.	The EP suggested a third translation that brought the context into the translation. One cannot 'get' a genetic condition (FT2: '…utsatt for å få denne tilstanden.') which was a misunderstanding by the translators caused by context-unawareness. FT1 used the word 'disponert' which is not a word commonly used in genetic counselling and was expected to cause confusion in patients.
20. 'I am able to make plans for the future'	Both FT1/2 translated 'able' to 'kan', which was deemed unsatisfactory.	The EP changed 'Kan' to 'I stand til'. These terms are qualitatively different from each other. 'I stand til' is the correct translation.
22. 'I am powerless to do anything about this condition in my family.'	No translation proved sufficient for the word 'powerless'.	The EP discussed different phrasings, including 'maktesløs', but decided to keep the suggested item from FT2 due to similarities with the Danish translation.
23. 'I understand what concerns brought me to the clinical genetics service.'	The two translations translated 'concerns' to either 'bekymringer' or 'vurderinger'.	The EP decided that FT1's 'vurderinger' was correct, after discussing how 'concerns' should be interpreted in terms of semantic equivalence.

The EP discussions, as reported in table 5, produced one reconciled version, named FTX, that can be found in appendix 3. FTX was further sent to the translation company for back translation.

4.2 - Back translation results

Reviewing the back translations (BT1 and BT2, see appendix 4 and 5) revealed some discrepancies between how the Norwegian version and the original measure was worded. The back translations were mostly very consistent and proved high conceptual and semantic equivalence to the original items. Items that back translated particularly well (where at least one back translation was nearly identical to the original item) were: 1, 5, 6, 10, 11, 12, 13, 14, 15, 17, 19, 20, 21 and 24. Note that the issues with items 14, 15 and 20 in the forward translation were resolved by the first EP discussions and proven by a satisfactory back translation. The results of the EP discussion of the remaining items are presented in table 6.

Table 6: Issues discovered and amended in step 3 – back translation and harmonization.

Item	Issues	Solution
2. 'I can explain what this condition means to people in my family who may need to know.'	Both BT1/2 translated 'innebærer' to 'entails', despite the FT1/2 had both translated 'means' to 'innebærer'.	The EP decided that, despite the back translation issues, 'innebærer' is a better word than 'betyr'. This is mainly due to which words are being used in the context of genetic counselling. Since the forward-translations were satisfactory, it was concluded that no meaning was lost in back translation. The item in FTX was therefore kept as-is.
3. 'I understand the impact of the condition on my child(ren)/any child I may have.'	'Konsekvenser' was back translated to consequences instead of impact.	The EP decided to change the word from 'konsekvenser' to 'betydning'.
4. 'When I think about the condition in my family, I get upset.'	Both BT1 and BT2 produced statements that starts with 'I get upset' which can be read differently from the original item with a different order of clauses.	The EP decided the best way to solve the issue was to change the order of the clauses. 'Blir jeg bekymret' was placed in the end rather than the start of the sentence, contrary to the forward translation. Typically, in Norwegian grammar, this statement would start with 'I get upset', but the EP considered it to be qualitatively different from the original statement. To avoid the possibility of affecting the respondent while reading the item, the sentence needs to start with asking the patient to think of the condition in the family.
7. 'I can control how this condition affects my family.'	Neither BT1 nor BT2 produced items similar to the original item. This made the EP question their decision from the forward translation.	The EP could not reach a unanimous solution for this issue, as there was too much uncertainty around the intent of the question. The solution was to leave this item unresolved until final input from the instrument developer.
8. 'I feel positive about the future.'	BT1/2 produced two items dissimilar to the original instrument.	The item was kept as-is due to idiomatic equivalence. The problem seemed to be the translators not considering the added idiomatic value for Norwegian language when back translating.
9. 'I am able to cope with having this condition in my family.'	The item in FTX did not back translate well, with neither BT1 nor BT2 choosing the word "cope".	It was kept as-is, due to the lack of a better word for 'cope' in Norwegian. The item was highlighted as one to discuss in particular with the PRG.

16. 'I can explain what this condition means to people outside my family who may need to know (e.g., teachers, social workers).'	Both BT1/2 translated 'innebærer' to 'entails', despite the FT1/2 had both translated 'means' to 'innebærer'.	See item 2 for solution.
18. 'I don't know who else in my family might be at risk for this condition.'	Conceptually inequivalent translations. Lack of contextual awareness from translators.	The suggested item 18 in FTX back translated well in BT1 but was still influenced by the missing context in BT2. As it was a lack of contextual awareness that influenced the unsatisfactory translation, the EP still decided that it was the best translation, and kept it as phrased in FTX.
22. 'I am powerless to do anything about this condition in my family.'	By keeping the suggested item from FT2, the following back translations were not satisfying.	The EP decided the best word for powerless was 'maktesløs', contrary to suggestions by translators. The item was also marked as one to discuss in particular with the PRG.
23. 'I understand what concerns brought me to the clinical genetics service.'	Both BT1/2 translated 'vurderinger' to 'assessments', which led the EP to question the decision made in FTX.	Failing to resolve the issue in the EP discussions, this item was brought to the instrument developer for clarification of intention. The word 'concerns' can be understood as both 'worries' and 'reasons'.

The EP discussions as reported in table 6 produced a synthesized, prefinal version, FTY, that was used for the cognitive interviews (see appendix 6).

4.3 - Cognitive debriefing results

Cognitive debriefing through interviews (hereby called cognitive interviews) were performed to test the prefinal version FTY on a group of patient representatives (PRG). The PRG consisted of eight volunteers from a patient population in Department of Medical Genetics, Haukeland University Hospital that have attended at least one genetic counselling session. The focus was on the patient's thoughts and perceptions of wording and their understanding of the content of each item in the instrument. The interviews utilized an interview guide provided by the 'Centre on patient-reported outcome data' at Haukeland University Hospital and were carried out through telephone conversations with each participant. In total, 8 respondents answered their phone when called on 21.01.22

(4 patients) and 28.01.22 (4 patients). The duration of the calls ranged from 15-40 minutes, with the participants having received the questionnaire and interview guide in advance.

For presentation of the results from the cognitive interviews, three categories have been selected: relevance, wording, and emotional response. If items were addressed by more than one patient or had been highlighted as important from the EP discussions, they were included in table 7. Items that had few or no comments related to them were items: 1, 2, 3, 4, 8, 11, 12, 16, 17, 18, 19, 20 and 22. Note that the issues with items 4, 8, 16, 18 and 22 in the back translation were all resolved in this step, with the patient representatives understanding the items as intended. The results of the feedback from the PRG on the remaining items are presented in table 7, and in the following text.

General comments that fall outside the scope of categories include similarity between items. Some patients found that items felt repetitive and similar, creating the feeling of already having given a response. Items 13 and 24 are both statements regarding decisions. Items 1, 14 and 23 are all statements of understanding the reason for referral/need for counselling and were mentioned by two participants. Furthermore, negatively worded items created confusion for several people, with the suggestion to change them to positive wording. As items are usually reversed intentionally by the instrument developer to counter response bias, such a change could not be implemented because it would weaken the validity of the instrument.

Table 7: Cognitive debriefing results

	Relevance	**Wording**	**Emotional response**	**Solution**
Item 5	Two participants commented on low relevance due to predictive testing	'Medical help' was misunderstood by the patients that had no diagnosis.	x	The item was kept as-is, to better include a broad, heterogenous group of patients across situations.
Item 6	Three patients commented on not understanding the relevance of the item. They could not imagine any situation	x	Four patients were quite provoked by the item and reported that they felt offended. The item was considered unnecessary. A few of	The comment on "imposing expectations" was deemed the most important to address. As no item could be removed, a suggested solution was changing the introduction of the

	where the response would be agreeable.		them used the phrase 'imposing expectations'.	questionnaire to better clarify that there are no correct answers.
Item 7	One patient reported increased relevance if it had been personally addressed, instead of towards the patient's family.	'Control' created some confusion in two patients.	x	Based on the feedback from the two participants, this could not be resolved before the meeting with instrument developer.
Item 9	x	'Cope' did not easily translate to Norwegian (takle). Two patients wanted to replace the word.	x	The EP discussed how it could be rephrased. It was kept as-is, due to most patients understanding and expressing a preference for the word, confirming conceptual equivalence.
Item 10	Relevance was low for three predictively tested patients, all waiting for their test results.	'Options' created confusion in patients that were waiting for a test result.	'Gain' was emotionally problematic (one patient). Implications were feelings of imposing expectations of positive emotions in a difficult situation. One patient also reported a lack of options is more realistic than having options. This was concerning surgical removal of breast tissue.	The item was kept as-is, to better include a broad, heterogenous group of patients across situations.
Item 15	Low relevance for three patients. Most other patients understood the item, and correctly assumed this was more relevant for other conditions than their own.	'Other forms of help' (as opposed to non-medical help) was unclear to two of the patients and needed to be clarified.	x	The item was kept as-is to include a broader, heterogenous group of patients across situations. The research group decided that a potential solution was to change the questionnaire introduction, and to ask for permission from the instrument developer.

Item 21	x	x	The tone of this item was perceived as instilling guilt in two patents. They used adjectives such as 'triggering' and 'blaming' One patient suggested rewording this item to imply a more positive connotation (see solution).	- 'Jeg er klar over at jeg ikke er ansvarlig for mine egne gener og hvem jeg viderefører dem til.' The suggested wording would be qualitatively different to the original item, where guilt is not mentioned. The best suggestion was to clarify this in the introduction of the questionnaire, as would be discussed with the instrument developer later.

The PRG's interpretation of the prefinal version FTY was compared to the original version GCOS-24, to identify where improvements were needed. Discrepancies were amended, as this is where the gap between professionals in the field and the patient population was exposed. Words, phrases, or items that were unclear or unfamiliar were changed. This step was necessary to add additional, valuable insights that were overlooked by the expert panel, and will, in turn, improve performance of the questionnaire in the target population.

4.4 – Finalization and clarification with the instrument developer

The last step before finalizing the prefinal version was to involve the instrument developer, as reported in step 15 (table 4), to discuss the items reported in table 6 and 7. Marion McAllister met with the research team on March 2nd, 2022. The discussion gave substantial support for the changes the EP had made, as well as decisions about wording. Changes were considered necessary where the EP could explain their decisions either through cross-cultural adaptation or structural differences in healthcare systems between target and source countries. The EP struggled with translating the phrase 'I can control', as there was unclarity about whether it should encompass either or both the feeling of experiencing control and being able to control. Version FTY phrased the item as 'kan kontrollere' but was not very well received by the PRG, as they felt they 'could obviously not control anything'.

McAllister clarified that it was most important that the item contained the feeling of control. Changing the item to the alternate phrasing: 'har kontroll over', would suggest truly being able to control something, and would still not solve the issues brought up by the PRG. In keeping the item as-is, semantic equivalence was retained.

Regarding item 23, McAllister confirmed that the intended meaning of 'concerns' is that there was an issue that arrests the attention of a doctor, leading to appropriate action. 'Vurderinger' is different to 'concerns' in that it is not negatively or positively charged, but 'bekymringer' is even more different as it is meant a feeling of anxiety or worry without any action attached to its meaning. Based on this clarification, the research group decided to keep the Norwegian word 'vurderinger' as this better encompasses both that an underlying reason has been considered relevant, combined with an action. McAllister also gave permission to change the introduction to the questionnaire as a solution to both the emotional and relevance issues reported by the PRG in items 5, 6, 10, 15 and 21. The initial introduction in the prefinal version was a direct translation of the original English language GCOS-24, without any edits made by the EP. In the project's final product, GCOS-24no, the introduction was changed to:

> Du vil nå se noen utsagn som handler om hva du sitter igjen med etter den genetiske veiledningen du har deltatt på. For hvert spørsmål du svarer på, tenk på informasjonen du fikk av din genetiske veileder. Sett ring rundt tallet som passer best for hvor enig du er med påstandene. Vennligst ta stilling til alle påstandene så godt du kan. Det finnes ingen riktige svar, og det er forventet at du skal kunne være både helt enig og helt uenig. Hvis du tenker at en påstand **ikke er relevant** for din tilstand, eller deg i din situasjon, velg **alternativ 4** (verken enig eller uenig).

From this step, the final questionnaire has been through professional translation in several steps, two expert panel discussions, cross-cultural adaptation, as well as the final patient perspective and input from the instrument developer has been considered. From this process, we are left with a Norwegian PROM: GCOS-24no (figure 4), which is ready for psychometric testing for application in both research and service evaluations of CGS in Norway.

The Genetic Counselling Outcome Scale (GCOS-24no)

Du vil nå se noen utsagn som handler om hva du sitter igjen med etter den genetiske veiledningen du har deltatt på. For hvert spørsmål du svarer på, tenk på informasjonen du fikk av din genetiske veileder. Sett ring rundt tallet som passer best for hvor enig du er med påstandene. Vennligst ta stilling til alle påstandene så godt du kan. Det finnes ingen riktige svar, og det er forventet at du skal kunne være både helt enig og helt uenig. Hvis du tenker at en påstand **ikke er relevant** for din tilstand, eller deg i din situasjon, velg **alternativ 4** (verken enig eller uenig).

1 = helt uenig
2 = uenig
3 = litt uenig
4 = verken enig eller uenig
5 = litt enig
6 = enig
7 = helt enig

		helt uenig	uenig	litt uenig	verken enig eller uenig	litt enig	enig	helt enig
1	Jeg har klart for meg hvorfor jeg deltar på genetisk veiledning.	1	2	3	4	5	6	7
2	Jeg kan forklare hva tilstanden innebærer, for de i familien min som kan ha behov for å vite det.	1	2	3	4	5	6	7
3	Jeg forstår hvilken betydning denne tilstanden kan ha for mine barn / eventuelle fremtidige barn.	1	2	3	4	5	6	7
4	Når jeg tenker på tilstanden i min familie, blir jeg opprørt.	1	2	3	4	5	6	7
5	Jeg vet ikke hvor jeg skal henvende meg for å få den medisinske hjelpen jeg / min familie trenger.	1	2	3	4	5	6	7
6	Jeg kan se at det har kommet noe godt ut av å ha denne tilstanden i familien.	1	2	3	4	5	6	7
7	Jeg har kontroll over hvordan denne tilstanden påvirker min familie.	1	2	3	4	5	6	7
8	Jeg ser lyst på fremtiden.	1	2	3	4	5	6	7
9	Jeg kan takle å ha denne tilstanden i familien.	1	2	3	4	5	6	7
10	Jeg vet ikke hva som er å vinne på hver av valgmulighetene jeg har.	1	2	3	4	5	6	7
11	Det gjør meg engstelig å ha denne tilstanden i familien.	1	2	3	4	5	6	7
12	Jeg vet ikke om denne tilstanden kan ramme andre slektninger (søsken, tanter, onkler, søskenbarn).	1	2	3	4	5	6	7
13	Når det gjelder tilstanden i familien, kan ingenting jeg beslutter endre fremtiden for mine barn / eventuelle fremtidige barn.	1	2	3	4	5	6	7
14	Jeg forstår hvorfor jeg er henvist til genetisk veiledning.	1	2	3	4	5	6	7
15	Jeg vet hvordan jeg kan skaffe andre former for hjelp jeg / min familie trenger (f.eks. undervisning/kurs, økonomisk -og sosial støtte).	1	2	3	4	5	6	7
16	Jeg kan forklare hva tilstanden innebærer til personer utenfor familien min som kan ha behov for å vite det (f.eks. lærere, sosionomer).	1	2	3	4	5	6	7
17	Jeg vet ikke hva jeg kan gjøre for å endre hvordan denne tilstanden påvirker meg / mine barn på.	1	2	3	4	5	6	7
18	Jeg vet ikke hvem andre i familien min som kan ha risiko for denne tilstanden.	1	2	3	4	5	6	7
19	Jeg har håp om at barna mine kan se frem til et givende familieliv.	1	2	3	4	5	6	7
20	Jeg er i stand til å legge planer for fremtiden.	1	2	3	4	5	6	7
21	Jeg har skyldfølelse fordi jeg har / kan ha gitt tilstanden videre til mine barn.	1	2	3	4	5	6	7
22	Jeg står maktesløs overfor tilstanden i familien.	1	2	3	4	5	6	7
23	Jeg forstår hvilke bekymringer som førte til min kontakt med den genetiske veiledningstjenesten.	1	2	3	4	5	6	7
24	Jeg kan ta beslutninger om denne tilstanden som kan forandre fremtiden for mine barn / eventuelle fremtidige barn.	1	2	3	4	5	6	7

Figure 5: The present study's final product: GCOS-24no

5. Discussion

The present study found that: (1) to measure the outcomes of genetic counselling, it is essential to have a valid tool that contains dimensions representative of the goals of the intervention; (2) translation of a PROM benefits from the use of specific guidelines when ensuring optimal performance in the target country; (3) such an adaptation has to involve representatives from the relevant patient group in addition to language and genetics professionals. This contributed to ensuring that the Norwegian version, GCOS-24no provides a scoring comparable to translations published worldwide. Through the process, the EP discussions proved valuable in detecting discrepancies, securing equivalence and preservation of the empowerment construct. In the forward translation, it became apparent that the Norwegian language has some limitations for equivalence through translation alone. For the EP to choose the optimal wording, the reference used was often the empowerment construct. Back-translation revealed some discrepancies in wording where the translations were not always adequate. The changes made by the EP in this step was focused on cultural adaptation and adding the context of CGS. Without such discussions with professionals, the items in the questionnaire would have lost contextual meaning.

The cognitive interviews served as valuable insight into how the translated questionnaire is perceived in the target population. Some issues were raised by the volunteers in the PRG, and made the basis for further discussion with instrument developer Marion McAllister. There were limitations to how comprehensively the research group could change the instrument based on PRG feedback alone, so compromises had to be made. Changing the introduction to the questionnaire, as approved by the instrument developer, served as such a compromise, hopefully solving the issues raised by the PRG, without compromising the psychometric properties. Considering the modifications done between the prefinal version FTY and the final GCOS-24no, the psychometric properties of the instrument could have been negatively affected without the patient perspective and McAllister's viewpoint. Following is a summary of the items that created issues and in which steps of the process they were resolved. Further, sections 5.1 – 5.4 will discuss the reasoning behind the solutions to the issues and provide the context and basis for such

resolutions. After disclosing and clarifying potential issues, edits were made and the new version was formed: GCOS-24no.

Items 14, 15, 20 created issues in the forward translation but were resolved through EP discussion, and back translated well. Items 8, 18, 22 and 23 were problematic in both forward and back translation, and problems were amended only when confirmed and understood by the PRG. Items 2, 3, 4 were not perceived as problematic in the first EP discussion but raised questions after back translation proved insufficient. The changes made by the EP to these items proved feasible after cognitive interviews. During the cognitive interviews with patient representatives, they were instructed to critically evaluate and share their understanding of the items in questionnaire version FTY (appendix 6). Some minor, but important issues were raised, and formed the basis for further discussion with the instrument developer. Item 15 was once again brought up as an issue in the cognitive interviews and was only resolved after discussing the change of the questionnaire introduction with McAllister. This served as a solution to the patient representatives' issues with items 5, 6, 10, 15 and 21. Items 7 and 9 were problematic in all steps of the process and was resolved only in the final version GCOS-24no after discussion with Marion McAllister.

5.1 – Preservation of the Empowerment construct through translation

Before psychometric testing, the discussion of validity of the instrument will benefit from addressing the underlying score that is produced for each respondent of the questionnaire. Traditionally the validity of a questionnaire depends on content, construct and criterion validity (Boparai et al., 2018). Empowerment, the construct overarching the five dimensions in both GCOS-24 and GOS, captures the potential outcomes of CGS. For the questionnaire to maintain its construct validity through translation, it was important that the EP successfully preserved this construct, carrying it over to GCOS-24no. The genetic counselling definition from Fraser (1974) is comparable to the empowerment construct defined by McAllister, Dunn, et al. (2011), as they contain similar outcomes. They also correlate highly with how the Norwegian genetic counselling practice values outcome from care. Norwegian healthcare is regulated by, among other, the Biotechnology Act (2003) and the Patient's Rights Act (1999). In general, patient-centred care is highly valued in the healthcare system, which is reflected in the legislation. This is especially relevant in

genetic counselling, where every condition and individual has varying needs, and the information, care and follow-up should be tailored accordingly. The patients should be involved in all steps of decisions about their own health and treatments. This is only possible once sufficient information and support has been given, leaving the patient increasingly empowered.

The first goal towards empowerment is to give the patient the sufficient knowledge and understanding about the condition in question. Informed decision making is a main goal in genetic counselling. Stated in the Patient's Rights Act (1999, § 3-2), the patient's right to information is highly regarded. It is important that the patient receives necessary information for full insight into their condition and content of the health care. It is not enough just providing such information; the genetic counsellor is legally obligated to considerately adapt the information to the patient's needs. The Patient's Rights Act (1999, § 3-5) regards the form of information, where the information given should be provided as accurately, and clearly as possible. The patient should be left with a realistic impression of the weight of importance and content of the information given. Providing such information in a sufficient way may increase knowledge and leaves the patient feeling like they have gained cognitive control. This may further empower the patients to make decisions about major life events (McAllister & Dearing, 2015).

Such major life events may be, whether the patient should take on an extra loan, move to another country, expand their family, re-educate, etc. For the patient to be able to decide about the options available, all relevant information that may influence the decision needs to be provided to the patient. Thus, decisional control regards not only the decision to undergo genetic testing, but also important life decisions that can be affected by health status and the condition in question (McAllister & Dearing, 2015). This aligns with the goals of genetic counselling, to enable the patient to make such decisions. The behavioural control dimension is meant to consequently make the patients feel like they can take appropriate action and become active participants in managing the family condition. Such appropriate action and the patient's role are values preserved in the Patient's Rights Act (1999, § 3-1) regarding the patient's right to participate in the implementation of their health care. These goals are affiliated; once the patient has decisional control, they are better equipped to navigate their own treatment plan. Like decisional control, the patients

need to be informed about their options in order to experience behavioural control. The patient should have gained such an ability after participating in genetic counselling. It is crucial that the patient can utilize this new information as a tool for navigating the different options available, and make an informed choice based on the facts available.

As a final consideration, the dimensions of hope for the future and emotional regulation will differ depending upon both the characteristics of the patient and the nature of the content in the counselling session. Every individual applies their own cognition to the information given (Forgas, 2017). If the patient is already in a negative affective state, the information might be considered threatening. The baseline affective state impacts the individual's ability to emotionally regulate and may consequently decrease hopefulness. Counsellors provide context to patients in genetic counselling so that they are better equipped to achieve emotional control and hope for the future (McAllister & Dearing, 2015). By giving the patient positive, future-oriented information, the counsellor can direct the patient's feeling of the future towards hope. Such information can be about opportunities to participate in ongoing research about their family's condition, penetrance and variability, patient support groups, and promises of updates on the expanding knowledge and new research on their family's condition. When such information is provided in counselling, patients feel they can cope better with their circumstances, which gives them hope. Receiving not only information, but support from the genetic counsellor and post-test support systems is useful for emotional regulation (McAllister et al., 2008).

Considering the similarities between the goals of genetic counselling as stated in Fraser (1974), the Norwegian regulations and the empowerment construct, the GCOS-24 is a highly accurate measure for patient-reported outcomes of genetic counselling. The GCOS-24no can give an indication of how well the Norwegian practice attains its goal, facilitating informed decision making in patients, leaving them hopeful and capable of coping with their condition both in the present moment and for the future. After all, the services provided will solely be as good as the reported outcomes from patients, following the intervention.

5.2 – Cross-cultural adaptation

There are differences between the NHS, Britain's Healthcare system in which GCOS-24 is validated, and the Norwegian healthcare system where GCOS-24no is to be applied. Considering these differences, some cross-cultural adaptations needed to be made in addition to linguistic. A benefit of the adaptation method is that it captures these subtle differences that the translation alone might not be capable of addressing. Lee et al. (2019, p. 1) suggest that 'translation alone is insufficient to narrow the subtle gaps caused by differences in culture and linguistic style'. It is crucial to also consider 'adaptation', which addresses the translatability of the questionnaire. It is defined as 'the extent to which a questionnaire can be meaningfully translated to achieve equivalence to the source text, and yet remains culturally and linguistically appropriate in the target country' (Conway et al. (2014) as cited in Lee et al. (2019, p. 5). In addition to qualitative and structural differences between countries and their respective healthcare systems, McAllister, Dunn, et al. (2011, p. 129) states that 'empowerment is likely to be influenced by culture'. Cross-cultural differences can provide explanations for surprising results, which was seen especially for results from the cognitive interviews.

Such differences identified in the cognitive interviews were mostly regarding the positive and negative valence of the questions belonging to the 'hope for the future' and 'emotional regulation' dimensions. Several respondents reported feeling that these statements placed expectations on how they were supposed to feel, as noted in table 7, items 6, 10 and 21 under emotional response. This feedback was especially present when there was an incongruence in the emotional valence of the patient and the item. With questions addressing emotions with a positive valence, such as hope and positivity, the respondents that were initially not feeling hopeful or positive were quite offended to be asked such questions. Correspondingly, when the respondents were expressing hopefulness and positivity, they were offended when responding to negatively focused items, addressing emotions such as guilt and anxiety. It appears the general perception of the patient representatives was that they felt uncomfortable when not giving the 'correct' answer. The phenomenon, 'social desirability-bias' is defined in APA dictionary as 'the bias or tendency of individuals to present themselves in a manner that will be viewed favourably by

others' (American Psychological Association n.d.). This desire can manifest in dishonesty in response trends for self-reported questionnaires.

Although the PRG did not actually respond to the questionnaire, they indicated they would find it difficult to do so. These uncomfortable feelings may have arisen when they considered their honest reply to be different than what they perceived to be socially acceptable. Such a response trend is a confounder that can reduce the validity of the instrument. This phenomenon is present in most populations, as people in general aim to please either the interviewer, themselves, society, or the in-group they identify with. Social desirability as an issue in research that uses self-reported measures has been reported and explored by many authors (Badejo et al., 2022; Bergen & Labonté, 2020; Bou Malham & Saucier, 2016; van de Mortel, 2008). This phenomenon is not culturally delimited nor specific for a Norwegian sample. However, it is interesting to discuss why this was present in our study and not reported as an issue in the sample used for developing the original instrument (McAllister, Wood, et al., 2011). The PRG showed a trend in regarding the most agreeable answer the correct one. This trend can be somewhat explained by cross-cultural differences. Norwegians, when compared to 30 other countries worldwide, score the third highest on agreeableness in the Big Five personality test, ranking only below Sweden and Flemish Belgium (Bartram, 2013). Agreeableness entails cooperation in addition to other sub-traits, which is unsurprising to find in the Norwegian population. Since the items are worded as statements, and not questions with a yes or no answer, this may distort perception. It may manufacture the reported feeling of imposing expectations. Even though this was not intended by the developers, the conclusion was drawn by the patient representatives. This may be indicative of the notion that the participants' perception of the items, can be attributed to cultural differences.

This issue was further resolved through changing the introduction of the GCOS-24no. In the final version, it was emphasized that there are no correct answers, and it is expected that one can both agree and disagree. This was also reported by Diness et al. (2017) where, in the interviews, they had to emphasize that there were no correct and incorrect perceptions. This was in response to the participants' concerns about how they were perceived and whether they had made a mistake. However, they do not report how they ended up dealing with the issue. Keeping the

aspect of cultural differences between the source and target country in mind while choosing preferred wording for the items, contributes to strengthening relevance for the target population. The target population in this study are CGS-users, represented by the participants of the PRG. The results from the cognitive interviews were very satisfying in terms of cultural understanding and relevance. Although there were comments on the relevance of some items, these were mostly condition specific as opposed to culturally specific. Keeping these items as they were intended, even though not relevant for all patients, will likely increase relevance for a broader, more heterogenous group of CGS-users, as this measure is not targeted toward specific genetic conditions. The characteristics of the study-sample where McAllister, Wood, et al. (2011) developed the GCOS-24, represents a broad range of genetic conditions, as should the sample we choose for future psychometric evaluation.

5.3 – Cognitive interviews and input from instrument developer

As Diness et al. (2017) discussed in their study, investigating how the patient representatives understand specific sentences is difficult. A phone interview as used in the present study, will only provide a crude impression of the participants' perception of complex questions (Diness et al., 2017). In the cognitive interviews in the present study, participants were asked specifically about complex words and items that created discussion in the EP meetings, and how they were perceived. This prevents overlooking valuable information and gives the participants the opportunity to either confirm or deny their understanding of the item as intended. Asking specifically about such items did initiate reflection from the participants about items they had not yet reflected upon. In addition to not overlooking valuable information, it was important not to overestimate the importance of feedback. To reflect the trends in responses, issues with items were only included in table 7 when they were raised by two or more respondents. This was to avoid focusing too heavily on the feedback of a single respondent. Most participants did not prepare for the interview, and it may be reasonable to question whether they felt obligated to give criticism on the spot. Whenever they were asked to elaborate on their comments, they had trouble explaining why they expressed low preference for wording, and few had suggestions for changing the items. The patients in our sample were few and were interviewed after only their first counselling session. It is possible that many

patients who underwent predictive testing were either anxious to know their results, or avoided such thoughts, which may have resulted in a less prepared interview.

The level of preparation varied between the participants, with only one having read the questionnaire and taken notes prior to the interview. This participant had nothing negative to report as she had discussed the translation with her bilingual (English-speaking) daughters and had even found and compared the original GCOS-24 to the prefinal version of the Norwegian translation. Overall, the PRG reported few difficulties with both understanding and interpreting the items in the cognitive interviews. This is an indication of a high-quality translation and adaptation, where the conceptual meaning of the original items was retained. The feedback from the cognitive interviews were mostly preference based, which is valuable information, but not enough to make changes that would compromise Beaton's equivalences. There were limited options for changing the items drastically, as the intention was not to improve the questionnaire, but translate and adapt the items. The EP decided at the start of the present project to prioritize retainment of the psychometric values and the empowerment construct. The main reasoning for this is to ensure the GCOS-24no's feasibility of comparing results across studies from different countries using the GCOS-24. The PRG in the present study suggested changes to the questionnaire that would alter the items to a degree where it would no longer be comparable to the original measure. It was decided to make no such edits that would compromise the original intention of the items. Most decisions to change the questionnaire after this step were based on relevance. Some significant cross-cultural differences were identified in the cognitive interviews, as presented in section 5.2. Additional findings from the cognitive interviews will be addressed in this section.

The PRG had some issues with item 6 (I can see that good things have come from having this condition in my family). Most participants could not think of examples of 'good things' until the interviewer gave examples. Payne et al. (2008) had similar findings, where their solution was to add examples of 'good things' to the item: '(e.g., early illness detection and personalized screening)'. The authors also added examples to item 10 (I don't know what could be gained from each of the options available to me). This could have served as a solution to the PRG's comments on the items where examples had to be supplied during the cognitive interviews. In

the end, the EP decided not to make such changes to the questionnaire. The reasoning is that patients should themselves reflect on such examples, without the influence of what's objectively regarded as good things. If the respondent cannot think of examples for item 6, that is a clear indication that the appropriate answer is 'strongly disagree'. Providing such examples can be the source of bias in questionnaire design. One must assume that adding anything to the items will potentially change the original meaning intended, and furthermore influence the respondents' perception. One category of bias that originates in question design, identified by Choi and Pak (2005) is leading questions. Such questions, where providing examples, may cause the respondents' to only focus on the examples provided, and will lead their response in a certain direction. This gives support to the EP's decision not to add examples, as this would potentially have contributed to response-bias, thus also compromising the validity of the questionnaire.

The most significant adjustment done to the questionnaire was changing the introduction. This was a solution to meet the challenges reported by the PRG in table 7, except for item 7 (I can control how this condition affects my family) and 9 (I am able to cope with having this condition in my family). Many comments regarded relevance, which should not be an issue considering alternative 4, which can be selected as a neutral response to irrelevant items. This is also stated explicitly in the introduction to the prefinal version FTY. A reasonable explanation could be that the PRG have overlooked the instructions for responding to the questionnaire. This could in turn affect response rates for single items, as well as promote fatigue in respondents. Since no items could be removed during translation and adaptation, the research group had to find an alternative solution to increase relevance. Removing the items would weaken the measure's application value. It would prevent the possibility of comparing results with studies from different countries that use the GCOS-24. Additionally, it would also decrease inclusivity of patients to whom the items are relevant. During the cognitive interviews, the patients were asked when mentioning relevance, if they felt comfortable choosing alternative 4 where they found the item irrelevant. Some participants considered this option as new information, and some said they would prefer if the item was not present. Bolding the words 'not relevant' and 'alternative 4' may increase the attention from the respondents to this information, hopefully leading to less patients overlooking this

option. This is a compromise in an effort to resolve the issues in a way that includes every potential patient.

Palmer et al. (2018) found in their study that GCOS-24 may not fully capture aspects of patients living with undiagnosed conditions, and that it fell short of the average in clinical importance when dealing with undiagnosed diseases. These findings agree with what the present study found in the cognitive interviews. Those patients that commented on the relevance of items 5[5] and 10[6] in the FTY were all predictively tested with no test results available at the time of the interview, meaning they were living with undiagnosed conditions or unsure test results. Additionally, the word 'condition' caused confusion among patients whose condition was still undisclosed. Item 5 refers to the medical help needed, which will be affected by the carrier status of the patient. If the patient's genetic test is negative, they might not need medical help for themselves. Furthermore, if they have an unclear phenotype that requires more general testing, they might not know what medical help can be offered yet. Item 10 focuses on the benefits of the options available to patients. If the counsellor is aware of what genetic condition is likely for the patient, they should be informed of what options are available for post-test support (support-groups, screening program, annual specialist checks).

After the first counselling session, the options and medical help available may be less apparent to the patient and the counsellor than it would be once more information has been gathered. When asked about items 5 and 10 after the first counselling session, they might find it challenging to answer, and consequently evaluate the items' relevance as low. The results from the cognitive interviews in the present study corroborate the findings of Palmer et al. (2018), assuming other factors did not significantly influence the patient's evaluation of relevance for the items. The solution to this problem was again adjusting the introduction of the questionnaire. It was specified that when the item is irrelevant to 'your condition, or you in your current situation', please choose alternative 4. This encompasses both patients with conditions like in item 15 (I know how to get the non-medical help I/my family need(s)), where there is no applicable non-medical help available, as well as

[5] I don't know where to get the medical help I/my family need(s)
[6] I don't know what could be gained from each of the options available to me

undiagnosed patients that may struggle with items requiring them to think about a specific condition.

After performing the cognitive interviews, the research group had substantial support for issues to be brought to the instrument developer. Initially the method proposes a meeting with the instrument developer for clarification during review of the back translation (step 10) before producing the prefinal version of the translated questionnaire. The research group decided to postpone this step until after the cognitive interviews, as this is where all potential issues and questions have become transparent. Through the meeting, McAllister asked specifically about the term 'condition', which was highlighted as hard to translate into several other languages. The term was unproblematic in the present project, both during translation and the cognitive interviews, with patients understanding the word 'tilstand' as intended. The Norwegian language word 'tilstand' inhabits semantic equivalence to the original language term 'condition'. It was understood by all patients as a genetic condition and was no source of confusion in the present study. This is different from the findings of Diness et al. (2017) and Muñoz-Cabello et al. (2018) where they reported issues with finding a suitable translation for the word. In the end, the terms selected were perceived as intended by the target population in their respective projects. Voorwinden et al. (2019) did not perform cognitive interviews. Their study dealt with statistical analyses and testing the psychometric properties of the questionnaire after translation, and no such issues were reported. The meeting with McAllister resulted in no further major changes, other than the ones reported in the previous sections.

5.4 – Strengths and limitations of the present study

As disclosed in the methods section, some changes were made to the originally proposed methods. Translators were initially excluded from the EP for convenience and instructions from stakeholders yet ended up strengthening the quality of the product and increasing transparency in each step of the process. By separating the translation and the adaptation, the two groups could focus entirely on their respective task, not compromising its translation quality to address adaptation, and vice versa. The back translation would also have been compromised if the translators were informed of the empowerment construct. 'The two translators

should neither be aware nor be informed of the concepts explored, and preferably without medical background' (Beaton et al., 2000, p. 3188). If we were to include the translators in the EP meetings, they would be highly context aware, which would be a source for information bias as well as increase the risk of eliciting unexpected meanings of the items in the translated instrument. Regarding the forward translation, the method proposes one context aware, and one unaware translator. It is unknown whether including one or both translators in the EP for the forward translation would have produced a different result. Regardless, translation and supplying context were performed in parallel, and addressed in every version of the translation. Keeping the translation and expert panel discussions strictly separate has provided a transparent process where every step is easily identifiable without overlapping.

An advantageous result from the project is that the translation and adaptation of the GCOS-24 results in two products with different potentials of application, ready for psychometric testing. The Genomics Outcome Scale (GOS), developed by Grant et al. (2019) is a questionnaire in short form, based on the GCOS-24. Using qualitative analyses and item selection of the six most important items from GCOS-24 valued by patients, GOS also measures empowerment. While the GCOS-24no will be used for research and outcome evaluations of genetic counselling in the out-patient clinic, GOS applies as a more general, less burdensome, and easily applicable instrument for use also outside the traditional counselling session. During the psychometric evaluation of the GCOS-24no, it is possible to extract the six GOS-items: 4, 16, 17, 18, 20 and 24, and run separate statistical tests for these translated items. The only change that needs to be done is rephrasing items 17 and 18 into agreeable statements. The MSc candidate will be involved in these future projects with writing and publication of the articles that report results from these psychometric tests. The Department of Medical Genetics, Haukeland University Hospital have already been in contact with the research group currently translating the GCOS-24 to Swedish, as well as the authors that published the Danish translation. Interest has been expressed from all three countries for further Scandinavian cooperation on research projects utilizing the instrument.

The strength of the present study lies also within the strength of the methods. This project remains anchored in an approach using two translation methods,

following guidelines proposed by recognized authors (Beaton et al., 2000; Wild et al., 2005) that have been used in several translations of questionnaires (Bing-Jonsson et al., 2018; Diness et al., 2017; Hagell et al., 2010; Lee et al., 2019). The present study produced a product that through translation and adaptation harmonizes with the product from Diness et al. (2017) in Danish, a country and language culturally similar to Norway. From the results of their studies and ours combined, it can be concluded that the current methods are sufficient both in answering the aims of the present study and produce reliable results that are comparable to other studies. Utilizing not only a translation method, but also a method for adaptation, the present study ensured that the GCOS-24no would stand the test of culture in addition to language when implemented in Norwegian CGS. Both GCOS-24 and GOS have been assessed for sensitivity to change and construct validity (McAllister, Wood, et al., 2011; Ting et al., 2021).

Further, the present study has some limitations at this stage of the questionnaire, which may be exposed when psychometric testing is performed. The patients in our PRG sample were few and were interviewed only after their first counselling session. This is not representative for how the questionnaire will be used in CGS, as patients will be measured in all stages of the counselling process. Including more patients would strengthen the ground for making changes to the instrument, if more of the participants gave similar feedback. Having a broader sample, including patients at different stages of the genetic counselling process, could have contributed to a better insight into how the questionnaire could have been adapted. Our sample consisted mostly of predictively tested patients, where the majority were cancer related. It could have been interesting to see how the items with feedback on the relevance would have been perceived from patients being counselled for even more complicated conditions. There are conditions more severe, that require the patient to receive more information on services and support systems available. More information may result in more knowledge and a clearer perception of what certain items were referring to. However, these issues will reveal themselves when analysing responses from a bigger sample. It is important to note that psychometric testing should include a large, diverse, and representative sample. The reliability and validity of the GCOS-24no has not yet been established in a Norwegian sample, and only after

performing such statistical analyses, its strengths and weaknesses can be truly revealed.

6. Conclusion

Bringing back the aim of the present study, seen together with the results discussed in the previous section, it is reasonable to conclude that the aim has been answered successfully. The instrument of McAllister, Wood, et al. (2011): GCOS-24 has successfully been translated according to Wild et al. (2005) and Beaton et al. (2000)'s methods for translation and cross-cultural adaptation of patient-reported outcome measures. The translation was conducted by including relevant participants, acknowledging the issues raised by them, and resolving them by considering the significance of the measure's underlying purposes. By ensuring that the changes made did not negatively affect the measure, the goal of translation was achieved.

The measure had to maintain equivalence, preserve the empowerment construct, adapt to Norwegian language and culture, and retain psychometric properties through translation. Resolving these issues required the EP to think dynamically, sometimes trading one principle of translation in favour of another through constantly evaluating where attention should be focused. It was sometimes challenging to figure out in which step it would be most valuable to make certain changes, that in many cases would resolve one issue, but might elicit another. Hence, to have a hierarchy of importance for these principles was necessary. The hierarchy formed the foundation for the EP's decisions, which were based on how it was assumed the measure would perform the best in the target population. Any conclusions about the psychometric properties of GCOS-24no are not possible before the proper statistical analyses have been performed.

6.1 – Future research

The psychometric evaluation of the GCOS-24 will be the final proof of the performance of the product in the target population. After this, the practical and applicational value of the instrument can be explored. It would be interesting to try and replicate the findings of other authors, to further strengthen the evidence for the versatility of the GCOS-24. The psychometric tests performed on the British GCOS-24

concluded that it is a potentially useful PROM for evaluating outcomes of CGS (McAllister, Wood, et al., 2011). There was a similar, uneven distribution of patients with different genetic conditions across the various studies that have validated their translated instrument. Cancer-risks constituted 60.3% of the sample in the original GCOS-24 psychometric evaluation (McAllister, Wood, et al., 2011). Voorwinden et al. (2019) include a large, diverse, and representative sample in their validation. Their sample was being counselled for a broad range of genetic conditions, where 50% were cancer-risks. Diness et al. (2017) have similar group distribution between conditions, with 52,5% of the sample was counselled for oncogenetics.

Even though cancer groups were significantly bigger than other groups in all three studies, this may not be an unrealistic representation of the populations. This may also be the case for Norway CGS referrals. In Haukeland University hospital, 55% of all referrals to the out-patient clinic in 2021 were related to oncogenetics (Thorgrimsen-Stensvold, Ø., Administrative manager at the Department of Medical Genetics, personal communication, May 2022). McAllister, Wood, et al. (2011) disclaim that further testing is needed before it can be unreservedly recommended for routine evaluation of CGS. Such tests should focus on longer follow-up studies, with non-cancer as well as cancer genetics samples. These are all indications of how the GCOS-24no may be used in Norwegian genetic counselling. It could be interesting to look at outcomes from other conditions, not just cancer, as less information exists on outcomes from, eye conditions, for example, that only represented 3% of the sample.

6.2 – Implications for practice

There is undeniable, yet unexplored value in having the GCOS-24no, a validated PROM for use in Norwegian genetic counselling. A major strength is in the outcome of this study, where the final product of GCOS-24no provides a measure for outcome variables, that is the empowerment score for each respondent. Through this study, the Departments of Medical Genetics across Norway will have gained such an instrument. The discussions in the present project try to prove the feasibility of empowering the patient, and facilitating patient-centred care, using the GCOS-24no as an evaluation for these goals. These strong conceptual correlations give indication as to what the instrument can be used for. It can explore the contribution that genetic

counsellors make in empowering the patient, strengthening positive outcomes from the intervention. It can compare between individuals, conditions, departments and even countries, how these outcomes vary. The variations in outcomes can give an indication as to where service improvement is needed and save valuable time in making such assessments. This instrument is an important contribution to genetic counselling in Norway and puts the healthcare system one step closer to improving through patient-centred care.

7. References

American Psychological Association (n.d.). 'instrument', 'measure', 'dimension', 'social desirability'. In *APA Dictionary of Psychology*. Retrieved May 4th 2022, from https://dictionary.apa.org/

Badejo, M. A., Ramtin, S., Rossano, A., Ring, D., Koenig, K., & Crijns, T. J. (2022). Does Adjusting for Social Desirability Reduce Ceiling Effects and Increase Variation of Patient-Reported Experience Measures? *J Patient Exp, 9*, 23743735221079144. https://doi.org/10.1177/23743735221079144

Balint, E. (1969, May). The possibilities of patient-centered medicine. *J R Coll Gen Pract, 17*(82), 269-276.

Bartram, D. (2013). Scalar Equivalence of OPQ32:Big Five Profiles of 31 Countries. *Journal of Cross-Cultural Psychology, 44*(1), 61-83. https://doi.org/10.1177/0022022111430258

Beaton, D. E., Bombardier, C., Guillemin, F., & Ferraz, M. B. (2000, Dec 15). Guidelines for the process of cross-cultural adaptation of self-report measures. *Spine (Phila Pa 1976), 25*(24), 3186-3191. https://doi.org/10.1097/00007632-200012150-00014

Bergen, N., & Labonté, R. (2020, Apr). "Everything Is Perfect, and We Have No Problems": Detecting and Limiting Social Desirability Bias in Qualitative Research. *Qual Health Res, 30*(5), 783-792. https://doi.org/10.1177/1049732319889354

Bernhardt, B. A., Biesecker, B. B., & Mastromarino, C. L. (2000, Sep 18). Goals, benefits, and outcomes of genetic counseling: client and genetic counselor assessment. *Am J Med Genet, 94*(3), 189-197. https://doi.org/10.1002/1096-8628(20000918)94:3<189::aid-ajmg3>3.0.co;2-e

Bing-Jonsson, P. C., Slater, P., McCormack, B., & Fagerström, L. (2018, Jul 16). Norwegian translation, cultural adaption and testing of the Person-centred Practice Inventory - Staff (PCPI-S). *BMC Health Serv Res, 18*(1), 555. https://doi.org/10.1186/s12913-018-3374-5

Bjorvatn, C., Eide, G. E., Hanestad, B. R., Øyen, N., Havik, O. E., Carlsson, A., & Berglund, G. (2007). Risk Perception, Worry and Satisfaction Related to Genetic Counseling for Hereditary Cancer. *Journal of Genetic Counseling, 16*(2), 211-222. https://doi.org/https://doi.org/10.1007/s10897-006-9061-4

Boparai, J. K., Singh, S., & Kathuria, P. (2018). How to Design and Validate A Questionnaire: A Guide. *Curr Clin Pharmacol, 13*(4), 210-215. https://doi.org/10.2174/1574884713666180807151328

Bou Malham, P., & Saucier, G. (2016, Dec). The conceptual link between social desirability and cultural normativity. *Int J Psychol, 51*(6), 474-480. https://doi.org/10.1002/ijop.12261

Choi, B. C., & Pak, A. W. (2005, Jan). A catalog of biases in questionnaires. *Prev Chronic Dis, 2*(1), A13.

Costal Tirado, A., McDermott, A. M., Thomas, C., Ferrick, D., Harris, J., Edwards, A., & McAllister, M. (2017, Oct). Using Patient-Reported Outcome Measures for Quality Improvement in Clinical Genetics: an Exploratory Study. *J Genet Couns, 26*(5), 1017-1028. https://doi.org/10.1007/s10897-017-0079-6

D'Orazio, M. (2021). Distances with mixed type variables some modfied Gower's coeficcients. *Cornell University*, 1-17. https://doi.org/https://doi.org/10.48550/arXiv.2101.02481

Diness, B. R., Overbeck, G., Hjortshøj, T. D., Hammer, T. B., Timshel, S., Sørensen, E., & McAllister, M. (2017, Oct). Translation and Adaptation of the Genetic Counselling Outcome Scale (GCOS-24) for Use in Denmark. *J Genet Couns, 26*(5), 1080-1089. https://doi.org/10.1007/s10897-017-0086-7

Forgas, J. P. (2017). Mood Effects on Cognition: Affective Influences on the Content and Process of Information Processing and Behavior. In M. Jeon (Ed.), *Emtoions and Affect in Human Factors and Human-Computer Iteration* (pp. 89-122). Academic Press. https://doi.org/https://doi.org/10.1016/B978-0-12-801851-4.00003-3.

Fraser, F. C. (1974, Sep). Genetic counseling. *Am J Hum Genet, 26*(5), 636-659.

Grant, P. E., Pampaka, M., Payne, K., Clarke, A., & McAllister, M. (2019, May). Developing a short-form of the Genetic Counselling Outcome Scale: The Genomics Outcome Scale. *Eur J Med Genet, 62*(5), 324-334. https://doi.org/10.1016/j.ejmg.2018.11.015

Hagell, P., Hedin, P. J., Meads, D. M., Nyberg, L., & McKenna, S. P. (2010, Jun-Jul). Effects of method of translation of patient-reported health outcome questionnaires: a randomized study of the translation of the Rheumatoid Arthritis Quality of Life (RAQoL) Instrument for Sweden. *Value Health, 13*(4), 424-430. https://doi.org/10.1111/j.1524-4733.2009.00677.x

Inglis, A., Koehn, D., McGillivray, B., Stewart, S. E., & Austin, J. (2015, Mar). Evaluating a unique, specialist psychiatric genetic counseling clinic: uptake and impact. *Clin Genet, 87*(3), 218-224. https://doi.org/10.1111/cge.12415

Lee, W. L., Chinna, K., Lim Abdullah, K., & Zainal Abidin, I. (2019, Feb). The forward-backward and dual-panel translation methods are comparable in producing semantic equivalent versions of a heart quality of life questionnaire. *Int J Nurs Pract, 25*(1), e12715. https://doi.org/10.1111/ijn.12715

Macleod, R., Craufurd, D., & Booth, K. (2002, Mar). Patients' perceptions of what makes genetic counselling effective: an interpretative phenomenological analysis. *J Health Psychol, 7*(2), 145-156. https://doi.org/10.1177/1359105302007002454

McAllister, M. (2016). Genomics and society. In C. R. Kumar D. (Ed.), *London*. Elsevier Inc.

McAllister, M., & Dearing, A. (2015, Aug). Patient reported outcomes and patient empowerment in clinical genetics services. *Clin Genet, 88*(2), 114-121. https://doi.org/10.1111/cge.12520

McAllister, M., Dunn, G., & Todd, C. (2011, Feb). Empowerment: qualitative underpinning of a new clinical genetics-specific patient-reported outcome. *Eur J Hum Genet, 19*(2), 125-130. https://doi.org/10.1038/ejhg.2010.160

McAllister, M., Payne, K., Macleod, R., Nicholls, S., Dian, D., & Davies, L. (2008, Oct). Patient empowerment in clinical genetics services. *J Health Psychol, 13*(7), 895-905. https://doi.org/10.1177/1359105308095063

McAllister, M., Wood, A. M., Dunn, G., Shiloh, S., & Todd, C. (2011, May). The Genetic Counseling Outcome Scale: a new patient-reported outcome measure for clinical genetics services. *Clin Genet, 79*(5), 413-424. https://doi.org/10.1111/j.1399-0004.2011.01636.x

Muñoz-Cabello, P., García-Miñaúr, S., Espinel-Vallejo, M. E., Fernández-Franco, L., Stephens, A., Santos-Simarro, F., Lapunzina-Badía, P., & McAllister, M. (2018, Jun). Translation and Cross-Cultural Adaptation with Preliminary Validation of GCOS-24 for Use in Spain. *J Genet Couns, 27*(3), 732-743. https://doi.org/10.1007/s10897-017-0154-z

Palmer, C. G. S., McConkie-Rosell, A., Holm, I. A., LeBlanc, K., Sinsheimer, J. S., Briere, L. C., Dorrani, N., Herzog, M. R., Lincoln, S., Schoch, K., Spillmann, R. C., &

Brokamp, E. (2018, Sep). Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network. *J Genet Couns, 27*(5), 1087-1101. https://doi.org/10.1007/s10897-018-0228-6

Payne, K., Nicholls, S., McAllister, M., Macleod, R., Donnai, D., & Davies, L. M. (2008, May-Jun). Outcome measurement in clinical genetics services: a systematic review of validated measures. *Value Health, 11*(3), 497-508. https://doi.org/10.1111/j.1524-4733.2007.00259.x

Payne, K., Nicholls, S. G., McAllister, M., MacLeod, R., Ellis, I., Donnai, D., & Davies, L. M. (2007, Nov). Outcome measures for clinical genetics services: a comparison of genetics healthcare professionals and patients' views. *Health Policy, 84*(1), 112-122. https://doi.org/10.1016/j.healthpol.2007.03.005

Pedersen, T., Soini, S., Laufey, H. G., Stoll, J., & Herrmann, J. R. (2022). *Legislation on Biotechnology in the Nordic countries - An overview 2022* (Legislation on Biotechnology in the Nordic countries, Issue 9). http://urn.kb.se/resolve?urn=urn:nbn:se:norden:org:diva-12413

Santana, M. J., Manalili, K., Jolley, R. J., Zelinsky, S., Quan, H., & Lu, M. (2018, Apr). How to practice person-centred care: A conceptual framework. *Health Expect, 21*(2), 429-440. https://doi.org/10.1111/hex.12640

Segundo-Ribeiro, M., Bacalá, B. T., Alvarenga, W. A., Nascimento, L. C., McAllister, M., & Flória-Santos, M. (2020, Nov). Adaptation and preliminary validation of the genetic counseling outcome scale (GCOS-24) in a Brazilian genetic counseling setting. *Eur J Med Genet, 63*(11), 104018. https://doi.org/10.1016/j.ejmg.2020.104018

Siglen, E., Vetti, H. H., Lunde, A. B. F., Hatlebrekke, T. A., Strømsvik, N., Hamang, A., Hovland, S. T., Rettberg, J. W., Steen, V. M., & Bjorvatn, C. (2021, Oct 6). Ask Rosa - The making of a digital genetic conversation tool, a chatbot, about

hereditary breast and ovarian cancer. *Patient Educ Couns*. https://doi.org/10.1016/j.pec.2021.09.027

Skirton, H. (2001, Aug). The Client's Perspective of Genetic Counseling-A Grounded Theory Study. *J Genet Couns, 10*(4), 311-329. https://doi.org/10.1023/a:1016677110500

Slomp, C., Morris, E., Inglis, A., Lehman, A., & Austin, J. (2018, Apr). Patient outcomes of genetic counseling: Assessing the impact of different approaches to family history collection. *Clin Genet, 93*(4), 830-836. https://doi.org/10.1111/cge.13176

The Act Relating to the Application of Biotechnology in Medicine, (2003). https://lovdata.no/dokument/NL/lov/2003-12-05-100

The Act of 2 July 1999 No. 63 relating to Patients' Rights (1999). https://lovdata.no/dokument/NL/lov/1999-07-02-63/

Ting, M. S. O., Clarke, A., & McAllister, M. (2021, May 1). Assessing sensitivity to change of the genomics outcome scale (GOS). *J Genet Couns*. https://doi.org/10.1002/jgc4.1429

Tseng, E. K., & Hicks, L. K. (2016, Aug). Value Based Care and Patient-Centered Care: Divergent or Complementary? *Curr Hematol Malig Rep, 11*(4), 303-310. https://doi.org/10.1007/s11899-016-0333-2

van de Mortel, T. F. (2008). Faking It: Social Desirability Response Bias in Self-report Research. *The Australian Journal of Advanced Nursing, 25*(4), 40-48. https://search.informit.org/doi/10.3316/informit.210155003844269

Voorwinden, J. S., Plantinga, M., Krijnen, W., Ausems, M., Knoers, N., Velthuizen, M., Birnie, E., Lucassen, A. M., van Langen, I. M., & Ranchor, A. V. (2019, May). A validated PROM in genetic counselling: the psychometric properties of the

Dutch version of the Genetic Counselling Outcome Scale. *Eur J Hum Genet, 27*(5), 681-690. https://doi.org/10.1038/s41431-018-0318-9

Weldring, T., & Smith, S. M. (2013). Patient-Reported Outcomes (PROs) and Patient-Reported Outcome Measures (PROMs). *Health Serv Insights, 6*, 61-68. https://doi.org/10.4137/hsi.S11093

Wild, D., Grove, A., Martin, M., Eremenco, S., McElroy, S., Verjee-Lorenz, A., & Erikson, P. (2005, Mar-Apr). Principles of Good Practice for the Translation and Cultural Adaptation Process for Patient-Reported Outcomes (PRO) Measures: report of the ISPOR Task Force for Translation and Cultural Adaptation. *Value Health, 8*(2), 94-104. https://doi.org/10.1111/j.1524-4733.2005.04054.x

Yuen, J., Lee, S. Y., Courtney, E., Lim, J., Soh, H., Li, S. T., Chen, Y., McAllister, M., Fenwick, E. K., & Ngeow, J. (2020, Feb). Evaluating empowerment in genetic counseling using patient-reported outcomes. *Clin Genet, 97*(2), 246-256. https://doi.org/10.1111/cge.13646

8. Appendices

See separate attached documents.