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ORIGINAL ARTICLE

Cross-cultural adaptation of the Genetic Counseling Outcome Scale (GCOS-24) for use in Canada: A qualitative study

Laura Redondo  | Marion McAllisterCentre for Medical Education, Cardiff
University, Cardiff, UK**Correspondence**Laura Redondo, Centre for Medical
Education, Cardiff University, Cardiff, UK.
Email: lauraredondo95@gmail.com**Abstract**

The Genetic Counselling Outcome Scale (GCOS)-24 is a patient-reported outcome measure (PROM) developed and validated in the United Kingdom (UK). The aim of this study was to cross-culturally adapt GCOS-24 to Canadian Clinical Genetic Services (CGS). This was achieved through a qualitative study exploring whether the existing GCOS-24 maintains its intended meaning in a Canadian population and assessing whether GCOS-24 items could be better worded to meet the needs of members of families affected by genetic conditions in Canada. Thirteen participants were recruited from Canadian Patient Organizations supporting people and families affected by genetic conditions. Data were collected through semi-structured cognitive interviews, as these allow exploration of participants' comprehension, opinions, thoughts, and feelings regarding GCOS-24's instructions, response options, and the meaning/relevance of each item. Thematic analysis was utilized for data analysis, and an inductive approach to coding was followed to allow for themes to emerge from the data. Themes were organized in respect to their questionnaire item and further classified into their respective Empowerment dimension. The GCOS-24 instructions were found easy to understand by all thirteen participants. Although the response options were also found to be straightforward, the data suggest the questionnaire would benefit from the addition of a "non-applicable" option. Semantic validation of the GCOS-24 showed that items within the Cognitive Control and Emotional Regulation dimensions were found easy to understand by participants. However, items within the Decisional Control, Behavioural Control and Hope dimensions presented semantic difficulties. Participants provided feedback on syntactic changes to support understanding, and this feedback was used to develop a final Canadian-adapted version of GCOS-24, GCOS-Canada. This study provides the first step towards a valid, culturally adapted PROM for use in Canadian CGS service evaluation and research. GCOS-Canada would benefit from psychometric validation to ensure validity, reliability, responsiveness, minimal clinically important difference and internal consistency.

KEYWORDS

GCOS-24, GCOS-Canada, genetic counseling, patient-reported outcome measure

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1 | INTRODUCTION

Present-day genetic counseling sessions include the interpretation of family/medical histories; education about inheritance, testing, management, and resources; and counseling to promote informed choices (Metcalfe, 2018). The growth of genetic counseling as a field and thus the increase of available genetic tests and clinical genetic referrals has resulted in a search to establish a valid, reliable, and responsive method to evaluate genetic counseling interventions and associated testing (hereinafter referred to as clinical genetic services (CGS)). Valid, psychometrically sound measures of the benefits provided by CGS are key for service evaluation and improvement, patient-centered practice, and clinical budget allocation. However, the definition of successful genetic counseling is rarely stated explicitly, which has, in turn, limited its evaluation. To add to the intricacy, CGS often deal with occurrence or risk of a genetic condition in a family, and the treatment or cure of genetic conditions is not the main objective—although sometimes possible. Clinical genetics professionals have argued that traditional healthcare outcome measures, such as measures of morbidity and mortality, are not appropriate to measure such services and have focused more on evaluating psychosocial outcomes (McAllister et al., 2007; Payne et al., 2007; Redlinger-Grosse et al., 2016). The complexity of genetic counseling contributes to challenges in the published research investigating its outcomes. Although studies have shown that genetic counseling can lead to reductions in anxiety, cancer-related worry, and decisional conflict, as well as increased knowledge and patient satisfaction, the existing literature faces limitations (Madlensky et al., 2017; Payne et al., 2008). In their comprehensive literature review, Madlensky et al. (2017) highlighted that although a wide variety of outcomes can be considered in genetic counseling, there are challenges in using various measures. This diversity in measurement tools hinders the ability to make meaningful cross-study comparisons regarding the effectiveness and value of genetic counseling.

Standardized, validated patient-reported outcome measures (PROMs) have been introduced to provide evidence-based evaluation of healthcare interventions. These provide an important patient perspective when evaluating healthcare services. PROMs measure the patient's health status or health-related quality of life (HRQoL) directly from the patient's standpoint, without interpretation of their response by a healthcare professional (U.S. Department of Health and Human Services FDA Center for Drug Evaluation and Research et al., 2009). PROMs have been incorporated for routine use in a number of countries, such as the United Kingdom and the United States, with the aim of evaluating healthcare quality (Baumhauer & Bozic, 2016; Cella et al., 2007; Dawson et al., 2010).

Once a patient-reported outcome (PRO) is determined to be appropriate for the evaluation of a specific service, psychometric validation of a PROM to capture that PRO is important as inadequately designed PROMs can have adverse ethical consequences, as patients might be completing measures that are not properly capturing the patient's perspective (McKenna, 2011). For example, assessing the reliability of a PROM speaks to its ability to yield reproducible

What is known about this topic

Although the Genetic Counselling Outcome Scale (GCOS-24) has been used as a tool for research, service evaluation and genetics healthcare quality improvement, and researchers across the globe have translated and cross-culturally adapted GCOS-24 for use in their country of interest, the tool has yet to be cross-culturally adapted to a Canadian population. As genetic counselling continues to expand, so does the value of its assessment and using PROMs world-wide.

What this paper adds to this topic

This is the first study to cross-culturally adapt GCOS-24 for use in Canada, providing the first step to a valid, culturally adapted PROM for use in clinical genetics research, service evaluation, quality assessment, and budget allocation.

and consistent results and this is assessed by determining the internal consistency and test-retest reliability of the PROM. The validity of a PROM refers to the ability of the instrument to measure what it is intended to measure, and this is determined by assessing the PROM's face validity, construct validity and criterion validity (Shah et al., 2016). Responsiveness of a PROM speaks to its ability to detect change over time, and this is assessed by determining the effect size (e.g. by calculating Cohen's *d*). Evidence-based assessment of the psychometric properties is needed to ensure quality in both the process and results of outcome research and service evaluations.

A systematic review conducted by Payne et al. (2008) found a total of 67 validated outcome measures, each covering domains such as perceived personal control (PPC), information-recall, satisfaction, levels of anxiety, quality of life, and perception of risk. The review found none of these PROMs encompass all the benefits that a patient receives from CGS. Further, only 25 out of the 67 measures were psychometrically assessed for test-retest reliability, only five for responsiveness and only two for interpretability (McAllister & Dearing, 2015; Payne et al., 2008). The review also highlighted the lack of consistency in the field, as most PROMs used in genetic counseling research were used only once.

Qualitative grounded theory research in genetic counseling patient benefits proposed the construct of 'empowerment' as an outcome of genetic counseling and thus applicable to its evaluation. Empowerment is defined as "a set of beliefs that enable a person affected by a genetic condition, or at risk for developing or transmitting a genetic condition, to feel that they have some degree of control over and hope for the future" (McAllister et al., 2008, p. 901). The construct was developed through qualitative grounded theory research which aimed to further explore what patients expect from CGS and allowed the researcher to develop a psychosocial model (McAllister, 2001; McAllister et al., 2008). Additional qualitative research to explore the

validity, relevance and importance of the “empowerment” construct found it to be useful as a PRO, and redefined the construct to five dimensions: cognitive, decisional, and behavioral control; knowledge and understanding; decision-making; hope and emotional regulation (McAllister, Dunn, & Todd, 2011). The empowerment construct was then operationalized as a PROM, the Genetic Counselling Outcome Scale (hereinafter referred to as GCOS-24; Figure 1). GCOS-24 was designed to evaluate outcomes of CGS, encompassing the involvement of both clinical geneticists and genetic counselors (McAllister, Wood, et al., 2011). Both types of genetics health professionals provide both genetic counseling and testing in UK clinical genetics. While the focus of GCOS-24 is on the patient's perspective and experience within the context of clinical genetics, it acknowledges the collaborative nature of genetic counseling within CGS. Psychometric validation of GCO2-24 demonstrated that this PROM is valid, reliable, and responsive. (McAllister, Wood, et al., 2011). Studies have found that GCOS is useful for clinical service evaluation and quality improvement (Costal Tirado et al., 2017), well-accepted by clients and can be used to demonstrate that services provide measurable patient benefits (Inglis et al., 2015; Ison et al., 2019; McAllister et al., 2016).

Additionally, Thomas and McAllister (2019) demonstrated the minimum clinically important difference (MCID) for GCOS-24 to be 10.3 points, which contributes to the interpretability of the tool. Lastly, the psychometric properties of GCOS-24 were further optimized using Rasch analysis, which found the scale to be multidimensional in its overall form (Yuen et al., 2020). As a result, GCOS-24 is more psychometrically robust than any other measure currently available for evaluation of CGS. Further, the inclusion of the PPC dimensions and those of emotional regulation (which represents elements such as anxiety and guilt) and hope makes GCOS-24 more relevant than its predecessors when evaluating clinical genetics, as it incorporates domains identified by patients and other stakeholders as valued outcomes from genetic counseling.

GCOS-24 has been utilized as a tool for healthcare quality improvement, and researchers across the globe have translated and cross-culturally adapted GCOS-24 for use in Denmark (Diness et al., 2017), the Netherlands (Voorwinden et al., 2019), Spain (Muñoz-Cabello et al., 2018), Brazil (Segundo-Ribeiro et al., 2020), and Singapore (Yuen et al., 2020). Five studies have applied GCOS-24 to a Canadian population. Three of these utilized GCOS-24 for service evaluation, evaluating the services provided by the first specialist psychiatric genetic counseling clinic in British Columbia (Inglis et al., 2015), measuring the impact of counseling physical environment on patient outcomes (Morris et al., 2019), and assessing the impact of psychiatric genetic counseling in empowerment (Gerrard et al., 2020). The other two studies aimed to adapt GCOS-24 for autism spectrum disorders and related conditions (Yusuf et al., 2021) and use Rasch analysis to explore the fitness of purpose of GCOS-24 within a specialized psychiatric genetic counseling clinic (Borle et al., 2022). However, these studies did not explore the cross-cultural adaptation of GCOS-24 to the Canadian population. While one could make the argument that the populations of Canada and the UK share similarities due to their common language and historical

ties, researchers (Diness et al., 2017) and international guidelines for PROM translation and cultural adaptation (Beaton et al., 2000; Wild et al., 2005) emphasize the importance of appropriately adapting PROMs for similar populations and healthcare systems. Additionally, it should be noted that service delivery in Canada and the UK exhibit slight differences. Despite both being publicly funded, Canadian healthcare services vary across provinces, where each has its own healthcare insurance plan and regulations that determine the scope of practice for different professions. Similarly, genetic tests covered by the healthcare system vary provincially (Costa et al., 2021; Ormond et al., 2018). In Canada, the involvement of clinical geneticists and genetic counselors in the care of patients with genetic conditions can vary between centers and based on specific genetic conditions. In some cases, patients may receive care from either a clinical geneticist or a genetic counselor individually, while in other instances, they may see both professionals together. Contrastingly, in the UK, most genetic counselors practice within regional Genomic Medicine Centres, working together with clinical geneticists as well as autonomously with their own patient load (Ormond et al., 2018).

Considering the global utilization of GCOS-24 as a tool for healthcare quality improvement and its successful cross-cultural adaptations in various countries, it is crucial to explore its adaptation for use in Canadian CGS. While several studies have applied GCOS-24 to a Canadian population, these studies focused on specific applications without exploring its cross-cultural adaptation to the Canadian context. As genetic counseling continues to expand, so does the value of its assessment and using PROMs world-wide. Therefore, the present study aims to answer the following research question: *How can the English language Genetic Counseling Outcome Scale be adapted for use in Canadian Clinical Genetics services?*

The results of this study will thus address the gap in the literature and cross-culturally adapt GCOS-24 for use in Canada, to provide the first step towards an appropriate tool for evaluating CGS in Canada in a patient-centered manner.

2 | METHODS

2.1 | Study design

Qualitative methods were chosen as the study's aim was to discover meaning in the data and explore participants' perceptions and understanding of GCOS-24 (Neuman, 2003). Guidelines on translation and cross-cultural adaptation of PROMs recommend a process consisting of five stages: translation, synthesis, back translation, expert committee review, and pretesting (Beaton et al., 2000; Wild et al., 2005). As the present study aimed to adapt the British tool for use in English-speaking Canada, no translation is required. However, it is recommended that tools are cross-culturally adapted for use in another country other than the country of origin, even if the language is the same (Guillemin et al., 1993). For this reason, stages one to four do not apply to this study. In stage five, qualitative methods are utilized to field

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 The Genetic Counselling Outcome Scale (GCOS-24) (Adapted from McAllister, Wood, et al., 2011).

test the questionnaire of interest to ensure it retains its intended meaning, check participant understanding, and include participant feedback. Semi-structured cognitive interviews were conducted as these have been recommended and used in order to validate cross-cultural equivalence of survey questions (Beck et al., 2017; Willis, 2015).

The study protocol and recruitment materials were given a favorable ethical opinion by the Cardiff University School of Medicine Research Ethics Committee in September 2020 (reference number: SMREC 20/72).

2.2 | Participants

This study targeted the population of Canadian individuals affected with a genetic condition or with a genetic condition in their families. A purposive sampling technique was utilized to come up with target sample guidelines, as it allowed the selection of especially informative participants (Neuman, 2003). Participants eligible for the study included those 18 years or older, fluent in written and spoken English, and member of a Canadian Patient Organization catering to people and families affected by genetic conditions. Recruitment was carried out through the Canadian Organization of Rare Disorders, the Rare Disease Foundation, Ehlers-Danlos Canada, Neuromuscular Disease Network for Canada and the Ontario Rett Syndrome Association. A total target sample size of around 12–15 interviews was expected to reach data saturation (Guest et al., 2020) and comparable to similar studies (Diness et al., 2017; Muñoz-Cabello et al., 2018).

2.3 | Procedures

Data collection between October 2020 and February 2021 was carried out by LR via audio-only one-on-one, semi-structured cognitive interviews conducted through Cardiff University's Blackboard Collaborate, which allowed assessment of participants' comprehension of GCOS-24 items. Utilizing the "Share Screen" feature, LR interviewed the participant as they were reading the GCOS-24 directions and questions for the first time, to obtain the participants' initial reactions and feedback. All interviews were audio recorded using Blackboard Collaborate with participants' written informed consent. Interviews lasted between 60 and 150 min.

An interview guide with open-ended questions was used, adapted from Irwin et al.'s (2009) study, which evaluated a similar PROM instrument (see Appendix S1). In the present study, interview questions were used flexibly, omitting, or adapting them based on participants' needs. Participants were encouraged to "think aloud" while answering the interview questions, in accordance with international guidelines (Beaton et al., 2000; Wild et al., 2005).

2.4 | Data analysis

Audio recordings were transcribed and anonymized using pseudonyms by LR. The transcribed product was subjected to thematic analysis, which identifies themes or patterns within qualitative data (Braun & Clarke, 2006).

As interviews were being held, LR took anonymized field notes to complement the interview transcript. These notes allowed for data analysis to start while the data were being collected. Inductive coding was used to identify major patterns in participants' responses. This approach captures and identifies patterns within the data and allows themes to emerge (Nowell et al., 2017). These codes related to initial thoughts, feelings, possible misunderstandings, and feedback on each GCOS-24 item. The codes were then compared and combined between all datasets and used to develop a participant-response thematic map for each GCOS-24 item. Transcription extract examples were selected to serve as demonstrations of the chosen themes and sub-themes, which in turn relate to the research question. After data analysis, a summary was generated with all GCOS-24 items and their respective participant comments.

3 | RESULTS

3.1 | Sample characteristics and demographic information

It is estimated that a total of ~8807 individuals were invited through Canadian Patient Support Groups' social media platforms and mailing lists. Interest was expressed by 20 individuals (approx. 0.22%). Thirteen respondents provided informed consent and completed interviews. As a result, the estimated response rate for the study was 0.14%. The remaining seven individuals did not respond to the interview invitations and reminder e-mails, and thus did not participate in the study.

Demographic characteristics and clinical characteristics of the 13 participants are displayed in Table 1. To protect confidentiality, participants are identified with the letter P followed by a number which represents the order participants were interviewed. Proof of diagnosis was not required. However, all participants were patient members of Canadian patient organization for individuals with genetic conditions.

3.2 | Cognitive interviews and thematic analysis

Thematic analysis of interview transcripts identified themes related to the study aims of exploring whether GCOS-24 items maintain their intended meaning for Canadians, or whether items could be better worded to meet the needs of Canadians. Data saturation was achieved following the 13th interview, as no new themes were emerging from the data. The research findings on the GCOS-24

TABLE 1 Participant demographics.

Participant	Sex	Condition	Affected/unaffected?	Received or familiar with genetic counseling?	Seen by genetic counselor	Seen by geneticist
P1	Female	Son with SHANK3-related ID	Unaffected	Yes	Yes	Yes
P2	Female	EDS	Affected	Yes	No	Yes
P3	Female	Undisclosed	Affected	Yes	Yes	No
P4	Female	vEDS	Affected	Yes	Yes	No
P5	Female	EDS	Affected	Yes	No	Yes
P6	Female	EDS	Affected	Yes	Yes	No
P7	Female	EDS	Affected	Yes	Yes	No
P8	Female	EDS	Affected	No	No	Yes
P9	Female	Mitochondrial depletion syndrome	Affected	No	No	Yes
P10	Female	MURF	Affected	Yes	Yes	Yes
P11	Female	EDS	Affected	Yes	Yes	No
P12	Female	Myotonic Dystrophy	Affected	Yes	Yes	No
P13	Female	Daughter with Rett syndrome	Unaffected	Yes	Yes	Yes

instructions, response choices and items are outlined below accompanied by illustrative quotes taken from participants' interviews.

3.2.1 | GCOS-24 instructions and Likert scale

All participants (13/13) found the instructions for the questionnaire clear and easy to understand. Participants were asked their opinions on the questionnaire's Likert scale. Specifically, if they believed seven response options were optimal (1 = strongly disagree; 2 = disagree; 3 = slightly disagree; 4 = neither disagree nor agree; 5 = slightly agree; 6 = agree and 7 = strongly agree) or five would be better (removing options 3 and 5). Participants were split in the matter, with 7/13 participants expressing that seven response options were best as they provide both patients and clinicians with more specificity. Further, keeping the seven categories ensures comparability between the Canadian-adapted GCOS-24 and the original tool.

P4: Having variability allows for a more honest answer so that instead of just picking 4 on some things that I'm not quite sure, or whether I agree or disagree, I can go with the slightly. And I think that's more valuable in terms of getting information from me rather than having fewer choices.

3.2.2 | GCOS-24 item semantics

Cognitive control

CGS aim to educate patients in inheritance, condition occurrence/reoccurrence, testing, and management. This information can make participants feel they have regained some control over their lives, or that they have gained as much information about the condition as

they need. Within GCOS-24, items #1, #3, #12, #14, #18, and #23 were designed to capture Cognitive Control.

All items under the Cognitive Control dimension were deemed "easy to understand" by all participants (13/13). When answering "What does this item mean to you?" and "How would you answer this question?" five main themes were prevalent in the transcripts: "Future Plans", "Emotional Response to condition", "Genetic Counselling Impact", "Family" and "Understanding of referral".

The theme of "Future Plans" was present in answers to Item #3—where patients are asked if they understand the impact of the condition on any child(ren) they may have. Participants related this item to obtaining information about the genetic condition that would influence their decision to have children or consider other methods of conception.

P6: ...I guess it would make you reflect on whether you should pursue having children, depending on your condition and how you have been surviving with it. Yeah, it's kind of where my mind would go.

The impact genetic counseling has on participants' answers also emerged as a theme within this dimension, specifically on items that aim to assess participants' understanding of their condition and how it affects their relatives (Items #3, #12 and #18). Participants expressed that genetic counseling would provide them with the information they needed regarding the condition's mode of inheritance and what it entails. They expressed that before genetic counseling, they would probably not know this information, but their answers would change following the consult.

P13 on Item #12: ... I would say pre genetic counselling, we were not sure... we didn't know a lot about it. But post genetic counselling, we felt more

comfortable knowing that this was a spontaneous genetic mutation that caused the disorder.

Additionally, the theme 'Familial Implications of the Condition' emerged from participants' understanding of items #3, #12 and #18. Participants spoke about how the information they receive pertaining to the genetic condition, its nature, and its inheritance pattern, immediately prompts them to think about the implications it can have on other family members.

P11 on Item #3: If you understand the impact of the genetic condition, like you have just been diagnosed with a genetic condition, which would be passed down or could be passed on to your children, do you understand how it will or might impact them?... I like that one.

Another participant, when considering item 18, spoke to the communication of the implications of genetic test results:

P8: We found in my family that it was actually hard to initiate a conversation about it, you know, just to go up to an extended family member and say, 'Hey, I have got this, and it is in the family, you know, genetics and possibly is causing these symptoms? And then maybe you want to look into it'... you don't know how they're going to respond. So... it's difficult. It's difficult.

The last theme for this dimension, "Understanding of Referral", relates specifically to items 1, 14 and 23. Participants spoke to the understanding of their own symptoms that consequently brought them to genetic counseling, as well as their doctor's communication of the referral.

P1 on Item #23: That means that my doctor has... whatever doctor is referring me to genetics, that they have clearly articulated to me their rationale for referring us to genetics.

Further, Participants P1 and P7 expressed that since Items #1 and #14 already addressed patients' understanding of their referral to genetics, there was no need for Item #23.

P7 on Item #23: It's understandable. I'd scrap the whole thing, though.

P1 on Item #23: I wouldn't have the statement there, because that statement has already been made and at the beginning.

Comparing the themes emerging from interview data with the definition of the Cognitive Control dimension adds another layer

within semantic validation to support the participants' claims. The results suggest that this dimension's items maintained their intended meaning within the sample.

Decisional control

Within the empowerment framework, decisional control entails the patient's understanding of options for managing the condition, and the ability to make informed decisions (McAllister et al., 2008). These are not limited to decisions made in the healthcare setting, but also include major or minor life decisions that are influenced by the condition – such as decisions regarding children and marriage. In the GCOS-24 questionnaire, items #10, #13 and #24 were designed to capture decisional control.

All items within decisional control were found "difficult to understand" by a proportion of participants (10/13 participants for item #10; 5/13 participants for item #13 and 3/13 participants for item #24). When answering "What does this item mean to you?" and "How would you answer this question?" four main themes were derived from the transcripts: "Options" "Genetic Counselling Impact", "Empowerment" and "Understanding of conditions".

Within the theme of 'Options' participants who found the items difficult to understand were unsure what options or decisions the items were referring to.

P9 on Item #10: That one I don't really know. What options are you talking? It's unclear because I don't know what options they're talking about.

P8 on Item #24: I do not know what kind of decisions you could make that might change your children's future? Unless it is something whether you're going to have them or not? Or, or when you want to get them tested? Or whether it's about how they're treated early on, or anything like that?

When further prompted to "think aloud" and asked, "How would you answer this question?" participants would then assume or guess that the item was referring to treatment or intervention options available.

P4 on Item 10: I'm not super clear. I'm assuming this is a discussion of 'okay, in this we can do A, B or C.' And I guess if you have a condition that has options that... like for treatment, it like does it mean treatment options?

P1 on Item 24: Is it referring to interventions that may prevent an inheritance of a disorder? Are you talking about monitoring, to watch for complications that arise with the syndrome? Are you talking about them living to their fullest potential? Are you talking about taking preventative action so that, you know if the genetic diagnosis is breast cancer?

Participants also expressed that genetic counseling would have an impact on their answers. Some expressed that they would not know what the question meant before genetic counseling but hoped this would change after the consult.

P2 on Item #10: I'm thinking, 'what options?' You know, so pre, I'm thinking I couldn't answer that... Post? I may be able to answer that. So, I'd probably leave it blank at the time.

The *availability* of treatment/management options also played a role on this impact. P1, who had a diagnosis of EDS, acknowledged that for them, there were not any treatment options. However, there are other genetic counseling subtypes that might include discussion of options during the consult.

P1: It's going to be different for every scenario. I mean, what if this person is going into genetic counselling, because they've had prenatal testing, and they're pregnant, versus medical conditions versus but you know, for us, personally, there are no options, is what it is, and there are no treatments there... I'm going to assume a person would answer this question differently before their counselling appointment and after it.

Although these participants expressed difficulty understanding Item #10, the themes emerging from their interview transcripts suggest that upon further thought, participants 'guessed' the item was referring to treatment/management options. Syntactic changes can be made to this item to address the ambiguity.

The concept of "Empowerment" emerged from the data as a theme within items #13 and #24. Specifically, as item #13 is worded negatively ("Nothing I decide will change the future for my children/any children I might have") some participants strongly felt that "if you have a sick child, you are still going to try and do something."

P13: I feel I could make decisions, right? To change her future and health, therefore, I wasn't going to cure her of her genetic disability or her genetic, you know... but health care provider and you know, my background and my expertise...I did feel empowered, that we would do as much as we could to give her a full and happy life within the constraints of her disability.

Another recurring theme within items #13 and #24 was that of understanding the condition, which prevailed in the transcripts as participants expressed that the information they receive about their condition aids in their decision-making, as well as coping with the situation and its implications in their future.

P10 on Item #24: I can make decisions about my response to the knowledge that I have about the

condition. That's what I can impact. And that may... through my example, I may be able to give my son some better lifestyle kind of options that aren't currently available, right through mainstream medical, through the standard of care, right?

In summary, items within the decisional control dimension presented semantic difficulties due to otherwise common words, such as "options" in item 10 ("I don't know what could be gained from each of the options available to me") and "decide"/"decision" in items #13 and #24 ("In relation to the condition in my family, nothing I decide will change the future of my children/any children I might have"; "I can make decisions about the condition that may change my child(ren)'s future/the future of any child(ren) I may have."). Participants often got stuck in the idea that you cannot control genetics and there are no decisions that can change the future. Although Item #10 caused the most difficulty with 10/13 participants finding it difficult to understand, the overall findings within the dimension suggest that these items did not maintain its intended meaning within a proportion of the sample. Syntactic evaluation might lead to rewording or expanding (through the addition of examples) these items to address these limitations.

Behavioral control

Part of a patient feeling empowered includes their perceived ability to improve the situation arising from their genetic diagnosis. This includes effective use of health and social systems available, communicating genetic risks to people within and outside their families, and managing their condition on a day-to-day basis (McAllister et al., 2008). Clinical Genetics Services values the importance of family communication and signposting, or referring, to other sources of support. This information is often shared with patients during genetic consults to give them the necessary tools to adapt to the genetic condition in their family. Within GCOS-24, items #2, #5, #7, #9, #15, and #16 were designed to capture Behavioural Control.

Items #2, #5, #9, #15, #16, #17, and #22 were all found to be semantically easy to understand by all participants. Item #7 was found to be difficult to understand by participants P1, P2, P4, P7, P8 and P13 (6/13) due to the word 'control'. When answering 'What does this item mean to you?' and 'How would you answer this question?' five main themes were prevalent in the transcripts: 'Medical and Non-Medical Resources', 'Impact of Genetic Counselling', 'Control', 'Emotional Responses,' and 'Communication.'

Within the dimension of behavioural control, participants spoke to the importance of both medical and non-medical resources. Namely, the availability, awareness, and access to these services. Some participants mentioned the lack of medical resources available for their condition as a barrier to care.

P4 on Item #5: For rare conditions that's... that's the rigor, right? Like, there are no doctors. I mean, there are a few, but most people don't have access to them. So, I think for a lot of people going for more rare conditions, this would be an issue that's at the forefront.

Participants mentioned patients' awareness of these resources and indicated that clinical genetic services can play a role in providing these tools/materials.

P6 on Item #5: Yeah, I guess I would just think of whether my condition was treatable, as well. And, whether the geneticist ended up giving me any material to help me find resources probably come in there as well.

As results from other dimensions have showcased, participants were aware of the potential impact genetic counseling can have in how patients communicate about their condition with their families and others, and how they cope with the condition on a day-to-day basis. Participants spoke about how their answers to the items pertaining to Behavioral Control could change before and after genetic counseling:

P2 on Item #22: I think that the answer would be very different pre consult versus post consult, pre consult, I might feel powerless. And post consultation, I may feel like I've gained the knowledge to have some different outcomes for myself and for my family.

The concept of 'control' emerged as a main theme within this dimension. Of particular interest is the semantic difficulty described by participants when reading item #7 due to the word 'control'. Participants expressed how, with genetic conditions, there is an inherent *lack* of control, and nothing can be done to change it, which they believed was what Item #7 was suggesting:

P4 on Item #7: I'm not sure... because I don't know how anyone could control a condition that's genetic. So...a person could control how they tell their family, maybe, but I don't think they could do anything about the actual condition.

Within the 'control' theme, participants also expressed how knowledge, coping strategies and self-advocacy leads to their empowerment as patients:

P13 on Item #7: If my interpretation is accurate, that's... then I think, "can control" that that's me having a choice and choosing my response and either feel empowered or disempowered by this, and feeling, either, you know, "woe is me", or, okay, "let's make lemonade."

Additionally, some participants believed that the only way to control a genetic condition was deciding against having children, adopting, or utilizing other methods of conception.

P12 on Item #7: I can control? Well, yeah, I can choose not to have a kid with the bad egg that I've

got. I mean, ultimately, right? Yeah, I'm in control. I'm kind of in control. I chose to be in control of that.

The theme of emotional responses naturally emerged within the Behavioral Control dimension, specifically when participants were asked about items pertaining to coping with the diagnosis. Participants understood that coping mechanisms would inevitably come to play when living with a genetic diagnosis.

P13 on Item #9: Well, so the word cope, you know, that encompasses, a lot of facets, coping, socially, emotionally, and functionally and physically and all of those. So, that's what I interpret. I do interpret a broader meaning of the word cope.

However, they did not shy away from exploring some difficult feelings that arise on a day-to-day basis, such as guilt and uncertainty.

P2 on Item #7: You know, knowing that...diagnosis can affect families in a big way. And it can be heart-breaking. So, somebody will wonder, what can I do to help change that? And that sort of leads back into an earlier question about control. So, it's like, I don't know what I can do.

Lastly, the theme of "Communication" emerged from participants understanding of items within the "Behavioral Control" dimension. Specifically, items #2 and #16. Participants valued these items as they found it important for patients to talk about their condition both within and outside their families.

P3 on Item #2: I think just having to explain like different symptoms to my family or saying like, this is what I have, like, in my case, my condition has reduced life expectancy, so I can kind of explain that to them.

Item #7 within the Behavioral Control dimension presented semantic difficulties due to the word "control" and participants' perceived notion of its meaning. Like semantic difficulties within the Decisional Control dimension, participants often got stuck with the lack of control individuals have over their genetics. This difficulty can be addressed through syntactic modifications. Other than item #7, no other item within this dimension presented semantic difficulties. The themes emerging from participants' understanding of the items relate to the definition of the dimension, suggesting then that these items maintain their intended meaning within the sample.

Emotional regulation

The Emotional Regulation dimension in the empowerment framework includes the emotional effects a genetic diagnosis can have in a family, and how individuals and their families manage these (McAllister et al., 2008). CGS play a key role in addressing these emotions

and providing tools and support for families to cope. Within GCOS-24, items #4, #11 and #21 were designed to capture Emotional Regulation.

No participant found items #4, #11 and #21 difficult to understand. When answering "What does this item mean to you?" and "How would you answer this question?", three main themes were prevalent in the transcripts: "Genetic Counselling Impact", "Hopelessness" and "Negative Outcomes".

Participants acknowledged the impact genetic counseling can have on the way they feel and think about their genetic diagnosis. The overall sentiment was that the information given in these sessions, such as the explanation on the condition's mode of inheritance, can minimize feelings of anxiety, distress, and guilt.

P13 on Item #11: So, now [post-counselling], okay, but back then we would have answered that I agree that I was... anxious about the future and the unknown... and that was all part of that journey of coming to terms with grieving and coping and, and information gathering... knowledge! You know, educating, you know, myself and informing myself.

A recurring theme within these items was that of hopelessness, which prevailed in the transcripts as participants expressed feeling a lack of control over their condition, and a lack of optimism or "anything good happening" from obtaining a genetic diagnosis. The diagnosis of a genetic condition can cause feelings of distress, participants acknowledged these and the importance of developing coping mechanisms:

P4 on Item #4: ...Understanding the consequences in terms of how it's going to affect people, including myself. Because rarely is it like, 'oh, you're going to win a million dollars.' That's what your DNA says. It's never something lovely like that, right?

Contrastingly, some participants utilized the lack of control an individual has over their genetics as a reassuring piece to alleviate both their own feelings of guilt and that of others in their family.

P8 on Item #21: Um... in in, I'm thinking in my case, it doesn't apply but I was thinking also of my mom. And the fact that she always says, 'Oh, I feel guilty because look at you girls', about me and my sister, and I think you know, you're my mom and we choose to be born and don't feel guilty.

All participants spoke to the negative emotions that can be present throughout the process of receiving a genetic diagnosis. When asked what the item meant for them, participants drew from their personal experiences with their diagnosis and explored what they have felt throughout the process.

P7 on Item #4: I think it's certainly understandable. I mean, there's a lot of guilt and shame that can go along with the idea of passing down and fear for family members, if others are affected. And yeah, there's a lot of unpleasant emotions tied to that.

Participants also expressed how these items triggered an emotional response, making them "feel guilty all over again" or feel that they "should be upset".

P2 on Item #21: ...having to answer the question makes me feel guilty all over again. It is a heavy burden, right? I... in my personal case, I had no idea I had EDS or would develop the severity or pass it on to my child.

All participants (13/13) expressed the items under Emotional Regulation were easy to understand. Participants spoke to the challenging emotional aspects that come from the diagnosis of a genetic condition, highlighting the importance of obtaining more information and developing coping mechanisms. Consequently, when comparing the themes emerging from interview data with the definition of the Emotional Regulation dimension, the results suggest that this dimension's items maintained their intended meaning within the sample.

Hope

The empowerment dimension of 'Hope' entails patients' ability to look forward to the future, hopeful for a fulfilling life for themselves, their family members and their future descendants (McAllister et al., 2008). Within GCOS-24, items #6, #8, #19 and #20 were designed to measure Hope.

No participant found items #8 and #20 difficult to understand. Item #6 was found to be difficult to understand by participants P1, P4 and P10 (3/13) due to the phrase "good things". Item #19 was found to be difficult to understand by participants P1, P2, P4, and P8 (4/13) due to the phrase 'rewarding family life'. When answering "What does this item mean to you?" and "How would you answer this question?" three main themes were prevalent in the transcripts: 'Feelings of Hope', 'Tools' and 'Negative Outcomes'.

'Feelings of Hope' was a prevailing theme within this dimension, which was demonstrated in some capacity throughout all transcripts. When referring to hope, participants spoke about ensuring they provide their children a rewarding and happy life, the hope for treatment feeling empowered, and being able to cope with the implications of the condition:

P3 on Item #8: Do you feel optimistic? Like, do I feel... for the future there's going to be a cure, that I'll get a treatment, that I'll get better? That things won't happen?

These findings directly relate to the definition of the "Hope" dimension, which highlights the need for patients to have hope

for the future: not only for themselves but also for their family members.

A recurring theme within the Hope dimension was the understanding that when coping, managing, and making plans for the future, individuals with genetic conditions utilize various tools to feel empowered. Participants often made mention of being aware of resources available to cope with the condition in a healthy way and make plans for the future. Further, participants appreciate the power of information and further knowledge of their condition to make informed decisions.

P8 on Item #20: Um, I guess that you feel capable or confident in, in making plans or that you, you know, you have access to the resources you need to make plans? It's fairly broad, I suppose. Because I don't know what kind of plans it means. I guess it just means looking ahead. Do you feel... Do you feel okay, thinking ahead, that you're going to be okay?

Within the conversations of the hope and plans for the future, participants believed that although a positive mindset and access to tools can aid in coping and planning, the negative outcomes stemming from a genetic diagnosis still play a part. Most notably, some participants expressed feelings of uncertainty when answering items such as items #19 and #20, which specifically speak to the patient's ability to make plans for the future:

P7 on Item #20: Oh, goodness, like, I don't know what day I'm going to go anaphylactic. So, no. I mean, I hope so. But yeah, I mean, my husband and I have discussed this, that we think about things very, very differently, because he assumes, he will get old and I'm genuinely shocked to have made the age 40.

When speaking on Item #6 specifically, all participants spoke about negative emotions that can be present throughout the process of receiving a genetic diagnosis, namely feelings of distress. Item #6, "I can see that good things have come from having this condition in my family" came as a surprise to participants, as "nothing good comes from a genetic condition". Syntactic changes to the item might address these initial reactions, which might have prompted some participants (3/13) to classify the item as "difficult to understand."

P10 on Item #6: Well, what good has come from having EDS, in the family? I just, I put in my thing, so I can relate to it. I don't see any good. I don't know why you'd ask that question. Is there anything good coming from any condition that is genetic?

Items #6 and #19 were found to be difficult to understand by 3/13 participants and 4/13 participants, respectively. The semantic difficulties arising from these items pertained to the phrases

"good things" ("I can see that good things have come from having this condition in my family") and "rewarding family life" ("I am hopeful that my children can look forward to a rewarding family life"). These difficulties can be addressed through syntactic modifications. Once these are implemented, the understanding of the dimension within the sample will be consistent with the original intended meaning. The remaining items (#8 and #20) were semantically valid.

3.2.3 | Syntactic modifications to GCOS-24 items

During the cognitive interviews, participants were asked "How would you change the words to make it [the item] clearer?". This allowed participants to suggest syntactic changes, which, to their understanding, would enhance comprehension of the item. Table 2 includes proposed changes to the original GCOS-24 based on participant feedback and discussion with MM, original creator of the tool, to ensure items maintained their intended meaning.

3.2.4 | Participants' overall thoughts on GCOS-24

Before finalizing the cognitive interviews, participants were asked to provide an overall assessment of the questionnaire. Six out of 13 participants mentioned they believed GCOS-24 was a very useful tool, "very encompassing" and "thoughtfully looking at several factors from different perspectives":

P4: I think that that's thoughtfully kind of looking at several factors, from different perspectives and different questions. And I, and I think in terms of assessing how much or little the counselling impacted the patient, that it's, it's pretty strong in that regard. In my lay persons assessment.

Additionally, 9/13 participants reiterated the feedback they had provided across the interviews. For instance, several participants mentioned how items needed to be more specific and that the language used was too broad:

P1: So that would be my only other feedback is to be careful of the language and then to be specific, what your statement is really, the intent of your statement actually is so that you can get a better answer. It'll get a little closer to black and white in something that's really grey.

Further, participants emphasized the need to change "I don't know" items to "I know", as this would make the item easier to understand and answer:

TABLE 2 Proposed changes to GCOS-24 items.

#	Original GCOS-24 item	Proposed change for the Canadian-adapted GCOS questionnaire	Rationale
1	I am clear in my own mind why I am attending the Clinical Genetics Service.	I understand why I am attending genetic counseling .	Both "I am clear in my own mind" and "Clinical Genetics Service" presented syntactic difficulties for 12/13 participants. To our knowledge, the proposed changes address these without affecting the meaning of the item.
2	I can explain what the condition means to people in my family who may need to know.	I can explain what the condition means to people in my family who may need to know.	No change.
3	I understand the impact of the condition on my child(ren)/any child I may have.	I understand the possible impact of the condition on my child(ren)/any child I may have.	2/13 participants believed the item was too declarative and suggested the addition of "possible" before the word "impact". Although other 4/13 participants suggested syntactic changes to this item, these were thought to change the item's intended meaning.
4	When I think about the condition in my family, I get upset.	When I think about the condition in my family, I get upset.	No change.
5	I do not know where to go to get the medical help I/my family need(s).	I know where to go to get the medical help I/my family need(s).	7/13 Participants found items beginning with "I do not know" difficult to answer. For this reason, these have been changed to "I know".
6	I can see that good things have come from having this condition in my family.	I can see that good things (for example, personal growth) have come from having this condition in my family.	All participants, regardless of if they found the item difficult to understand or not, expressed the item made them wonder what good comes from having a genetic condition. The intended meaning for "good things" includes many aspects, such as personal growth. This was not clear to participants and the suggested changes proposed by 3/13 of them do not address this. Providing an example might address this limitation.
7	I can control how this condition affects my family.	I can control how this condition affects my family physically, medically, emotionally, and/or financially .	7/13 participants suggested the item needed to be more specific and expand on what "control" meant. The addition of examples might make the item easier to understand.
8	I feel positive about the future.	I feel positive about the future.	No change.
9	I am able to cope with having this condition in my family.	I can cope with having this condition in my family	4/13 participants suggested syntactic changes to item #9. Participant P10 proposed keeping the item simple and straightforward. This suggested change was preferred as it was thought not to change the intended meaning of the item.
10	I do not know what could be gained from each of the options available to me.	I know what could be gained from each of the options available to me (for example, genetic testing or treatment/management options).	10/13 participants found item #10 difficult to understand due to the lack of specificity on what the word "option" entails. Adding "genetic testing or treatment/management options" as examples might address this difficulty without potentially changing the intended meaning of the item.
11	Having this condition in my family makes me feel anxious.	Having this condition in my family makes me feel anxious.	No change. This item was found easy to understand by 13/13 participants.
12	I do not know if this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	I know how this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	7/13 participants found items beginning with "I do not know" difficult to answer. For this reason, these have been changed to "I know".
13	In relation to the condition in my family, nothing I decide will change the future for my children/any children I might have.	Nothing I decide about this condition will change the future for my children/any children I might have.	8/13 participants suggested syntactic changes to Item #13. 4/13 suggested simplifying and shortening the item. The preferred change addresses this and, to our knowledge, keeps the item's intended meaning.
14	I understand the reasons why my doctor referred me to the clinical genetics service.	I understand the reason(s) why my doctor referred me for genetic counseling .	Similar to item #1, "Clinical Genetics Service" was changed to "genetic counseling". Participant P1 suggested adding parentheses around the "s" to make the term both singular and plural. These suggested changes were preferred as they were thought not to change the intended meaning of the item.

TABLE 2 (Continued)

#	Original GCOS-24 item	Proposed change for the Canadian-adapted GCOS questionnaire	Rationale
15	I know how to get the non-medical help I/my family needs (e.g., educational, financial, social support).	I know how to get the non-medical help I/my family needs (for example, educational, financial, social support).	3/13 participants suggested syntactic changes to Item #15. Participant P7 suggested removing "e.g.," to encompass ESL populations. This suggested change was preferred as it likely would not change the intended meaning of the item.
16	I can explain what the condition means to people outside my family who may need to know (e.g., teachers, social workers).	I can explain what the condition means to people outside my family who may need to know (for example, teachers, social workers).	3/13 participants suggested syntactic changes to Item #15. Participant P7 suggested removing "e.g.," to encompass ESL populations. This suggested change was preferred as it likely would not change the intended meaning of the item.
17	I do not know what I can do to change how this condition affects me/my children.	I know what I can do to change how this condition affects me/my children	7/13 participants found items beginning with "I do not know" difficult to answer. For this reason, these have been changed to "I know".
18	I do not know who else in my family might be at risk for this condition.	I know who else in my family might be at risk for this condition.	7/13 participants found items beginning with "I do not know" difficult to answer. For this reason, these have been changed to "I know".
19	I am hopeful that my children can look forward to a rewarding family life.	I am hopeful that my children can look forward to a fulfilling life .	"Rewarding family life" was unclear to 8/13 participants. No specific change was suggested to the item due to participants' lack of understanding. Removing "rewarding family life" from the item might increase understanding.
20	I am able to make plans for the future.	I am able to make plans for the future.	No change.
21	I feel guilty because I might have passed this condition on to my children.	I feel guilty because my child(ren) might have inherited the condition.	3/13 participants suggested syntactic changes to Item #21. The phrase "I feel guilty because I might have passed this condition on..." was considered to give a "sense of responsibility." The preferred change addresses this and should keep the item's intended meaning.
22	I am powerless to do anything about this condition in my family.	I am powerless to do anything about this condition in my family (that is, physically, emotionally and/or medically).	4/13 participants suggested Item #22 needed to be more specific. The addition of examples might address this difficulty without potentially changing the intended meaning of the item.
23	I understand what concerns brought me to the clinical genetics service.	ITEM REMOVED	Participants found the item repetitive when compared to items #1 and #14. For this reason, the item has been removed from the Canadian adapted version of GCOS-24.
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.	I can make decisions (physical, medical, emotional, and/or financial) about the condition that may change the future of any child(ren) I may have .	8/13 participants proposed semantic changes to item #24. The removal of "my child(ren)'s future" was suggested by participants P1, P7 and P12. Further, the addition of examples might provide more specificity. These suggested changes were preferred as they likely do not change the intended meaning of the item.

Note: Bolded words indicate syntactic changes to the item.

P10: No, I mean, overall comments, I think I've been clear that remove the negatives and replace them with a positive would be helpful, I think, in a lot of cases.

there should be one like for people who can and who can't.

Lastly, participants who could not have children, found some items did not apply to them. Due to the number of items participants found to not be applicable to them, they suggested the creation of a different questionnaire for individuals who cannot have children. This feedback reinforces the need to add a N/A response choice:

P3: Maybe there should be like different ones, just because, like, some questions didn't really apply to me. Just because, like, I can't have kids, so like, maybe

3.3 | Summary

Results for the GCOS-24 instructions and response choices showed that (a) the instructions were clear to understand (aside from a formatting suggestion), (b) a 7-item Likert scale is preferred by most participants (7/13) and ensures comparability between the original tool and the Canadian-adapted version, and (c) the tool would benefit from a N/A response choice. The themes presented within each "Empowerment" dimension describe in detail participants' semantic

and syntactic understanding of each GCOS-24 item and their suggested item modifications. Following further analysis of the results (including participants' overall feedback) and discussion with MM to ensure that, to our understanding, no edits changed the original meaning of the item, a Canadian-adapted version of the GCOS-24 items, GCOS-Canada, was developed. Figure 2 presents the final version of GCOS-Canada.

4 | DISCUSSION

This is the first study to cross-culturally adapt the GCOS-24 for use in Canada. Its objectives were to explore whether the British GCOS-24 maintained its intended meaning in a Canadian population, and whether the items could be better worded to meet the needs of families affected by genetic conditions in Canada. Only five studies identified through our search utilized GCOS-24 in Canada and did so without cross-cultural adaptation of the tool (Borle et al., 2022; Gerrard et al., 2020; Inglis et al., 2015; Morris et al., 2019; Yusuf et al., 2021). This study not only addresses the gap in the literature but also provides the first step towards a Canadian-adapted version of GCOS-24 that, once psychometrically validated, can be used confidently in Canadian clinical genetic services and research. Thirteen members of families affected by genetic conditions participated in semi-structured cognitive interviews to assess understanding of GCOS-24 instructions, response options and items.

4.1 | Instructions and response choices

The number of response choices were maintained in most items based on participant feedback. This contrasts with the findings by Borle et al. (2022), who used Rasch measurement theory to explore GCOS-24's fitness for purpose within a specialized psychiatric genetic counseling service in Vancouver, Canada. The study found that the 7-item Likert scale did not work as intended for 23/24 items, with participants not being able to distinguish between the lower end of the scale (1–4) and the middle (5 and 6). As a result, a three-category response option was more favorable for this sample. Borle et al.'s (2022) findings are limited due to the study sample homogeneity, as participants were recruited from a single specialized center in one Canadian city, and the retrospective methodology, which did not allow ascertainment of any patient feedback on proposed changes to the scale. Although the present study's changes to the GCOS-24 are based on patient feedback, the sample is not representative of the entire Canadian population. This highlights an opportunity for future research on the validity of GCOS-Canada with a larger, more representative sample.

One important difference when compared to the original GCOS-24 is the addition of a “not applicable” response option to items asking about children as a result of participant feedback. This was not previously added because there was concern that it might contribute to missing data, which in turn would influence the psychometric

properties of the instrument if measured using classical test theory. As a result, participants were instructed to mark a neutral response, ‘4,’ if the item was not applicable to them. However, Yuen et al. (2020) recently psychometrically evaluated the Singapore-adapted GCOS-24 using Rasch analysis and found this method does not require complete data to generate measure estimates. In fact, the addition of a “not applicable” option to items relating to children improved the psychometric properties of the Singapore-adapted GCOS-24, as participants did not need to answer items that were not relevant to them (Yuen et al., 2020). These findings re-emerge in a different context within Grant et al. (2019)'s study, which aimed to develop a short form of GCOS-24, the Genomics Outcome Scale (GOS). This study found an increased peak in “Non-applicable” responses in GCOS-24 items that asked about children (Grant et al., 2019). Together, these findings reflect the reality that not all patients attending CGS have children, and, as the items within GCOS-24 cater to encompass the wide population seen in these services, the response options would benefit from the addition of a N/A option.

4.2 | GCOS-24 items

It was critical that the psychometric properties of GCOS-24 were retained. Syntactic modifications to the original scale incorporated participant feedback and further discussion with MM, creator of the original tool. However, there was limited room for modifications within the items on the scale—if the suggested change altered the meaning of the item, it was not modified. Only adjustments, additions and simplifications not found to alter the meaning of the item were made.

Semantic validation of the GCOS-24 showed that items within the Decisional Control dimension (items #10, #13 and #24) were not found to be easy to understand by a proportion of participants. This can be due to syntactic aspects, such as a lack of specificity and/or clarity, which the present study has aimed to address through rewording of the items. These results are concordant with other GCOS-24 cross-cultural validation and translation studies. Specifically, item #10 (‘I don't know what could be gained from each of the options available to me’) was found to be difficult to understand by multiple studies in different settings: Spain, Brazil and Singapore. These studies report that their participants found the item “unclear and inconsistent” and did not understand what the item meant by “options” (Muñoz-Cabello et al., 2018; Segundo-Ribeiro et al., 2020; Yuen et al., 2020).

Within the dimension of Decisional Control, participants expressed the same limitations in understanding item #13 (‘In relation to the condition in my family, nothing I decide will change the future for my children/any children I might have’), often wondering what “decisions” can be made that would affect the future. This finding is novel to this study, as other GCOS-24 cross-cultural adaptation and translations studies have only found difficulties with the negative sentence stem of item #13 (Muñoz-Cabello et al., 2018). Interestingly, the study by Muñoz-Cabello et al. (2018) did not modify the

The Canadian Genetic Counselling Outcome Scale (GCOS-Canada)

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.

Please only choose option 8, not applicable (N/A), for questions 3, 13, 19, 21 and 24 if you do not have children and do not intend to.

1 = strongly disagree
 2 = disagree
 3 = slightly disagree
 4 = neither agree nor disagree

5 = slightly agree
 6 = agree
 7 = strongly agree
 8 = not applicable (N/A)

strongly disagree disagree slightly disagree neither agree nor disagree slightly agree agree strongly agree not applicable

		strongly disagree	disagree	slightly disagree	neither agree nor disagree	slightly agree	agree	strongly agree	not applicable
1	I understand why I am attending genetic counselling.	1	2	3	4	5	6		
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 possible impact of the condition on my child(ren)/any child I may have.	1	2	3	4	5	6	7	8
4	When I think about the condition in my family, I get upset.	1	2	3	4	5	6	7	
5	I 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 (for example, personal growth) 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 physically, medically, emotionally, and/or financially.	1	2	3	4	5	6	7	
8	I feel positive about the future.	1	2	3	4	5	6	7	
9	I can cope with having this condition in my family.	1	2	3	4	5	6	7	
10	I know what could be gained from each of the options available to me (for example, genetic testing or treatment/management options).	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 know how this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	1	2	3	4	5	6	7	
13	Nothing I decide about this condition will change the future for my children/any children I might have.	1	2	3	4	5	6	7	8
14	I understand the reason(s) why my doctor referred me for genetic counselling.	1	2	3	4	5	6	7	
15	I know how to get the non-medical help I / my family needs (for example, 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 (for example, teachers, social workers).	1	2	3	4	5	6	7	
17	I know what I can do to change how this condition affects me / my children.	1	2	3	4	5	6	7	8
18	I 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 fulfilling life.	1	2	3	4	5	6	7	8
20	I am able to make plans for the future.	1	2	3	4	5	6	7	
21	I feel guilty because my child(ren) might have inherited the condition	1	2	3	4	5	6	7	8
22	I am powerless to do anything about this condition in my family (that is, physically, emotionally and/or medically).	1	2	3	4	5	6	7	
23	I can make decisions (physical, medical, emotional, and/or financial) about the condition that may change the future of any child(ren) I may have.	1	2	3	4	5	6	7	8

FIGURE 2 Final Canadian adaptation of GCOS-24: GCOS-Canada.

item to a positive statement, suggesting that modifying the item to a positive stem results in the loss of its intended meaning (Muñoz-Cabello et al., 2018). Likewise, no other GCOS-24 cross-cultural adaptation study has reported a difficulty in understanding item #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'), which makes the Decisional Control dimension being difficult to understand unique to this study. These findings could be attributed to all participants in the current study being either affected or having a child affected by rare genetic conditions, where treatment and management options are scarce. Studies have found that these individuals have lower PPC when compared to individuals with more manageable, common conditions, which might explain the lack of understanding of items pertaining to the availability of options and decision-making (Lipinski et al., 2006).

Interestingly, Grant et al. (2019) found that participants did not perceive items within the Decisional Control dimension as being of high value, resulting in no items pertaining to this dimension being included in the GOS (Grant et al., 2019). The cognitive interview sample from Grant et al. (2019)'s study and the present study are similar in that participants are affected by rare conditions, further highlighting the suggested trend. Decisional Control outcomes, such as decision-making, have been demonstrated to be of value to CGS patients (Macleod et al., 2002; McAllister et al., 2008). Further research is needed to test the reworded version of these items within and beyond this participant population.

This study's findings also support other published evidence regarding other dimensions of the Empowerment construct. The results pertaining to the Behavioral Control dimension suggest an overall semantic understanding of all items except for #7 ('I can control how this condition affects my family'), which was deemed difficult to understand by 6/13 participants. The use of the word "control" was received with confusion – similar to the findings on item #24 (and the word "decisions")—due to participants' "lack of control" over their condition. Muñoz et al. (2018), when translating and cross-culturally adapting GCOS-24 for use in Spain, also found participants did not understand what was meant by "control" in item #7, with one participant expressing that the item seems to imply an individual is in full control, when with genetic conditions, many things can feel out of control (Muñoz-Cabello et al., 2018). When developing items for use in health measurements, non-specific words and ideas can produce confusing ambiguity, while strong words such as "control" can bias the results. GCOS-24 aims to cater to a wide population of individuals with different reasons for attending genetic counseling, and thus its items need to be general enough to do this. The resulting Canadian adapted GCOS-24 hopefully addresses the difficulties present with the addition of examples.

Two items within the Hope dimension (items #6 and #19) also presented semantic difficulties within the present study's sample. Item #6 ('I can see that good things have come from having this condition in my family') was found difficult to understand as participants did not comprehend what "good things" meant. This finding was also identified in several previous studies cross-culturally adapting and

translating GCOS-24 for use in Singapore, Spain, and Denmark, respectively. Yuen et al. (2020), modified item #6 to include examples of "early detection and personalized screening" for a better understanding of what "good things" might refer to (Yuen et al., 2020). However, Yuen et al. (2020)'s adapted version was developed solely for use within a cancer genetics context. Comparably, Muñoz et al. (2018) took a different approach to enhance comprehension of item #6, by modifying it to a negative: "I can't see that good things have come from having this condition in my family" (Muñoz-Cabello et al., 2018). On the other hand, a study by Diness et al. (2017) found no indication that item #6 was felt to be distressing by their study sample but did highlight the emerging theme of participants understanding the item as "implying something good coming from the genetic condition" (Diness et al., 2017).

Interestingly, all dimensions except that of Hope had the theme of "Genetic Counselling Impact" emerging from participants' transcripts – regardless of semantic understanding. Within the interviews, all participants acknowledged at one point or another that genetic counseling would have an impact on their answers to each of the GCOS-24 items. The published evidence reviewed highlighted the consistent use of a restricted number of outcomes (i.e., information recall, satisfaction, reduced anxiety) – which do not encompass all the available benefits of using CGS. The present study builds upon the extensive qualitative research done throughout the development of GCOS-24 (McAllister et al., 2008; McAllister, Dunn, & Todd, 2011; McAllister, Wood, et al., 2011), and further demonstrates that the tool captures outcomes that are valued and found to be relevant by the patient population utilizing CGS. Within the "Hope" dimension, participants mentioned awareness of tools, resources, and information aiding in their planning for the future. However, no participant linked this back to the information provided in genetic counseling. Previous studies show conflicting evidence regarding this dimension in particular, with Costal-Tirado (2017) finding a significant increase in post-appointment GCOS-24 scores within items pertaining to this dimension (Costal Tirado et al., 2017), and Yuen et al. (2020) notably finding that genetic counseling had little to no impact on participants' feelings of hope (Yuen et al., 2020). It is difficult to say if syntactic modifications will address and impact these findings, which highlights the importance of psychometric validation of the Canadian-adapted tool. However, this does pose questions about different genetic counseling service-delivery models that might not focus on emotional support or hope-based interventions.

Another important difference is the reduction of items compared to the original GCOS-24: 23 instead of 24. Item #23 ("I understand what concerns brought me to the clinical genetics service") was deemed repetitive by participants, as items #1 and #14 also address participants' understanding of their referral. This finding is unique to the present study when compared to other cross-culturally adapted and translated GCOS-24 studies. Further, there was an overall participant confusion over items beginning with "I don't know", which has also been found through the published evidence (Diness et al., 2017; Grant et al., 2019; Muñoz-Cabello et al., 2018). Similarly, the study

by Borle et al. (2022) exploring GCOS-24 purpose of fit using Rasch analysis in a Canadian psychiatric genetic counseling service found that removing items 4, 11, 12, 17 and 18 showed excellent fit to the Rasch model, good reliability, and responsiveness to change. A large proportion of these items are negatively worded, which the authors believed attributed to their poor performance. These findings highlight one of the potential biases that may arise within questionnaire respondents, which is that of “acquiescence”, or participants' tendency of giving positive responses. Guidelines suggest that in order to minimize such response set, questionnaires can include a balance of positive and negative items (Streiner et al., 2015). The same guidelines advise caution when using negative sentence stems, as participants can find the concept of disagreeing with an item to indicate a positive answer complicated – which is what was seen within the present study's sample. The resulting Canadian-adapted GCOS-24 incorporates a balanced approach to minimizing acquiescence, by retaining two negative stem items that did not present semantic difficulties within the sample: Item #13 and Item #22, but also modifying the “I do not know” items to “I know” as these did present difficulties when participants attempted to answer them.

4.3 | Implications to practice

The adaptation of this tool to a Canadian population presents an opportunity for CGS to be evaluated using a valid outcome measure once the tool undergoes psychometric validation. A study by Costa et al. (2021) examined the Canadian genetic counseling workforce, finding an increased demand for genetic counselors in Canada, but the presence of funding barriers prevent clinics from creating additional positions. In Canada, there is public funding of all healthcare services, including CGS. As a result, the majority of genetic clinics rely on funding from the provincial government, which can present an inherent drawback, as provincial budgets are limited (Costa et al., 2021). Value-based healthcare is becoming a leading approach to budget allocation within the Canadian healthcare system (Prada, 2016). This method links how much money is spent on programs or services to the outcomes that matter most to patients. Consequently, the national implementation of CGS evaluation utilizing validated PROMs could support the economic evaluation and budget allocation of these services (Baumhauer & Bozic, 2016).

4.4 | Implications to further research

The present study's findings and its resulting item modifications reinforce the need to cross-culturally adapt PROMs to provide the most appropriate tool for patient-centred evaluation of CGS. The methodology used was in accordance to published guidelines and thus ensures the semantic equivalency of the Canadian-adapted GCOS-24 to the original tool (Beaton et al., 2000; Wild et al., 2005). However, this is only the first step in cross-culturally adapting a self-report measure – further research is needed to preliminarily test the

internal consistency of the GCOS-24 adaptation with a large sample representative of the Canadian population. Further, future research could benefit from utilizing Rasch analysis to assess the psychometric validation of the tool (Petrillo et al., 2015). This method will ensure that no missing values emerge from the addition of the N/A option, compared to classical test theory. A comprehensive psychometric validation of the tool including include test-retest reliability, responsiveness, establishment of the MCID, as well as structural and construct validity would aid in establishing the Canadian-adapted GCOS-24 as a useful tool to evaluate clinical genetics services in Canada. Lastly, the official languages of Canada are English and French. Cross-cultural adaption and translation of GCOS-24 for use in French-speaking Canada is needed to encompass all CGS within the country.

4.5 | Strengths and limitations

An important strength of the current study is that data saturation was achieved. By the 13th interview no new themes were emerging, supporting the validity of the results. Prolonged engagement with the data, audit trails of documents pertaining to codes, records of all data field notes and peer debriefing between LR and MM further increase the study's credibility and validity. Confirmability of the findings was demonstrated with transcript excerpts supporting each point within the Results section, and clear explanations behind any choices made throughout the entire study.

A main limitation of this study is that of sample homogeneity. All participants (13/13) were female and affected by or had a child with a rare genetic condition. Additionally, information on factors such as age, race, education level and socioeconomic status were not collected. The findings are therefore limited to the perspective of this population and do not reflect the views and opinions of all Canadians from families affected by genetic conditions. An additional limitation to the external validity of this study is the potential sample bias, as participants were recruited from Canadian Patient Organizations and these individuals are more engaged with research than most. Further research is needed to test the Canadian-adapted GCOS-24 in the wider Canadian population. Specifically, research is needed within the “at-risk” for, but themselves unaffected by a genetic condition, population—these individuals are a key demographic in CGS and were not represented in this study.

5 | CONCLUSION

This study's findings have successfully addressed the research question of: “How can the English language Genetic Counselling Outcome Scale be adapted for use in Canadian Clinical Genetics services?” By answering this question, the study aimed to cross-culturally adapt GCOS-24 for use in Canadian CGS. Through cognitive interviews and thematic analysis, semantic validations and syntactic modifications were made that resulted in the Canadian-adapted tool. Despite

the limitations, the findings of the study contribute to future practice and research, as it is the first step in providing a valid, culturally adapted outcome measure for use in CGS service evaluation, quality care provision and budget allocation. Future research with a larger and more representative sample size could psychometrically validate the tool to ensure validity, reliability, responsiveness, MCID and internal consistency. Since Canada is a bilingual country, further translation, and cross-cultural adaptation of GCOS-24 to the French-Canadian population is necessary.

AUTHOR CONTRIBUTIONS

Laura Redondo and Marion McAllister confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All authors gave final approval of this version to be published and agree 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.

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CONFLICT OF INTEREST STATEMENT

Authors LR and MM declare that they have no conflict of interest.

DATA AVAILABILITY STATEMENT

Data collected for this qualitative study are not available to be shared publicly to protect the participants' confidentiality.

ETHICS STATEMENT

Human studies and informed consent: Ethics approval to conduct this qualitative research was obtained by Cardiff University School of Medicine Research Ethics Committee in September 2020 (reference SMREC 20/72). Informed consent was obtained from all participants for being included in the study.

Animal studies: No non-human animal studies were carried out by the authors for this article.

ORCID

Laura Redondo  <https://orcid.org/0009-0008-3528-6973>

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SUPPORTING INFORMATION

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

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