Received: 23 August 2023 | Revised: 31 January 2024 | Accepted: 20 March 2024

DOI: 10.1002/jgc4.1896

National Society of Genetic Counselors WILEY

ORIGINAL ARTICLE

Translation, cross-cultural adaptation, and preliminary validation of a patient-reported outcome measure for genetic counseling outcomes in Sweden

Rebecka Pestoff[1,2,3] | Henrik Danielsson[4] | Marion McAllister[5] | Peter Johansson[6] | Cecilia Gunnarsson[1,2,7]

[1]Centre for Rare Diseases in Southeast Region of Sweden, Linköping University, Linköping, Sweden

[2]Division of Community Medicine, Department of Medical and Health Sciences, Linköping University, Linköping, Sweden

[3]Department of Clinical Genetics, Biomedical and Clinical Sciences, Linköping University, Linköping, Sweden

[4]Department of Behavioural Sciences and Learning, Linköping University, Linköping, Sweden

[5]Centre for Medical Education, School of Medicine, Cardiff University, Cardiff, UK

[6]Department of Health, Medicine and Caring Sciences, Linköping University, Linköping, Sweden

[7]Department of Clinical and Experimental Science, Linköping University, Linköping, Sweden

Correspondence
Rebecka Pestoff, Department of Clinical Genetics, Biomedical and Clinical Sciences, Linköping University, Linköping, Sweden.
Email: rebecka.pestoff@regionostergotland.se

Abstract

Genetic counseling is key for understanding the consequences of hereditary and genetic diseases and, therefore, crucial for patients, their families, and healthcare providers. Genetic counseling facilitates individuals' comprehension, decision-making, and adaptation to hereditary diseases. This study focuses on the Swedish adaptation of the Genetic Counseling Outcome Scale-24 (GCOS-24), an internationally validated, patient-reported outcome measure (PROM) for quantifying patient empowerment in genetic counseling. This study aimed to translate and cross-culturally adapt the GCOS-24 to measure patient-reported outcome from genetic counseling in Sweden. The adaptation process was meticulously conducted, adhering to international guidelines, with cross-cultural adaptation, translation, and back translation, to ensure semantic, conceptual, and idiomatic equivalence with the original English version. Face validity and understandability was assured using qualitative cognitive interviews conducted with patient representatives, and by a committee of experts in the field. The psychometric properties of the Swedish version of GCOS-24 (GCOS-24swe) were evaluated using a robust sample of 374 patients. These individuals received genetic counseling by telephone or video, necessitated by the constraints of the COVID-19 pandemic. Participants responded to GCOS-24swe both before and after genetic counseling. The GCOS-24swe demonstrated face validity, good internal consistency (Cronbach's alpha = 0.86), significant responsiveness (Cohen's $d = 0.65$, $p < 0.001$), and good construct validity. The study's findings underscore the GCOS-24swe's potential as an effective instrument in both clinical practice and research within Sweden. It offers a valuable means for assessing patient empowerment, a key goal of genetic counseling. Additional psychometric assessment of test–retest reliability and interpretability would further enhance the utility of GCOS-24swe.

KEYWORDS
clinical genetics, genetic counseling, outcome measures, patient empowerment

1 | INTRODUCTION

Genetic diseases profoundly impact individuals and their families. Genetic counseling is a crucial component for providing appropriate care and support. Understanding the complexities of genetic contributions to diseases, such as modes of inheritance and variations in expression, is vital. Genetic counseling plays an essential role in helping patients understand and adapt to the multifaceted nature of genetic conditions. This includes implications of genetic testing, living with a heritable disease, and making informed decisions (Fraser, 1974; Resta et al., 2006). The genetic counselor can provide a therapeutic relationship and empathic understanding of the patient's needs and concerns (Biesecker, 2020), thereby increasing patient empowerment. The Swedish context in which this study took place is patient-centered, similarly to international practice, for example, in the United Kingdom.

Reliable and validated patient-reported outcome measures (PROMs) are instrumental in capturing patient's perspective and evaluating the outcomes of care, including genetic counseling (Acquadro et al., 2001; Mokkink et al., 2010). A PROM can measure the benefits of clinical services and lead to improvements in service delivery. The patient–provider relationship and educational objectives can develop and reinforce patients' own psychosocial skills, and improve their knowledge and sense of control (Aujoulat et al., 2007). This can enhance their ability to make informed decisions, and to understand and adapt to medical, psychological, and familial implications regarding a genetic condition. Acquired knowledge and a sense of personal control leads to more active patients participating in their own and relatives' decision-making. The GCOS-24 is a widely used PROM used in genetic services around the world. It is specifically designed to measure patient empowerment, a central tenet of genetic counseling (McAllister, Dunn, & Todd, 2011).

The empowerment construct of GCOS-24 was grounded in comprehensive qualitative research involving patients and genetics service providers, and aimed to pinpoint the outcomes they value most (Hickmann et al., 2022; McAllister, Dunn, & Todd, 2011; McAllister, Wood, et al., 2011). The GCOS-24 captures five important aspects of patient empowerment: *Behavioral control; Decisional control; Cognitive control; Emotional regulation and Hope*, and aligns with the genetic counseling definition (McAllister et al., 2008; McAllister & Dearing, 2015). GCOS-24 was chosen because, to our knowledge, no previously published PROM captures patient empowerment in the clinical genetic setting (Payne et al., 2008) nor has undergone such extensive psychometric assessments. In English, the GCOS-24 shows validity, reliability, and sensitivity to change over time and medium-to-large effect size (Cohen's $d = 0.65$). It contains 24 items, with a 7-point Likert response scale (score range: 24–168) (McAllister & Dearing, 2015; McAllister, Wood, et al., 2011). Based on these qualities, the GCOS-24 was deemed the most suitable measure for the outcomes of genetic counseling in Sweden.

GCOS-24 has been extensively adapted and validated using different approaches in other cultural and clinical settings, including Brazil, Canada, Denmark, the Netherlands, Norway, Singapore, and Spain (Borle et al., 2022; Diness et al., 2017; Lleuger-Pujol et al., 2022; Løvik et al., 2022; Mochiki et al., 2023; Muñoz-Cabello et al., 2018; Pestoff et al., 2022; Redondo & McAllister, 2023; Segundo-Ribeiro et al., 2020; Voorwinden et al., 2019; Yuen et al., 2020). International use of GCOS-24 allows for evaluations of different genetic counseling contexts, languages, and service delivery models, after necessary case-mix adjustment (Burgess et al., 2019).

This current study aimed to perform a cross-cultural adaptation and preliminary validation of GCOS-24swe as a PROM for use in genetic counseling. Established psychometric properties determine the reliability, responsiveness, and factor structure of GCOS-24swe. Moreover, it enables Swedish healthcare professionals, researchers, and policymakers to make informed decisions and enhance the quality of genetic counseling services provided.

What is known about this topic:

Patient-reported outcome measures are useful in evaluating healthcare services. Previously, no validated measure was available for outcomes that correspond to the main tenets of genetic counseling in Swedish.

What this paper adds to the topic:

The genetic counseling outcome scale in Swedish shows reliability and validity for use in clinical care and provides a useful tool to evaluate and improve genetic services in Sweden.

2 | METHODS

To ensure the international applicability of GCOS-24, we conducted a comprehensive process encompassing translations, cross-cultural adaptations, and preliminary validations. Preserving the intended meaning of each item necessitated some modifications to the original instrument to maintain semantic, conceptual, and idiomatic equivalence (Beaton et al., 2000).

The process of cross-cultural adaptation and preliminary validation of the GCOS-24 in Swedish was approved by the Ethical Review Authority in Sweden for this project: *Dnr 2019–01051* and *Dnr 2020–05243*.

2.1 | Part I: Cross-cultural adaptation

Translation and cross-cultural adaptation were conducted to adapt GCOS-24 to the Swedish context and reach meaning equivalence with the English version. This was based on a modified protocol by Beaton et al. (2000) as shown in Table 1:

2.2 | Expert committee

An expert committee, consisting of clinicians and researchers fluent in both Swedish and English, played a pivotal role throughout the translation and adaptation process and in establishing face validity. Face validity means that the questionnaire items seem to measure the intended concept (Bolarinwa, 2015). The committee consisted of five experts in the field: *RP*, a certified genetic counselor and Ph.D. student; *CG*, a senior clinical geneticist and associate professor; *PJ*, a professor in Nursing Sciences; *PN*, a professor in Implementation Science; and *MM*, a professor in Genetic counseling and developer of the GCOS-24 in English. Written reports from each stage of the process were assessed, and a consensus was reached on a final version of the GCOS-24swe that proceeded to psychometric testing.

2.3 | Cognitive interviews with patient representatives

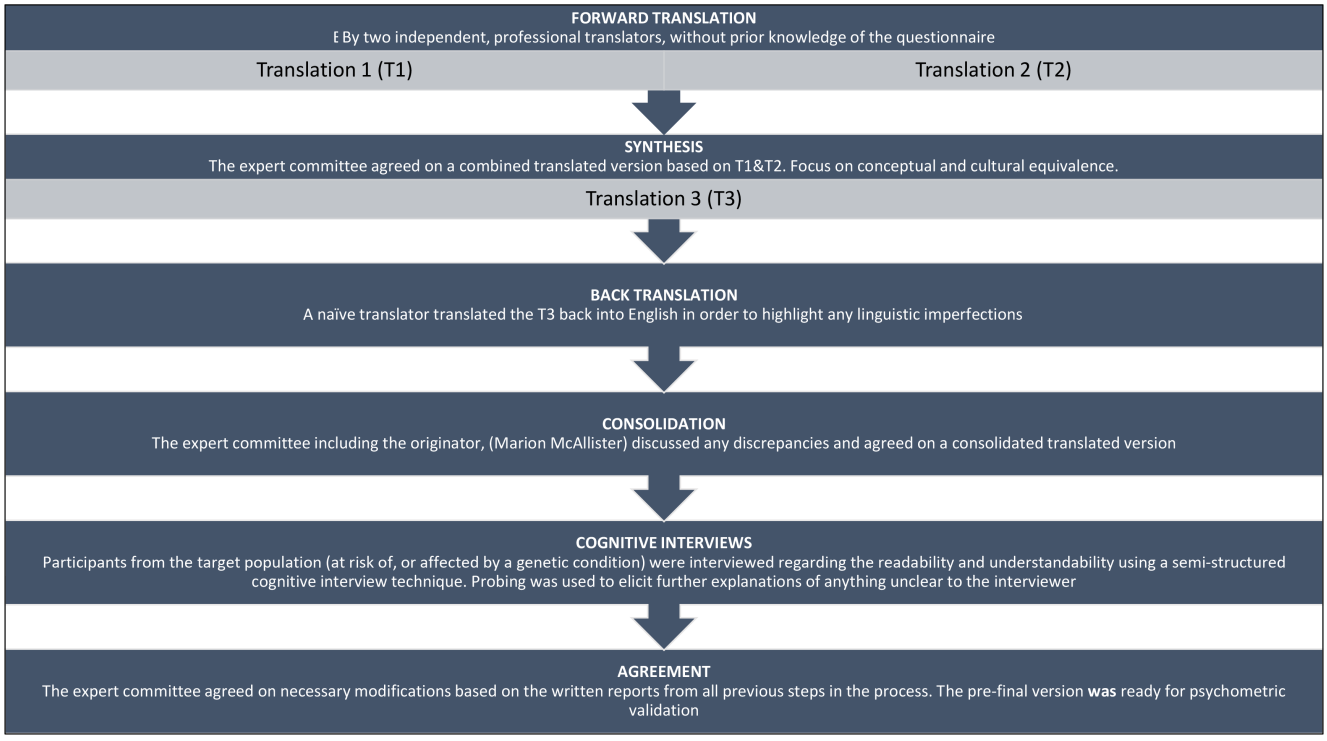
Cognitive interviews were conducted with patient representatives regarding the phrasing, format, and clarity of each questionnaire item. This process ensured face validity and enhanced readability, understanding, and identified issues related to item wording (Beatty & Willis, 2007; Hak et al., 2004). The probing technique was used to elicit participants' understanding and cognitive processes for each item. The data collected consisted of researchers' notes of issues per item and suggested modifications to resolve these issues (Willis, 2006).

2.4 | Participant selection for interviews

Strategic sampling was used to recruit a diverse group of patients for cognitive interviews, through the Rare Diseases Sweden Network (Sallsyntadiagnoser, 2022). Participants were informed about the study (Appendix S2) and represented a range of experiences related to genetic diseases (Wenemark, 2017). Included participants were 18 years of age or older; diagnosed with a genetic condition, or parent of a child with a genetic condition; and fluent in Swedish. Most participants had received genetic counseling previously. Participants were invited via email, including rights to withdraw from the study at any time, adhering to the ethical research principles outlined in the World Medical Association's Helsinki Declaration (Ethical Principles for Medical Research Involving Human Subjects, 1964).

Individual cognitive interviews were conducted at the participant's preferred location and included informed consent. First, the participant read each questionnaire item and then provided comments on the meaning of, and answers to, each item. In accordance with the method of cognitive interviewing, probing was used as needed (Appendix S3). The researcher took notes on each item (Appendix S4), summarized issues, and proposed modifications. These were discussed and reviewed by the expert committee (Beatty & Willis, 2007; Willis, 2006). Modifications were only made if they met the following criteria: (1) unanimous agreement in the committee, (2) consistent suggestions from two or more interviewees, and (3) not changing the idiomatic or conceptual meaning of the item (Irwin et al., 2009). These modifications

TABLE 1 Flow chart showing the protocol used for translation and cross-cultural adaptation (Steps# 1–6).



resulted in the new version of the questionnaire called the GCOS-24swe (Appendix S1).

2.5 | Part II: Psychometric evaluation

Furthermore, assessments encompassed evaluating internal consistency, responsiveness, and construct validity. *Internal consistency* assesses how items inter-relate and indicates the reliability of a scale. It is measured using Cronbach's alpha and is useful for multiple response options (Bolarinwa, 2015). *Responsiveness* measures a scale's ability to detect change over time, by comparing the scores before and after an intervention. It is measured by effect size, specifically Cohen's *d* (Bot et al., 2003). *Construct validity* examines how well the GCOS-24swe questionnaire measures the intended outcome, that is, whether the items load onto one dimension of empowerment, using confirmatory factor analysis (CFA). The CFA compared our data set with hypothetical constructs found in the literature (Bolarinwa, 2015).

Invited participants for the psychometric evaluation part of the study had a scheduled appointment for genetic counseling at a Department of Clinical Genetics (either at Karolinska University Hospital or Linköping University Hospital) in Sweden. Study information and invitation letters were distributed by site coordinators (Appendix S5). Genetic counseling consultations were conducted via telephone or video by trained clinical geneticists or genetic counselors. Participants consented by returning the completed GCOS-24swe questionnaire before their consultation. They were instructed to return the second questionnaire approximately 14 days after their consultation. Two postal reminders were sent if the post-appointment questionnaire was not returned.

Missing data is considered missing at random and imputed using multivariate imputation by chained equations (mice function in the R package mice) to create a complete dataset for all analyses (van Buuren & Groothuis-Oudshoorn, 2011). Previous studies in Dutch (Voorwinden et al., 2019), Chinese (Yuen et al., 2020), and English populations (McAllister, Wood, et al., 2011) were available at the time and provided information on item loadings and hypothetical construct dimensions. The Dutch version, Voorwinden et al. (2019) had identified six subscales, while Yuen et al. (2020) found two distinct scales. McAllister, Wood, et al. (2011) initially mentioned seven dimensions *but* recommended using the GCOS-24 as a unidimensional scale measuring the overarching construct of empowerment.

Psychometric results were available for the four aforementioned models (*Dutch with 6 factors; Chinese with 2*; *English with 7 and 1*). They were used for comparisons of construct models and were adjusted by adding residual covariances between items if the modification index >4. This ensured an improved fit without altering the factor structure, as the adjustment accounted for potential correlations between the error variances of items. The models were evaluated using various fit indices, including the Comparative Fit Index (CFI), root mean square error of approximation (RMSEA), and χ^2/df ratio. Cutoff criteria were applied to determine acceptable fit, such as a CFI of ≥0.90, an upper 90% confidence interval of RMSEA <0.08, and a χ^2/df ratio of ≤3 (Schreiber et al., 2006). All CFAs were conducted using R ((RCore Team, 2022) version 4.2.1) and RStudio (RStudio Team, 2020) with the lavaan package ((Rosseel, 2012) version 0.6.12). The mice package, ((van Buuren & Groothuis-Oudshoorn, 2011) version 3.14.0); papaja package, ((Aust & Barth, 2022) version 0.1.1); and the psych package ((Revelle, 2022) version 2.2.5) were utilized for data imputation. This analysis determined the most suitable construct model to fit the data from the GCOS-24swe.

3 | RESULTS

3.1 | Part I: Cross-cultural adaptation and translation process

Following the procedure delineated by Beaton et al. (2000) and detailed in our *Methods* section, we ensured semantic, idiomatic, and contextual equivalence, alongside face validity, while maintaining fidelity to the original English version of GCOS-24. The expert committee confirmed that the GCOS-24swe maintained face validity throughout the translation and adaptation process. Participants in the cognitive interviews were all females and were either affected by a genetic condition themselves (two-thirds) or mothers of affected children (one-third). The interviews took between 43 and 78 min, and the whole adaptation process can be found in Appendix S6.

3.2 | Examples of issues identified: Translation of "condition"

Throughout the translation and adaptation process, issues were identified. In the GCOS-24, the word "condition" appeared in items #2, 3, 4, 6, 7, 9, 11, 12, 13, 16, 17, 18, 21, 22, and 24. Initially, the word "condition" was translated to [tillstånd] in Swedish, which literately means *permission/condition/state*. To further clarify that it referred to a medical/genetic condition, the expert committee decided to add "medical" in front of "condition," resulting in the Swedish translation [medicinskt tillstånd]. Another possible translation of "condition" could have been [diagnos], meaning "diagnosis" in English. However, since not all individuals seen for genetic counseling have received a medical diagnosis themselves or in their family, this information is unknown to many. Additionally, not all patients perceive their genetic condition as a diagnosis, as the term may carry a negative connotation for some individuals. To address this, respondents in the interviews were asked about their preferred translation for "condition." The same semantic issue was also noted in the Danish translation conducted by Diness et al. (2017), emphasizing the resemblance between the Swedish and Danish languages.

3.3 | Negative wording

The expert committee acknowledged the potential challenges posed to respondents by negatively worded items. Previously reported similar findings (Diness et al., 2017; Grant et al., 2019; Wenemark, 2017) indicated avoiding negatively worded items if possible. This issue was also highlighted in the cognitive interviews. However, a Norwegian publication (Løvik et al., 2022) arrived at a different conclusion and did not rephrase negative items.

For example, the following items #10, 12, 17, and 18 were negatively worded in the English GCOS-24. All our respondents supported changing the wording of these negatively framed items into positive framing, as shown in the following quotes:

> I wish this question wasn't negatively framed....
>
> Respondent #1

> ok, if negative framing is removed....
>
> Respondent #4

> ...negative framing, it is strange to turn it around in my head, difficult to answer.
>
> Respondent #5

> Remove [replace] the negation.
>
> Respondent #6

Some items included words that in themselves were negatively loaded. Nevertheless, respondents considered these items easy to understand and did not need to be changed to a positive framing. The following items and words: item # 4 (word "*upset*"), 11 ("*anxious*"), 21 ("*guilty*"), and 22 ("*powerless*") scores therefore need to be reversed upon analysis.

3.3.1 | Clarification through examples

Items # 5, 10, 12, 15, and 16 prompted queries regarding specific options or decisions. Upon discussion in the expert committee and with interview respondents, it was agreed on adding examples in parenthesis. The reason being if respondents do not understand their options this will reduce the number of responses to these items.

> Item #5: Give examples: such as prophylactic operations, and counseling.
>
> Respondent 4

> Item #10: I don't know which the different options are.
>
> Respondent 3

> Item #15: Give examples: living assistance, social support, patient organizations.
>
> Respondent 2

> Item #16: Give examples....
>
> Respondent 1

3.3.2 | Other issues

Minor grammatical and linguistic adaptations were made following the expert committee's recommendations after the

TABLE 2 Participant characteristics of respondents to GCOS-24swe.

Participant characteristics	Total invitations % (*n*)	Only pre-responses[a] % (*n*)	Both responses (pre and post)[b] % (*n*)
Respondents	100 (742)	50 (374)	34 (254)
Female		73 (273)	80 (204)
Mean age		47.06 (SD 16.69)	47.02 (SD 16.72)
Parent		75 (281)	77 (195)
Has the condition		45 (168)	49 (125)
Does not have the condition		23 (86)	22 (56)
Does not know if they have the condition		32 (120)	28 (72)
Referral hereditary cancer		64 (234)	67 (169)
Other common Referral reasons	Coagulatory defect; Neuromuscular disease; Huntington's Disease; Epilepsy; Infertility or chromosomal abnormality; Eye disorder; Maturity Onset Diabetes of the Young (MODY); Renal disease		

[a]Before genetic counseling consultation.

[b]Before and after genetic counseling consultation.

synthesis, consolidation, and cognitive interviewing steps (as found in Appendix S6). Several suggestions made by the expert committee during the translation phase were also supported by findings from the cognitive interviews. A step-wise adaptation of individual items was performed based on expert committee reasoning and interview responses. This resulted in the GCOS-24swe version found in Appendix S1.

3.4 | Part II: Statistical analysis

A convenience sampling method was employed, and 742 patients met the inclusion criteria and were invited to participate in the study. Recruitment took place during the COVID-19 pandemic between May 2020 and December 2021.

The first questionnaire was returned by 374 invited participants resulting in a 50% response rate. Of those, 254 returned the second questionnaire, resulting in a 68% response rate. Most participants were female with a mean age of 47 years, and almost two-thirds had a referral regarding oncogenetics, as shown in Table 2.

Fewer than 10% had missing data, and the proportion of respondents with missing data was very low (0.34%) and therefore treated as missing at random. The analysis of internal consistency yielded a standardized Cronbach's alpha coefficient of 0.86, indicating good reliability ($0.90 > \alpha \geq 0.80$) (George & Mallery, 2003). To assess responsiveness, Cohen's *d* was calculated to 0.65 ($p > 0.001$) and indicated a medium-to-large effect size (cutoffs: small = 0.2; medium = 0.5, large = 0.8 (Cohen, 1992)). The mean improvement in scores was +12.87 (95% CI [10.74, 14.99]) and showed that the GCOS-24swe captured change over time, when before and after genetic counseling were compared. Furthermore, confirmatory factor analysis (CFA) demonstrated construct validity as the items in the GCOS-24swe loaded onto a single dimension. These evaluations were based on the assumption that genetic counseling enhances patient empowerment and that the GCOS-24swe serves as a valid outcome measure. The one-factor and 7-dimension models based (McAllister, Wood, et al., 2011) exhibited the best fit to our data and met the predetermined cutoff values for all fit indices (Table 3). As comparisons, four models from previously published studies were used: 1 and 7 factors in McAllister, Wood, et al., 2011, six factors in Voorwinden et al. (2019), and two factors in Yuen et al. (2020). Yuen's two-factor model was the second best fit (meeting the criterion for RMSEA but not cutoffs for the other indices). The models with six and seven factors did not demonstrate satisfactory fit for any of the indices and indicated a poor fit for the current sample with the predetermined thresholds. The fit indices for the CFAs of the four models can be found in Table 3. All models were adjusted by incorporating residual covariances; however, the McAllister1 model required the most adjustments, contributing to its superior fit. Despite this, our interpretation of the results supported the conclusion that the McAllister1 model was the best fit for our study's data to measure the empowerment construct.

4 | DISCUSSION

The present study evaluated the psychometric properties of the GCOS-24swe questionnaire, specifically tailored for the Swedish-speaking population. Specifically, face validity, internal consistency, responsiveness, and construct validity were examined. The cross-cultural adaptation process encompassed contributions from an expert committee, translation and back translation, insights from six qualitative patient interviews, and a confirmatory factor analysis based on 374 patient responses.

4.1 | Reliability and validity of GCOS-24swe

The GCOS-24swe was found to be reliable and valid. The cross-cultural adaptation process led to some changes compared to the English version of GCOS-24. The main one was to reverse negatively framed items to positive ones (Grant et al., 2019). Identifying the optimal translation for the word *condition* posed a challenge; however, the suggested Swedish term [medicinskt tillstånd] was unanimously chosen by both the patients and the expert committee as the best semantic, cultural, and conceptual equivalent. Analysis demonstrated that the adapted questionnaire had good internal consistency (Cronbach's alpha of 0.86), and suggests that the items in the questionnaire consistently

TABLE 3 Fit indices for the tested models.

Model	χ^2	df	χ^2/df	CFI	RMSEA	RMSEA 90% CI	AIC
McAllister7	874.68	61	14.34	0.824	0.091	0.097	30,134
Yuen	494.52	99	5.00	0.924	0.062	**0.069**	31,133
Voorwinden	483.56	47	10.29	0.879	0.088	0.096	23,186
McAllister1	351.19	121	**2.90**	**0.955**	0.051	**0.059**	31,034

Note: Bold values indicate that they meet the cutoffs for acceptable fit: ≤3 for χ^2/df, (≥0.95) for CFI, and <0.08 for upper 90% confidence interval for RMSEA. Additional fit indices without specific cutoff values were also reported for descriptive purposes.

Abbreviations: AIC, akaike information criterion; CFI, the comparative fit index; CI, confidence interval; df, degrees of freedom, RMSEA, root mean square error of approximation.

measure the same underlying construct of empowerment. This is similar to the Cronbach's alpha 0.87 originally reported (McAllister & Dearing, 2015), and higher than reported in the Spanish study (0.71) (Segundo-Ribeiro et al., 2020). This suggests that the GCOS-24swe is a reliable tool for assessing patient-reported outcomes among individuals undergoing genetic counseling in the Swedish population.

After genetic counseling, the GCOS-24swe demonstrated responsiveness (Cohen's $d = 0.65$), and a moderate, but significant improvement of (+12.87) in patient empowerment. This is to compare with the other GCOS-24 validation studies, which reported the following effect sizes: 0.70 (McAllister, Wood, et al., 2011), *"significant"* (Yuen et al., 2020), and 0.30 (Voorwinden et al., 2019). The mean improvement showed a significant benefit for patients of genetic counseling.

The factor analysis of GCOS-24swe showed that it measured empowerment as a single overarching concept and demonstrated a good fit for the data (for fit indices *RMSEA*, df, X^2, *and CFI*, see Table 3). This is consistent with the original recommendations for use of the measure that found seven factors that are ordered under one construct of empowerment (McAllister, Wood, et al., 2011). However, the validation studies in different cultural and linguistic contexts found other constructs that may be better suited for those specific patient populations. The study by Yuen et al. (2020) used a Rasch model and suggested dividing the overall score into two domains including *Cognitive control* and *Emotional Control*, although with imperfect fit. Meanwhile, Voorwinden et al. (2019) suggested a 6-factor structure and removed six items due to low factor loading or low internal consistency (items #13, 15, 22, 24, 6, 7) (Voorwinden et al., 2019; Yuen et al., 2020). Based on our findings (face validity, interviewees understanding, CFA, Cronbach's alpha, and Cohen's *d*), the GCOS-24swe is considered a valid and reliable outcome measure for genetic counseling in Swedish. In Sweden, genetic counseling is still evolving and is not yet mandated by law, unlike in neighboring Norway. The validated GCOS-24swe offers a robust evidence base for refining current practices and policies, and substantiates the impact of genetic counseling in Sweden, as well as in other countries where such services are in the developmental phase.

4.2 | Strengths and limitations

The diligence with which the cross-cultural adaptation of the questionnaire was performed was a strength in this study. As was the number of participants for the statistical validation ($n = 374$) and the relatively low rate of missing data, which was less than 10%.

There were also some limitations in this study. Firstly, the participants in the statistical validation ($n = 374$) were a convenience sample that only had digital consultations, via telephone or video due to the COVID-19 pandemic (Pestoff et al., 2022). Demographics such as education, income, and ethnic background were not collected, which made it difficult to know how representative the sample was. The participants were overrepresented by women (73%) and referrals related to hereditary cancer (63%). However, these percentages were comparable to previously published studies in the field (Diness et al., 2017). The study relied mostly on self-reported measures, which may be subject to response biases and social desirability effects. Only six cognitive interviews were performed, although the literature recommends conducting 8–10 interviews (Beatty & Willis, 2007; Willis, 2006). Recruitment of eligible respondents proved challenging, so the expert committee decided to proceed after six conducted interviews as participants' responses were very much aligned. Not all respondents to GCOS-24swe had children, meaning that several questions did not apply to those individuals. Instead, these respondents were instructed to reply option 4, *neither agree nor disagree*, whenever a question was not relevant regarding children. This limitation was identified previously in other studies on the translation of GCOS-24 (Grant et al., 2019). Another limitation of this study was that no test–retest reliability or interpretability (MCID) assessments were performed, which is why further validation of the GCOS-24swe is recommended.

4.3 | Clinical implications

The findings of this study hold significant clinical implications for the practice of genetic counseling in Sweden. The validated GCOS-24swe provides clinicians with a standardized outcome measure on the impact of genetic counseling on patients. It can inform potential service improvements and evidence-based practice in the field of genetic counseling and promote the quality of care provided and improve patient outcomes. Comparative analysis of other cultures and settings can guide clinicians in adapting their counseling approaches to be culturally sensitive and responsive.

4.4 | Future research

Future research could use Rasch measurement theory to assess the fitness of GCOS-24swe (Borle et al., 2022) and should incorporate longitudinal studies using the GCOS-24swe to elucidate the long-term effects of genetic counseling. While the current study assessed responsiveness by comparing pre- and post-counseling outcomes, a longitudinal design would allow investigating empowerment changes over an extended period. It is also important to include a cohort of face-to-face counseling sessions, to gain a better understanding of how empowerment changes are affected by different contexts or service delivery models.

4.5 | Conclusion

In conclusion, this research provided evidence for the validity and reliability of the Swedish version of the Genetic Counseling Outcome Scale (GCOS-24swe) as a patient-reported outcome measure for genetic counseling. Our findings reveal that the GCOS-24swe exhibits

good internal consistency and responsiveness to change over time. The confirmatory factor analysis supported a unidimensional model of empowerment, suggesting that the GCOS-24swe captured the intended construct and main tenet of genetic counseling, specifically empowerment. This research thus presents the GCOS-24swe as a culturally adapted and validated tool for evaluating the outcomes of genetic counseling interventions in Sweden. Nevertheless, we recommend further psychometric assessment, including test–retest analysis and interpretability studies, to enhance its applicability.

AUTHOR CONTRIBUTIONS

Author Rebecka Pestoff confirms that she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

ACKNOWLEDGMENTS

Our deepest appreciation for the use of the GCOS-24 instrument that was approved by Marion McAllister (original author) and Weil and Sons publishing company. Also many thanks to participating patients and clinical staff at the Departments of Clinical Genetics at Linköping University Hospital and Karolinska University Hospital, Sweden, for giving both their valuable time and thoughts. A special thank you to all participants and the expert committee. This work was conducted to fulfill a degree requirement for the main author's PhD.

FUNDING INFORMATION

Funding for this project was provided by the County Council of Östergötland, Sweden, for PhD student Rebecka Pestoff.

CONFLICT OF INTEREST STATEMENT

The authors declare no competing interests.

DATA AVAILABILITY STATEMENT

Data are available upon request.

ETHICAL APPROVAL

Human Studies and Informed Consent: Participants were informed that participating was entirely voluntary and anonymous and would not affect their clinical care. Implied informed consent was obtained for all individuals who voluntarily completed and submitted their responses. They were also informed of their rights to decline participation (*by not responding at all*), accept participation (*by returning a completed questionnaire*), and revoke participation at any time. It was deemed not harmful to patients taking part in interviews or responding to the questionnaires. Original data are available upon request to the main author. The Ethical Review Authority in Sweden granted ethical approval for this project: *Dnr 2019–01051* and *Dnr 2020–05243*.

ORCID

Rebecka Pestoff https://orcid.org/0000-0001-7192-4532

REFERENCES

Acquadro, C., Berzon, R., Dubois, D., Leidy, N. K., Marquis, P., Revicki, D., & Rothman, M. (2001). Incorporating the patient's perspective into drug development and communication: An ad hoc task force report of the patient-reported outcomes (PRO) harmonization group meeting at the Food and Drug Administration. *Value in Health*, *6*, 522–531.

Aujoulat, I., d'Hoore, W., & Deccache, A. (2007). Patient empowerment in theory and practice: Polysemy or cacophony? *Patient Education and Counseling*, *66*(1), 13–20. https://doi.org/10.1016/j.pec.2006.09.008

Aust, F., & Barth, M. (2022). PAPAJA*: Prepare reproducible APA journal articles with R Markdown*. Retrieved from https://github.com/crsh/papaja

Beaton, D. E., Bombardier, C., Guillemin, F., & Bosi Ferraz, M. (2000). Guidelines for the process of cross-cultural adaptation of self-report measures. *Spine*, *25*(24), 3186–3191.

Beatty, P. C., & Willis, G. B. (2007). Research synthesis: The practice of cognitive interviewing. *Public Opinion Quarterly*, *71*(2), 287–311. https://doi.org/10.1093/poq/nfm006

Biesecker, B. (2020). Genetic counseling and the central tenets of practice. *Cold Spring Harbor Perspectives in Medicine*, *10*(3), a038968. https://doi.org/10.1101/cshperspect.a038968

Bolarinwa, O. A. (2015). Principles and methods of validity and reliability testing of questionnaires used in social and health science researches. *The Nigerian Postgraduate Medical Journal*, *22*(4), 195–201. https://doi.org/10.4103/1117-1936.173959

Borle, K., Austin, J., & Barbic, S. (2022). Using Rasch measurement theory to explore the fitness for purpose of the genetic counseling outcome scale: A tale of two scales. *Quality of Life Research*, *32*, 895–904. https://doi.org/10.1007/s11136-022-03289-7

Bot, S. D. M., Terwee, C. B., van der Windt, D., Boutert, L. M., Dekkert, J., & De Vet, H. C. W. (2003). Psychometric evaluation of self-report questionnaires – the development of a checklist. In *Proceeding of the second workshop on research methodology* (pp. 161-168).

Burgess, R., Bishop, A., Lewis, M., & Hill, J. (2019). Models used for case-mix adjustment of patient reported outcome measures (PROMs) in musculoskeletal healthcare: A systematic review of the literature. *Physiotherapy*, *105*(2), 137–146. https://doi.org/10.1016/j.physio.2018.10.002

Cohen, J. (1992). A power primer. *Psychological Bulletin*, *112*, 155–159. https://doi.org/10.1037//0033-2909.112.1.155

Diness, B. R., Overbeck, G., Hjortshoj, T. D., Hammer, T. B., Timshel, S., Sorensen, E., & McAllister, M. (2017). Translation and adaptation of the genetic counseling outcome scale (GCOS-24) for use in Denmark. *Journal of Genetic Counseling*, *26*(5), 1080–1089. https://doi.org/10.1007/s10897-017-0086-7

Fraser, F. C. (1974). Genetic counseling. *American Journal of Human Genetics*, *26*(5), 636–659.

George, D., & Mallery, P. (2003). *SPSS for Windows step by step: A simple guide and reference, 11.0 update* (4th ed.). Boston, MA: Allyn & Bacon.

Grant, P. E., Pampaka, M., Payne, K., Clarke, A., & McAllister, M. (2019). Developing a short-form of the genetic counseling outcome scale: The genomics outcome scale. *European Journal of Medical Genetics*, *62*(5), 324–334. https://doi.org/10.1016/j.ejmg.2018.11.015

Hak, T., van der Veer, K., & Jansen, H. (2004). The Three-Step Test-Interview (Tsti): An observational instrument for pretesting self-completion questionnaires. *Report series research in management*.

Hickmann, E., Richter, P., & Schlieter, H. (2022). All together now – patient engagement, patient empowerment, and associated terms

in personal healthcare. *BMC Health Services Research*, *22*(1), 1116. https://doi.org/10.1186/s12913-022-08501-5

Irwin, D. E., Varni, J. W., Yeatts, K., & DeWalt, D. A. (2009). Cognitive interviewing methodology in the development of a pediatric item bank: A patient reported outcomes measurement information system (PROMIS) study. *Health and Quality of Life Outcomes*, *7*, 3. https://doi.org/10.1186/1477-7525-7-3

Lleuger-Pujol, R., Castello, E. O., Franco, L. F., Vallejo, M. E. E., Cabello, P. M., Simarro, F. S., McAllister, M., & Garcia-Minaur, S. (2022). Further validation and psychometric properties of the Spanish adaptation of the genetic counseling outcome scale. *Journal of Genetic Counseling*, *31*(1), 71–81. https://doi.org/10.1002/jgc4.1452

Løvik, I. K., Siglen, E., & Bjorvatn, C. (2022). *Adaptive translation of the genetic counseling outcome scale – A patient-reported outcome measure for application in genetic counseling in Norway* (Master's thesis, University of Bergen, Norway).

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

McAllister, M., Dunn, G., & Todd, C. (2011). Empowerment: Qualitative underpinning of a new clinical genetics-specific patient-reported outcome. *European Journal of Human Genetics*, *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). Patient empowerment in clinical genetics services. *Journal of Health Psychology*, *13*(7), 895–905. https://doi.org/10.1177/1359105308095063

McAllister, M., Wood, A. M., Dunn, G., Shiloh, S., & Todd, C. (2011). The genetic counseling outcome scale: A new patient-reported outcome measure for clinical genetics services. *Clinical Genetics*, *79*(5), 413–424. https://doi.org/10.1111/j.1399-0004.2011.01636.x

Mochiki, I., Okugawa, Y., Hashizume, R., Imai, H., Ikejiri, M., Ogura, T., Nakatani, K., & Hori, H. (2023). Psychological characteristics of Japanese patients and their family members receiving genetic counseling: A single-institute exploratory study. *Journal of Genetic Counseling*, *32*, 128–139. https://doi.org/10.1002/jgc4.1629

Mokkink, L. B., Terwee, C. B., Knol, D. L., Stratford, P. W., Alonso, J., Patrick, D. L., Bouter, L. M., & De Vet, H. C. W. (2010). The COSMIN checklist for evaluating the methodological quality of studies on measurement properties: A clarification of its content. *BMC Medical Research Methodology*, *10*, 22. https://doi.org/10.1186/1471-2288-10-22

Muñoz-Cabello, P., García-Miñaúr, S., Espinel-Vallejo, M. E., Fernández-Franco, L., Stephens, A., Santos-Simarro, F., Lapunzina-Badía, P., & McAllister, M. (2018). Translation and cross-cultural adaptation with preliminary validation of GCOS-24 for use in Spain. *Journal of Genetic Counseling*, *27*(3), 732–743. https://doi.org/10.1007/s10897-017-0154-z

National Society of Genetic Counselors' Definition Task Force, Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A new definition of genetic counseling: National Society of genetic Counselors' task force report. *Journal of Genetic Counseling*, *15*(2), 77–83. https://doi.org/10.1007/s10897-005-9014-3

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

Pestoff, R., P., Danielsson, H., Neher, M., & Gunnarsson, C. (2022). Rapid implementation of Telegenetic counseling in the COVID-19 and Swedish healthcare context: A feasibility study. *Frontiers in Health Services*, *2*, 848512. https://doi.org/10.3389/frhs.2022.848512

RCore Team. (2022). R: A language and environment for statistical computing. Retrieved from https://www.R-project.org/. https://www.R-project.org/

Redondo, L., & McAllister, M. (2023). Cross-cultural adaptation of the genetic counseling outcome scale (GCOS-24) for use in Canada: A qualitative study. *Journal of Genetic Counseling*, 1–20. https://doi.org/10.1002/jgc4.1771

Revelle, W. (2022). *Psych: Procedures for psychological, psychometric, and personality research*. Retrieved from https://CRAN.R-project.org/package=psych. https://CRAN.R-project.org/package=psych

Rosseel, Y. (2012). Lavaan: An R package for structural equation modeling. *Journal of Statistical Software*, *48*(2), 1–26. https://doi.org/10.18637/jss.v048.i02

RStudio Team. (2020). RStudio: Integrated development environment for R. Retrieved from http://www.rstudio.com/

Sallsyntadiagnoser. (2022). Rare Diseases Sweden: Regional Networks. Retrieved from https://www.sallsyntadiagnoser.se/regionala-natverk/

Schreiber, J. B., Nora, A., Stage, F. K., Barlow, E. A., & King, J. (2006). Reporting structural equation modeling and confirmatory factor analysis results: A review. *The Journal of Educational Research*, *99*(6), 323–338. https://doi.org/10.3200/JOER.99.6.323-338

Segundo-Ribeiro, M., Bacala, B. T., Alvarenga, W. A., Nascimento, L. C., McAllister, M., & Floria-Santos, M. (2020). Adaptation and preliminary validation of the genetic counseling outcome scale (GCOS-24) in a Brazilian genetic counseling setting. *European Journal of Medical Genetics*, *63*(11), 104018. https://doi.org/10.1016/j.ejmg.2020.104018

van Buuren, S., & Groothuis-Oudshoorn, K. (2011). Mice: Multivariate imputation by chained equations in r. *Journal of Statistical Software*, *45*(3), 1–67. https://doi.org/10.18637/jss.v045.i03

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). A validated PROM in genetic counseling: The psychometric properties of the Dutch version of the genetic counseling outcome scale. *European Journal of Human Genetics*, *27*(5), 681–690. https://doi.org/10.1038/s41431-018-0318-9

Wenemark, M. (2017). *Enkätmetodik med respondenten i fokus*. Studentlitteratur.

Willis, G. B. (2006). Cognitive interviewing as a tool for improving the informed consent process. *Journal of Empirical Research on Human Research Ethics*, *1*, 9–24.

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

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

How to cite this article: Pestoff, R., Danielsson, H., McAllister, M., Johansson, P., & Gunnarsson, C. (2024). Translation, cross-cultural adaptation, and preliminary validation of a patient-reported outcome measure for genetic counseling outcomes in Sweden. *Journal of Genetic Counseling*, *00*, 1–9. https://doi.org/10.1002/jgc4.1896