BRIEF REPORT

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Assessing sensitivity to change of the genomics outcome scale (GOS)

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Abstract

The Genetic Counseling Outcome Scale (GCOS-24) is a 24-item patient-reported outcome measure (PROM) that was developed to evaluate genetic counseling and testing services by measuring the construct of empowerment. The Genomics Outcome Scale (GOS) is a 6-item version of GCOS-24 that was designed to provide a PROM for use both within and outside clinical genetics services and reduce respondent burden. However, unlike GCOS-24, the sensitivity to change of the GOS has not yet been assessed in appropriate clinical settings. We carried out pre- and post-clinic surveys using the GOS to assess sensitivity to change of the GOS and produce before-and-after GOS data as part of a service evaluation. The survey was sent to patients attending the genetic counseling clinic for a first appointment at the All Wales Medical Genetic Service from 8 April 2019 to 18 September 2019. Patients attending disease management clinics, where genetic issues were not the primary concern, were excluded from this study. A total of 138 respondents were included in the final analysis. The result shows that empowerment scores, measured using the GOS, were significantly higher (p<0.05) after clinic attendance. The GOS shows good sensitivity to change, with a medium-to-large effect size (Cohen's $d = 0.73$). The result also shows that the service is delivering measurable benefits for its service users.

KEYWORDS

communication, genetic counseling, genetics services, patient-reported outcome measure

What is known about this topic

The Genomics Outcome Scale (GOS) is a six-item questionnaire that was designed as a shorter version of GCOS-24 to provide a PROM that can be used both within and outside of clinical genetics services and reduce respondent burden. Correlation between the two scales demonstrated that GOS maintains the ability of GCOS-24 to capture the construct of empowerment ($r = 0.838$).

What this paper adds to the topic

This study has shown good sensitivity to change of GOS, with a medium-to-large effect size (Cohen's $d = 0.73$). It also demonstrates that the All Wales Medical Genetics Service is delivering measurable benefits to its service users.

1 | INTRODUCTION

Patient-reported outcome measu (PROM) are increasingly implemented in healthcare systems to evaluate the effectiveness and quality of care (Meadows, 2011). The Genetic Counseling Outcome Scale (GCOS-24) is a 24-item PROM that was developed to evaluate genetic counseling and testing services (McAllister et al., 2011). GCOS-24 was designed to measure the construct of empowerment, comprising five sub-dimensions of outcomes valued by patients: cognitive, decisional and behavioral control, emotional regulation, and hope (McAllister et al., ,2010, 2011). Each item on the GCOS-24 is rated on a 7-point Likert scale (1 = strongly disagree, 7 = strongly agree) (McAllister et al., 2011). Scores range from 24 to 168 with higher scores indicating higher levels of empowerment (McAllister et al., 2011). GCOS-24 has been demonstrated to have good test–retest liability, sensitivity to change, and construct validity. (McAllister et al., 2011)

GCOS-24 has been translated into Danish, Spanish, Dutch, and Brazilian Portuguese (Diness et al., 2017; Muñoz-Cabello et al., 2017; Segundo-Ribeiro et al., 2020; Voorwinden et al., 2019). It is used extensively in clinical and research settings in both the UK and internationally. GCOS-24 was used in six of the 25 UK regional clinical genetics service in 2011–12 as part of a service evaluation exercise (McAllister, 2016). It has also been used to evaluate specialist genetic services, including psychiatric, cardiovascular, and cancer genetic services (Inglis et al., 2015; Ison et al., 2019; Yuen et al., 2020).

The Genomics Outcome Scale (GOS) was designed as a shorter version of GCOS-24 to provide a PROM that can be used both within and outside of clinical genetics services and reduce respondent burden (Grant et al., 2019). GOS was created through qualitative cognitive interviews, analysis of an existing data set of GCOS-24 responses using Samejima's Graded Response Model (GRM), and Rasch modelling (Grant et al., 2019). Correlation between the two scales demonstrated that GOS maintains the ability of GCOS-24 to capture the construct of empowerment ($r = 0.838$) (Grant et al., 2019).

However, unlike GCOS-24, the construct validity and sensitivity to change of the GOS have not yet been assessed (Grant et al., 2019).

This study aims to (a) assess sensitivity to change of GOS and (b) provide before-and-after GOS data to the NHS All Wales Medical Genetics Service (AWMGS) as part of a service evaluation.

AWMGS is a clinical genetics service offering genetic counseling and testing in families where a genetic condition is suspected or known to be present, including autosomal dominant, recessive and X-linked conditions, learning disability, chromosome abnormalities, and developmental delay. Patient may be seen by a clinical geneticist or a genetic counselor. Previous research has demonstrated that patients attending AWMGS experience increased level of empowerment following clinic attendance, as captured by GCOS-24 (Costal Tirado et al., 2017), and so we would expect that if GOS is sensitive to changes in empowerment, that GOS would also capture increased level of empowerment amongst patients after AWMGS clinic attendance. From 8 April 2019 to 18 September 2019 , patients attending the genetic counseling clinic for a first appointment at AWMGS were sent a pre-appointment GOS (Figure 1) by post, with completed pre-clinic questionnaires handed in on arrival at clinic. Patients who had forgotten to bring the completed pre-appointment GOS (Figure 1) were given a new pre-appointment GOS at the clinic to fill in on the spot. All patients completing and returning a pre-appointment GOS (Figure 1) were sent a post-appointment GOS together with a reply-paid envelope 4 weeks after clinic attendance. Patients attending disease management clinics, where genetic issues were not the primary concern, were excluded from this study.

Unfortunately, it has not been possible to calculate a participation rate or a completion rate. This is because there are so many locations across Wales in which clinics are held, with some in use every day and others only once per 1-2 months, and they have different secretarial and administrative staff involved. Practices have therefore diverged in terms of the proportion of patients given the questionnaires at each stage.

The before-and-after GOS data were inserted into a Microsoft Excel spreadsheet. The data were then analyzed using a paired samples t-test with IBM SPSS. After reversing the score for Question 3, the score of all six items was added up to provide a total empowerment score. We tested a hypothesis that empowerment scores would be significantly higher after clinic attendance. If the null hypothesis were rejected, this would show sensitivity to change of the
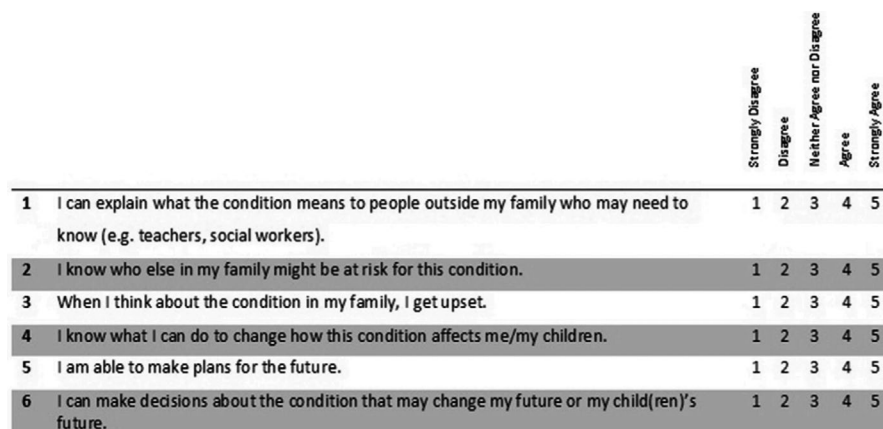

		Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
1	I can explain what the condition means to people outside my family who may need to know (e.g. teachers, social workers).	1	2	3	4	5
2	I know who else in my family might be at risk for this condition.	1	2	3	4	5
3	When I think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

FIGURE 1 The genomics outcome scale (GOS)

GOS. The effect size of the GOS was calculated using Cohen's d formula ($d = M_1 − M_2/s_{pooled}$).

A total of 138 paired GOS questionnaires were collected for this study. Of the 138 paired questionnaires, 20 had incomplete data and were excluded from further analysis.

The mean empowerment scores pre- and post-clinic were 20.11 and 23.08, respectively (See Figure 2). This study shows that empowerment scores increased by an average of 2.97 points after the appointment. Post-appointment scores tend to be distributed more on the high end of the scale compared to pre-appointment scores. (See Figure 3) The pre-clinic standard deviation was slightly wider (4.585) than post-clinic (3.530).

To determine if empowerment scores pre and post-appointment were significantly different, we conducted a paired samples t-test with the p-value threshold for statistical significance set at 0.05. As the p-value was 0.000, we conclude that empowerment scores were significantly higher after clinical attendance (See Table 1). The null hypothesis was rejected in favor of the alternative hypothesis. Cohen's d was calculated to be $d = 0.73$, demonstrating a medium-to-large effect size.

Figure 4 demonstrates an increase in the average score of each item on the GOS, except Question 3. We further analyzed the data by performing a paired samples t-test for each individual item on the GOS. Whilst GOS has been designed to generate a total scale score, item-level data were requested by AWMGS as part of this service evaluation. This follows the earlier experience gained by AWMGS of a service evaluation using GCOS-24, which found that patients achieved significant improvement in post-appointment scores for most GCOS-24 items except for those items designed to capture emotional regulation.

This supported the clinical team to reflect upon their practice and consider areas where practice improvements could be made to maximize patient benefits. (Costal Tirado et al., 2017). Corrections for multiple comparisons were made using the Benjamini–Hochberg method (Bejamini & Hochberg, 1995, see Table 1). By setting the p-value threshold for statistical significance at 0.05, Table 1 shows that the post-clinic scores were significantly higher for all items, except Question 3 (When I think about the condition in my family, I get upset).

Post-appointment empowerment scores were significantly higher than pre-appointment empowerment scores with an average increase of 3.26 points. This study has shown good sensitivity to change of GOS, with a medium-to-large effect size and some evidence of construct validity of the GOS. GOS has previously been shown to capture empowerment (Grant et al., 2019). Research has also demonstrated that empowerment is a valued outcome from clinical genetics and genetic counseling services (McAllister et al., 2010). The present study has demonstrated not only that GOS is sensitive to change in empowerment levels, but has also demonstrated that patients attending AWMGS have higher levels of empowerment following clinical attendance. This therefore demonstrates that participants in this study have derived a measurable benefit from AWMGS attendance.

The effect size of this study ($d = 0.73$) compares favorably with the original GCOS-24 study done in the UK ($d = 0.70$) (McAllister et al., 2011). Published studies that have used the GCOS-24 have shown a wide range of effect size. Studies in psychiatric and cardiovascular genetic counseling demonstrated a large effect size ($d = 1.10$ and 0.94 respectively), whereas a 2018 Spanish study and a 2017 UK study showed medium-to-large effect size ($d = 0.70$ and 0.64 respectively) (Costal Tirado et al., 2017; Gerrard et al., 2020; Ison et al., 2019; Muñoz-Cabello et al., 2017). This variation in effect size could be due to variation in patient populations and practice models of genetic counseling in different centers.

Post-appointment empowerment scores were significantly higher for all questions, except Q3 (When I think about the condition in my family, I get upset), which addresses the subdomain of emotional regulation within the concept of empowerment (McAllister et al., 2011). This could signal a need for additional counseling at AWMGS to address deeper levels of emotional distress, or it could mean that emotional regulation is more intractable amongst patients of clinical genetics services because of the particular challenges that they face (Costal Tirado et al., 2017; Yuen et al., 2020). However, the GOS has been designed in such a way that it is the total score that is important, not the scores on the individual items.

There is currently conflicting evidence in the literature regarding the subdomain of emotional regulation of the GCOS-24. A study done in Vancouver, BC, has shown that the subdomains of empowerment on which psychiatric genetic counseling had the largest effect were powerlessness and emotional regulation (Gerrard et al., 2020).
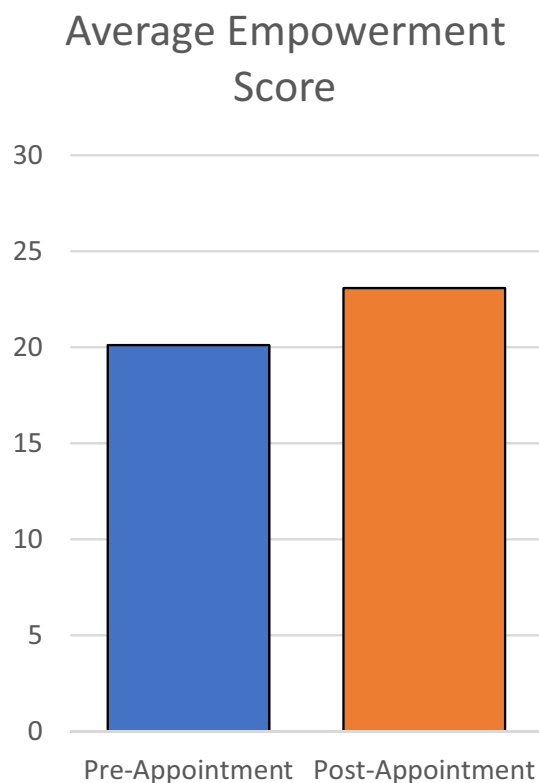

FIGURE 2 Average empowerment (total) scores pre- and post-appointment
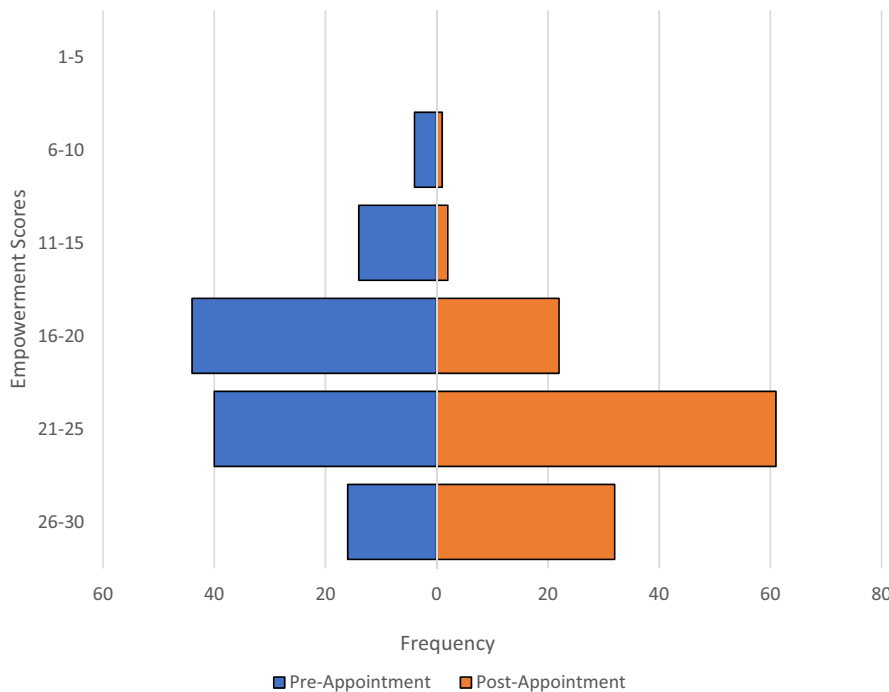
Distribution of Empowerment Scores

FIGURE 3 Distribution of empowerment scores pre- and post-appointment

TABLE 1 Paired samples T-test for empowerment score

| | Mean / Mean Difference | Standard Deviation | Standard Error Mean | 95% Confidence Interval | | t | Sig. (2-tailed) | Adjusted *p*-value[a] |
				Lower	Upper			
Pre Total Score	20.11	4.585	0.422					
Post Total Score	23.08	3.530	0.325					
Post Total Score – Pre Total Score	2.966	4.683	0.431	2.112	3.820	6.880	0.000	.000
Q1 Post – Q1 Pre	0.576	1.135	0.105	0.369	0.783	5.514	0.000	.000
Q2 Post – Q2 Pre	0.669	1.365	0.126	0.421	0.918	5.326	0.000	.000
Q3 Post – Q3 Pre	−0.144	1.142	0.105	−0.352	0.064	−1.371	0.173	.173
Q4 Post – Q4 Pre	0.847	1.337	0.123	0.604	1.091	6.884	0.000	.000
Q5 Post – Q5 Pre	0.466	1.182	0.109	0.251	0.682	4.285	0.000	.000
Q6 Post – Q6 Pre	0.551	1.418	0.131	0.292	0.809	4.220	0.000	.000

[a]Adjusted using Benjamini–Hochberg method (Benjamini & Hochberg, 1995).

This contrasts with the findings of studies in cancer genetics in Singapore and general clinical genetics in the UK, which showed no significant improvement in the emotional regulation sub-scale after genetic counseling (Costal Tirado et al., 2017; Yuen et al., 2020). The variability of the impact of genetic counseling on emotional regulation may be because of differences in the style of genetic counseling in different settings. As these studies were done in different countries, and with different types of patients, the variability may also be due to clinical and/or sociocultural differences between the participants. As Gerrard et al., (2020) commented, the differences in outcome observed between different patient populations would be a fruitful area for further research.

There were some important limitations to this study. Firstly, the extent to which the GOS survey was completed by all eligible patients is unknown. Whilst this is an important limitation, it does not significantly weaken the study findings because an adequate number of responses were collected to enable assessment of sensitivity to change of GOS. The study was also geographically limited to Wales, so the findings may not be reflective of genetic counseling services in other parts of the UK or internationally.

This study was also conducted purely in a clinical genetics setting. As the GOS is designed to be used in both clinical genetics settings and in other clinical specialties or research contexts where genetic counseling and / or testing are offered, it will be

Average Scores for Each Item
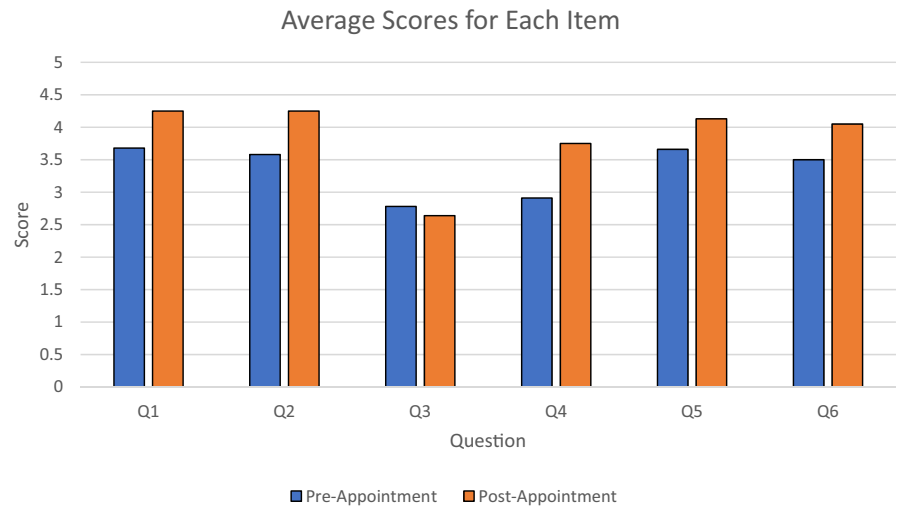
important to assess the usefulness of GOS in these other settings, for example, where genetic/genomic testing is offered in other medical specialties ('mainstreaming'), for example, oncology, pediatrics and neurology. It would also be useful to conduct follow-up work to establish (i) the construct validity of GOS by testing hypotheses regarding how responses correlate / do not correlate with responses to other measures of similar constructs, that is, locus of control, depression, satisfaction of life (ii) the test–retest reliability and (iii) the minimum clinically important difference (MCID) for the GOS.

AUTHOR CONTRIBUTIONS

Michael Sing Onn Ting: Analysis and interpretation of data, writing of manuscript; Angus Clarke: Design of the study, data collection, supervision of Michael Sing Onn Ting in writing the manuscript, manuscript preparation; Marion McAllister: Design of the study, supervision of Michael Sing Onn Ting in the analysis of the data and in writing manuscript, manuscript preparation. Author Michael Sing Onn Ting confirms that he 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 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.

COMPLIANCE WITH ETHICAL STANDARDS

CONFLICT OF INTEREST

The authors have no conflict of interest to declare.

HUMAN STUDIES AND INFORMED CONSENT

All procedures performed in this study involving human participants were in accordance with the ethical standards of Cardiff & Vale University Health Board Research & Development Office and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

ANIMAL STUDIES

No non-human animal studies were carried out by the authors of this article.

DATA AVAILABILITY STATEMENT

Data available on request from the authors: The data that support the findings of this study are available from the corresponding author upon reasonable request.

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