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Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples

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Authors' Contributions

Authors, Allyn McConkie-Rosell, Vandana Shashi, and Stephen Hooper, 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 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.

AMR, VS, and SRH contributed equally to project in terms of study design, measurement development, analysis of study data, coordination of project, and manuscript preparation and drafting. KS, JS, and CP assisted with conceptualization and revision of the GEmS measure. KS, RS, HC, JS, KT, QT, CP all participated in drafting of the manuscript. KS, RS, HC, JS, assisted in data collection and analysis of the data. The Undiagnosed Diseases Network is responsible for application (patient) allocation to the Duke UDN site. All authors read and approved the final manuscript.

Compliance with Ethical Standards

Conflict of Interest

Allyn McConkie-Rosell, Kelly Schoch, Heidi Cope, Jennifer Sullivan, Rebecca C. Spillmann, Khoon G. Tan, Christina G. S. Palmer, Undiagnosed Diseases Network, Stephen R. Hooper, and Vandana Shashi declare that they have no conflict of interest.

Human Studies and Informed Consent

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. The data gathered for this study were approved by the Institutional Review Boards of the National Human Genome Research Institute (15-HG-0130) and Duke University Medical Center (Pro00056651 and Pro00032301). Informed consent was obtained from all individual participants included in the study.

Animal Studies: none

Data Availability: The datasets generated and analyzed during the current study are not publicly available due to patient confidentiality, but can be redacted and made available from the corresponding author upon reasonable request.

Members of the Undiagnosed Diseases Network

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Clinical Application of a Scale to Assess Genomic Healthcare Empowerment (GEmS): Process and Illustrative Case Examples

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Abstract

The Genome Empowerment Scale (GEmS), developed as a research tool, assesses perspectives of parents of children with undiagnosed disorders about to undergo exome or genome sequencing related to the process of empowerment. We defined genomic healthcare empowerment as: perceived ability to understand and seek new information related to the genomic sequencing, manage emotions related to the diagnostic process and outcomes, and utilize genomic sequencing information to the betterment of the individual/child and family. The GEmS consists of four scales, two are primarily emotion focused (Meaning of a Diagnosis, and Emotional Management of the Process) and two are action oriented (Seeking Information and Support, and Implications and Planning). The purpose of this research was to provide a strategy for interpreting results from the GEmS, and present illustrative cases. These illustrations should serve to facilitate use of the GEmS in the clinical and research arena, particularly with respect to guiding genetic counseling processes for parents of children with undiagnosed conditions.

Keywords

Exome and genomic sequencing; undiagnosed disorders; healthcare empowerment; genetic counseling; parental perspectives; rare disorders

Introduction

Next-generation genomic sequencing (inclusive of exome and genome sequencing) has changed the diagnostic paradigm for patients presenting with suspected genetic conditions

Jennifer Wambach, Jijun Wan, Lee-kai Wang, Michael F. Wangler, Patricia A. Ward, Daniel Wegner, Mark Wener, Tara Wenger, Katherine Wesseling Perry, Monte Westerfield, Matthew T. Wheeler, Jordan Whitlock, Lynne A. Wolfe, Jeremy D. Woods, Shinya Yamamoto, John Yang, Guoyun Yu, Diane B. Zastrow, Chunli Zhao, Stephan Zuchner

that are refractory to traditional diagnostic approaches (Shashi et al., 2013). The benefits include a diagnosis rate of 30–40% (Clark et al., 2018; Gilissen, Hoischen, Brunner, & Veltman, 2011; Lee, Deignan, Dorrani, & et al., 2014; Need et al., 2012; Yang, Muzny, Xia, & et al., 2014), identification of new disease-causing genes and in some instances, new treatments or changes in medical management. While exome (ES) and genome sequencing (GS) holds great promise as a diagnostic tool, it poses unique challenges as well. There can be considerable variation in what individuals and families expect from the sequencing; how they understand the results; and how they might use the results for the betterment of themselves and families. Such variation can be a function of a myriad of psychosocial factors affecting both parents of affected children and adult probands that include feelings about the inherent uncertainty of the diagnostic journey, worries about missed treatment possibilities, the presence of anxiety or depression, and the level of self-efficacy (McConkie-Rosell et al., 2018; McConkie-Rosell et al., 2016). Thus, expectations of the sequencing, understanding the results, and using the results for the betterment of the individuals and the families can vary considerably from one individual to the other. These factors, in conjunction with the complexities of the results, such as variant pathogenic classification, and potential for diagnostic results to change with reanalysis (Brett et al., 2018; Shashi et al., 2015), create a need for increased identification and management of these psychosocial factors linked to the well-being of individuals undergoing this diagnostic process. With our focus on parents of undiagnosed children, we have noted parents are able to maintain positive coping self-efficacy, and remain engaged in their child's healthcare, and are tolerant of uncertainty, despite a third of parents reporting symptoms of anxiety and depression (McConkie-Rosell et al., 2018). These findings underscore the importance of practicing clinical geneticists, genetic counselors, and other providers (e.g., social workers) ascertaining the expectations, emotional management, and perceptions of the ability to understand and utilize the information derived from genomic sequencing in order to optimize potential benefits for the families.

Clinical genetic counselors face the challenges of busy clinical practices, with consequent time constraints (Attard, Carmany, & Trepanier, 2019; Maiese, Keehn, Lyon, Flannery, & Watson, 2019; Sukenik-Halevy, Ludman, Ben-Shachar, & Raas-Rothschild, 2016) that may limit exploration of parental expectations, their understanding of the process, abilities to cope and manage the process and diagnostic outcomes, and types of outcomes that result from ES/GS. Additionally, the complexity of the discussion for ES/GS consent is difficult to balance with specific tailoring of the genetic counseling due to psychosocial factors such as the individual's coping strategies and social support (Macnamara et al., 2019; Schmidlen et al., 2018). These findings led us to develop the Genome Empowerment Scale (GEmS) (McConkie-Rosell et al., 2019) as patient-reported tool to assess genomic healthcare empowerment in adult probands/parents of children with undiagnosed disorders about to undergo ES/GS.

We chose empowerment as the theoretical foundation for the GEmS because it is both a process and an outcome, is key to healthcare autonomy and decision making (Johnson, 2011; Paloma Garcimartin & Linas-Alonso, 2017), and critical to a patient centered approach to genetic counseling (Veach, Bartels, & Leroy, 2007). Empowerment is a social construct that has been defined in many different contexts and in clinical genetics and

genetic counseling, as encompassing the dimensions of cognitive, decisional and behavioral control, emotional regulation, and hope (McAllister & Dearing, 2015; McAllister, Wood, Dunn, Shiloh, & Todd, 2011). The process of empowerment includes developing: the knowledge, skills, and confidence in oneself to emotionally manage the life or health challenges that are being presented, the self-confidence to be able to identify and utilize resources, and connect to and learn from similar others (Johnson, 2011; McConkie-Rosell & Sullivan, 1999; Minooei, Ghazavi, Abdeyazdan, Gheissari, & Hemati, 2016; Paloma Garcimartin & Linas-Alonso, 2017). Critical to the process of empowerment is education, engaging with health providers and seeking out information about the illness/condition, and being an active partner in decision making (Johnson et al., 2007). Strategies to facilitate healthcare empowerment are designed to develop skills and knowledge needed so that participation in healthcare is characterized as (1) engaged, (2) informed, (3) collaborative, (4) committed, and (5) tolerant of uncertainty (Johnson, 2011).

We defined Genomic Healthcare Empowerment as: *the perceived ability to understand and seek new information related to the genomic sequencing, manage emotions related to the diagnostic process and outcomes, and utilize genomic sequencing information to the betterment of the individual/child and family* (McConkie-Rosell et al., 2019). The GEmS was designed to identify malleable areas of need related to genomic healthcare empowerment process, which may be amenable to targeted genetic counseling. This paper provides a strategy for interpreting results from the GEmS, illustrated by case examples.

Description of the Genome Empowerment Scale (GEmS)

As reported previously, validation assessments of the GEmS with parents of children undergoing ES/GS for diagnostic purposes found that it had good psychometric properties (McConkie-Rosell et al., 2019) and was consistent with the theoretical foundation of healthcare empowerment (Johnson, Rose, Dilworth, & Neilands, 2012; Johnson, 2011). The GEmS consists of 28 items, each evaluated on a Likert scale from 1 (low)-7 (high). The GEmS also has two open ended items. In general, Meaning of a Diagnosis and Emotional Management are both emotion-focused scales while Seeking information and Support and Implications and Planning are action-oriented. The four scales and relationship to empowerment are in Table 1. See Supplement Table 1 for GEmS measure.

GEmS Interpretation Strategy and Case Identification

Human Subjects Ethics approval was obtained from the Institutional Review Boards of the National Human Genome Research Institute (15-HG-0130) and Duke University Medical Center (Pro00056651; Pro00032301). Statistical analyses were performed with SPSS version 26.0 (IBM, SPSS). Data collection occurred from November 2016 to April 2019.

Parents of children enrolled in the Duke clinical site of the Undiagnosed Diseases Network (UDN) https://undiagnosed.hms.harvard.edu) and the Duke Genomic Sequencing Clinic were enrolled for the GEmS validation study. We have previously reported findings on Anxiety (GAD-7) and Depression (PHQ-9), coping self-efficacy, and health care empowerment in a subset of this cohort (McConkie-Rosell et al., 2018).

Interpretation Strategy:

Total raw scores were tabulated for each scale on the GEmS and descriptive statistics (frequencies, means, and standard deviations) were used to summarize the data. All raw scores were transformed into z-scores based on the available data from all participants (Mean = 0, SD = 1). This transformation allowed us to classify an individual's score as 0 (Average) and ≥ or ≤1 SD (Low or High) based on their z score. The total raw score ranges for z-score for each of the scales for the categories of Low, Average, and High can be seen in Table 2. For details on conversion of raw scores to z- scores see supplement Table 2. The resulting distribution of average and high/low of each scale was 15–20% either High or Low, with the majority in the average range. To provide an index of reliability we calculated the Standard Error of Measurement (SEM) for the z-scores for each GEmS scale using a 90% Confidence Interval: Meaning of a Diagnosis = ± 0.67; Emotional Management of the Process = ± 0.73; Seeking Information and Support = ± 0.82; and Implications and Planning = ± 0.92. The SEM provides a range for how an obtained GEmS rating may be spread around a "true" score. We then systematically reviewed all cases to discern potential patterns of each of the GEmS scales, taking into account high, low, or average z- scores and the previously identified inter-correlations with other measures used for validation of the GEmS (McConkie-Rosell et al., 2019). Meaning of a Diagnosis scale is inversely correlated with Emotional Management scale and is positively correlated with Seeking Information and Support and Implications and Planning scales, with the latter two scales positively correlated with each other. We also previously found that scores on the Meaning of a Diagnosis scale were positively correlated with greater reported symptoms of anxiety and depression as assessed by the PHQ-9 (Kroenke, Spitzer, & Williams, 2001) and GAD-7 (Spitzer, Kroenke, Williams, & Lowe, 2006). Scores on the Emotional Management scale were inversely correlated with reported symptoms of anxiety and depression (i.e., lower scores on Emotional Management suggested greater symptoms) and positively correlated with scores on the Coping Self-Efficacy (CSE) (Chesney, Neilands, Chambers, Taylor, & Folkman, 2006) and Healthcare Engagement and Tolerance for Uncertainty on the Health Care Empowerment Inventory (HCEI) (Johnson et al., 2012).

Based on this process, we developed the following cascade model to guide the interpretation of the GEmS:

1. Convert raw score to z-score for each scale;
2. Classify z-scores as average (z = 0 < 1 SD), high (z ≥ 1 SD), or low (z ≤ −1 SD);
3. Identify scales that are average, high, or low;
4. Visually screen items for outliers, defined as a raw score of either ≤2 or ≥6;
5. Review responses to open-ended questions to determine if a specific concern is reported and consider any other relevant data (e.g., assessments for anxiety, depression, family functioning, etc.);
6. Synthesize the above data and consider adjusting the genetic counseling session to focus additional time on the topics comprising the high and low scales.

It should be noted that we used z-scores of 1 SD as our cutoff to facilitate interpretation and to address issues that may be in the top 15% of concerns; however, as a continuous measure using the z-scores other cut-points can be utilized based on the z-scores (e.g., 1.5 SD) (see Supplement Table 2). *It is important to keep in mind that the z-scores presented are standardized to this unique population of parents who have children with undiagnosed disorders and not to parents in the general population or parents of children with diagnosed health conditions. Thus, even an "average" GEmS score should not be immediately dismissed as nonproblematic.*

Case Identification:

For the case selection for this paper, we used the pool of cases from 178 parents who had completed the GEmS, 158 of these parents were part of the validation study. The characteristics of the sample demographics were as previously described (McConkie-Rosell et al., 2019) where the majority of participants were mothers, 66% (118/178); Caucasian 87% (155/178); and 55% (98/178) had prior experience with a non-diagnostic ES. The mean age for the parents was 40.80 ± 8.58 years and for the proband was 9.32± 6.85. It is not possible to provide examples of all possible combinations of the scales; therefore, we selected four cases, which illustrated average, high, and low z-scores for each scale. We have labeled the cases with descriptors for the purposes of illustration; however these are our arbitrary descriptors and others may be more appropriate.

Illustrative Cases

Confident Realist:

Frequency: This pattern, in which all scores are in the average range, was common, with a frequency of 26 percent (46/178). Sixty-three percent (29/46) had a prior non-diagnostic ES for their child. The mean age of the proband was 7.93±4.0 years, with 63% (29/46) of the respondents being mothers and 37% (17/46) fathers. (Figure1)

This is from a mother with a 9-year child.

Z scores scales: All are within ± 1 SD

Relationship among the scales: Balanced

Screen Items for outliers: Strong positive perception of relationship with medical genetics team and local health providers. No current plans for using a diagnosis for reproductive planning for self or family.

Open-ended items: No comments provided.

Other relevant data: This parent reported symptoms in the range of mild depression on the PHQ 9 rating scale. No prior experience with ES.

Data synthesis: This all average pattern suggests a parent who is realistic about the possibility that the ES will result in a diagnosis, is able to partner with the genetics team,

and feels confident in her ability to emotionally manage the outcome with an understanding of the possibility of an uncertain result or no diagnosis. It also suggests a parent who is confident in his/her abilities to seek the help and support he/she may need and plan next steps once the sequencing is resulted.

This pattern is important to highlight because these middle range scores represent the "average" for the parents of children undergoing ES/GS for diagnostic purposes. These parents may still have a number of concerns, but just not more so than other parents going through the same diagnostic process. While this pattern does not suggest a specific area to target as part of the genetic counseling process, the screening for outliers of individual items may suggest specific areas that might be explored. For example, while this parent does not have plans to use information from ES/GS for reproductive planning, if these items were endorsed it would suggest a topic that should be addressed in the genetic counseling

Engaged But Worried Planner:

Frequency: Eleven percent (20/178) of the patterns were found to be high on Meaning of a Diagnosis with average or low scores on Emotional Management with Seeking Information and Support and Implications and Planning also average or high. Fifty-five percent (11/20) of these parents had prior experience with a non-diagnostic ES. The proband mean age was 9.9±6.73 years with 75% (15/20) of respondents being mothers and 25 % (5/20) fathers. (Figure 2) This pattern of high Meaning with low Emotional Management is correlated with greater self-reported symptoms of anxiety and depression on the GAD7 and PHQ9 (McConkie-Rosell et al., 2019) and is consistent with our previous findings that parents remain highly engaged in the diagnostic process and their child's care, despite their own emotional health (McConkie-Rosell et al., 2018).

This is from a mother of a 22-year old.

Z scores scales: All are > 1SD

Relationship among the scales: High Meaning, Seeking Information and Support, and Implications and Planning paired with low Emotional Management.

Screen Items for outliers: The items suggest this parent has very high expectations of a diagnosis with low tolerance for uncertainty and the discomfort of the ongoing search for a diagnosis. High interest in support from other families and seeking new information. A diagnosis is important for reproductive planning for her family.

Open-ended items: "Having more information on my child's condition will allow me to get her to people who can help her." "There could be a treatment that could improve my child's life substantially"

Other relevant data: This mother endorsed symptoms in the range of mild depression on the PHQ9 rating scale. No prior experience with ES

Data synthesis: This pattern is suggestive of a parent who has a high value attached to a diagnosis and has expectation that a diagnosis will lead to information that can be used to improve her child's management and at the same time, is also reporting that the search for a diagnosis has been difficult, and she may exhibit low tolerance for uncertainty as well as a diagnosis for which little may be known. The high expectation of the sequencing results may not be met as currently the majority of patients will continue to be undiagnosed even after undergoing ES/GS and if a diagnosis is achieved it may be a novel gene or an ultra-rare disorder for which only limited information is presently known. This parent's scores are also high on the action scales. This suggests a parent who is confident in her ability to take next steps to utilize information once the ES/GS is resulted. Both the importance of a diagnosis and her confidence in taking next steps is supported by her response to the open-ended item.

This parent may benefit from an exploration of her downstream expectations of a diagnosis, including how it might change management of her child. Genetic counseling can also explore this parent's perceptions about common outcomes of ES/GS, comparing what might be learned if the diagnosis is a well-known gene versus the discovery of a novel gene/ultra-rare disease, or a gene variant of uncertain significance that might need further follow-up and how these may affect reproductive risks and planning in the future. The genetic counseling should also explore feelings about not having a diagnosis and facilitate emotional coping strategies. Because of the high interest in seeking information and support, exploring with this parent ways to identify reliable and accurate information might be helpful to prevent misinformation/misunderstanding of a gene finding and, ultimately, significant disappointment.

Resigned Acceptor

Frequency: Thirteen percent (24/178) of the patterns showed low Meaning and high or average Emotional Management with Seeking Information and Support and Implications and Implications and Planning either low or average. Fifty percent (12/24) of these parents had experienced a prior non-diagnostic ES for their child. The proband mean age was 6.8± 0.5 years with 58% of the respondents being (14/24) mothers and 42% (10/24) fathers. (Figure 3) This pattern appears to be the inverse of the Engaged Worried Planner.

This is from a mother of an 11 year-old child.

Z scores scales: > 1SD, except for Implications and Planning

Relationship among the scales: Low Meaning of a Diagnosis, Seeking Information and Support, with high Emotional Management

Screen Items for outliers: Low expectation that a diagnosis will occur and low importance of a diagnosis. Low confidence in abilities to use or find new information and no plans to use the information for reproductive planning for self/family.

Open-ended items: "More information may make treatment and strategies for care more efficient and effective". "Possible medication to lessen the impact of her conditions, a gauge of her level of functioning to determine how much independence she can have."

Other relevant data: This parent did not endorse symptoms of anxiety or depression on the PHQ9 or GAD 7 rating scales. No prior experience with ES.

Data synthesis: The low score on Meaning of a Diagnosis suggests a parent who has little belief that the ES/GS will result in a diagnosis for her child. The high score on Emotional Management suggests that this parent is tolerant of diagnostic uncertainty and is confident that he/she can emotionally manage whatever is learned from the ES/GC, however, the low score on Seeking Information and Support suggests she is not confident in her abilities to take the next steps in securing support and/or information regarding the diagnostic outcome of the sequencing. This pattern suggests a parent who may have accepted that a diagnosis may never happen and may be unprepared if one is identified. In contrast, her response on the open-ended item and whether she expects the sequencing to improve her child's life indicate that she retains hope that the ES/GS will result in a diagnosis that will lead to improved management for her child.

The genetic counseling process for this parent should consider focusing on establishing a trusting relationship with her and build confidence that once the ES/GS results are obtained there will be support and practical assistance in how the information can be used and how to find it (i.e., locating other families, learning more about the gene and implications for the child and family with a plan for "next steps").

Disengaged or Overwhelmed

Frequency: While this pattern was seen only once [0.5% (1/178)], it requires further consideration to determine if it represents disengagement from the diagnostic process or emotional exhaustion. (Figure 4) The converse pattern of all scales being high was not identified.

This is a from a father with a 4-year-old child.

Z scores scales: All scores > 1SD.

Relationship among the scales: All scores low

Screen Items for outliers: Low expectation of a diagnosis and low perception of severity of the illness, with the importance of a diagnosis high, perception that the diagnosis process has been difficult and low tolerance for a diagnosis in which little is known. Low on confidence in ability to understand, find or use resulting information.

Open-ended item: No comments

Other relevant data: This parent did not endorse symptoms of either anxiety or depression on the GAD 7 and PHQ9. Previous non-diagnostic ES.

Data synthesis: The low scores across Meaning of a Diagnosis and the two action scales with low Emotional Management may suggest either disengagement from the ES/GS testing

and the associated diagnostic process or a parent who is overwhelmed with it. This pattern varies from the “Resigned Acceptor” because of the low score on Emotional Management.

Despite the low expectation and low Emotional Management, this parent is continuing to seek a diagnosis, with the importance of having one being rated as high. Genetic counseling exploring why he is continuing with the diagnostic process (i.e., what or who is the driving force to continue) as well as on the difficulties inherent in the ongoing diagnostic odyssey and the emotional toll it can take on the parent, child, and the family. This pattern may also suggest a parent who “needs a break” from the diagnostic process.

Discussion

For children who are undiagnosed, but suspected to have a genetic disorder, parents bring with them, on their diagnostic journey, their concerns for their child, and individual factors such as confidence in ability to comprehend complex information and to seek out and utilize information, social support(s), and emotional management strategies. Genetic counselors and clinical geneticists are tasked with providing parents with the information about ES/GS needed to make informed decisions for their child. Seasoned clinicians/genetic counselors also know that parents vary in their expectations, ability to emotionally manage and utilize ES/GS results, and that there may be underlying psychosocial factors influencing the process, but the time constraints of a busy practice and the lack of a standardized measure to assess these aspects make it difficult for them to provide further assistance. Our clinical and research experience also suggests that parents may not always express their worries about genomic sequencing, and thus these parental concerns may not be readily identified in a fast-paced clinical setting (McConkie-Rosell et al., 2018; McConkie-Rosell et al., 2016).

The GEmS is a tool designed to measure genomic empowerment, via caregiver ratings, which can be deployed in a clinical or research setting. It is easy to score, with current z-scores (see Supplementary Table 2) based on a sample of parents of children referred for evaluation of undiagnosed disorders for which ES/GS was being performed for diagnostic purposes. We recommend a cascade approach for the interpretation for the GEmS. To illustrate this interpretative approach, we specifically chose four cases from our available data to demonstrate the relationship among the scales and the associated interpretive process. These cases were chosen for illustrative purposes and many other possible combinations exist. In addition to the overall z scores and assessing the relationship among the scales, screening individual items for outliers may suggest a topic for further discussion or suggest a strength, which can be built upon in the genetic counseling process. The responses to the open-ended items can also be useful in gaining insight into the scores on the scales. This approach to the interpretation of the GEmS scales can facilitate the identification of needed steps in the process of empowerment that can be used to inform the focus of the genetic counseling.

Integrating the GeMS into practice

Using a health care empowerment framework, the GEmS can help to identify parental concerns that can be targeted during genetic counseling prior to embarking on ES/GS. The genetic counselor and the parent can jointly review these potential concerns. For example,

reviewing z scores with parents may provide an opportunity for parents to compare their expectations, emotional management, and utility related to genomic sequencing relative to the community of "similar others;" that is, other parents who are searching for a diagnosis for their child. Identification with others facing the same or similar situation is an important step to empowerment (Gutiérrez, 1994). Reviewing findings with the parent also may facilitate building a collaborative relationship, also an important step to empowerment (Gutiérrez, 1994; McConkie-Rosell & Sullivan, 1999). We believe that the GEmS can provide additional information to assist in the genetic counseling process and guide referrals for further psychosocial supports. Clinicians using the GEmS should pay particular attention to the relationships among the scales that can manifest, particularly when paired with other medical, family, and health-related information, to understand and encourage genomic healthcare empowerment in families of children with undiagnosed disorders.

Limitations

The data upon which the z-scores were developed for the GEmS were obtained from a sample of parents whose children were undergoing ES/GS as part of a research study, the majority of whom were included in the validation study. The majority of parents in this study were white and had a college education, which does not reflect the diversity typically seen in a clinical setting. Thus, it remains a question as to whether parents from other racial and ethnic backgrounds, those who are research naïve, or those who have not experienced a protracted search for a diagnosis would respond similarly. It will be important to collect data from a second, larger and more diverse cohort, to develop normative data and increase generalizability to other populations and experiences as it is possible that the z scores obtained from such a cohort could differ from those presented. Our cascade model for the interpretation of the GEmS is a preliminary scoring approach; data from a larger cohort would allow us to conduct analysis that would provide statistical reliability of the number and type of relevant patterns and resulting profiles (e.g. Latent Profile Analysis). It was also not possible to account for all possible influences of the GEmS scores, such as if the respondent is the primary caregiver, age of the affected child, length of time spent searching for a diagnosis, etc.; these are all potentially variables that should be the focus of future research. The interpretation of the illustrative GEmS cases are ours and alternative interpretations can be made.

The clinical interpretation of rating scales includes response biases and the GEmS is not immune to such biases. Parents completed the GEmS, after informed consent and initial genetic counseling had occurred, with genetic counselors who have extensive experience with ES/GS. We cannot exclude that this process influenced the responses on the GEmS. The GEmS was not designed to ascertain a specific diagnosis (e.g., anxiety and or depression), although low scores on the Emotional Management scale have been shown to be highly correlated with self-reported symptoms of anxiety and depression (McConkie-Rosell et al., 2019), and high scores on Meaning of a Diagnosis with symptoms of anxiety and should prompt consideration and discussion of this possibility. The current version of the GEmS cannot be used for assessment after ES/GS results have been obtained, although the post-test GEmS scale is in development.

Future Directions

The presentation of the various case illustrations showcases the potential utility of this rating scale. Future research will focus on how or if these scores and associated patterns might change once the results of the ES/GS are known, i.e.; are the scales sensitive to change depending on the diagnostic outcome? In this regard, we have developed follow-up versions of the GEmS, which are specific to diagnostic outcome (i.e., no diagnosis, probable diagnosis, definite diagnosis). These versions are designed to assess change in the scales once ES/GS results are received and is the focus of an ongoing study. It was also notable that although the case illustrating low z- scores across all four scales was presented, it was found only once; and, similarly, the case illustrating all scores being high was not identified in our sample. Because the z- scores represent the norms of parents who are in the midst of a diagnostic odyssey, it is possible that the extreme low on all scales may be rare in parents who are actively seeking a diagnosis versus parents who are no longer actively looking. The lack of all high scores also should be further explored.

Conclusion

Based on a health care empowerment theoretical framework, the GEmS, yields information that can be used for a refined and patient-specific genetic counseling discussion prior to ES/GS. The GEmS may be useful in identifying issues that a parent/patient might be thinking, but not be verbalizing. Just as ES/GS have changed the diagnostic algorithms, genetic counselors also need to adjust their counseling practices in order to meet the needs of patients and their families, and the GEmS has the potential to assist in that process. The GEmS has demonstrated potential for utility for these clinical settings and should provide additional information for the genetic counselor with the goal of maximizing the outcome for families, regardless of diagnostic outcome.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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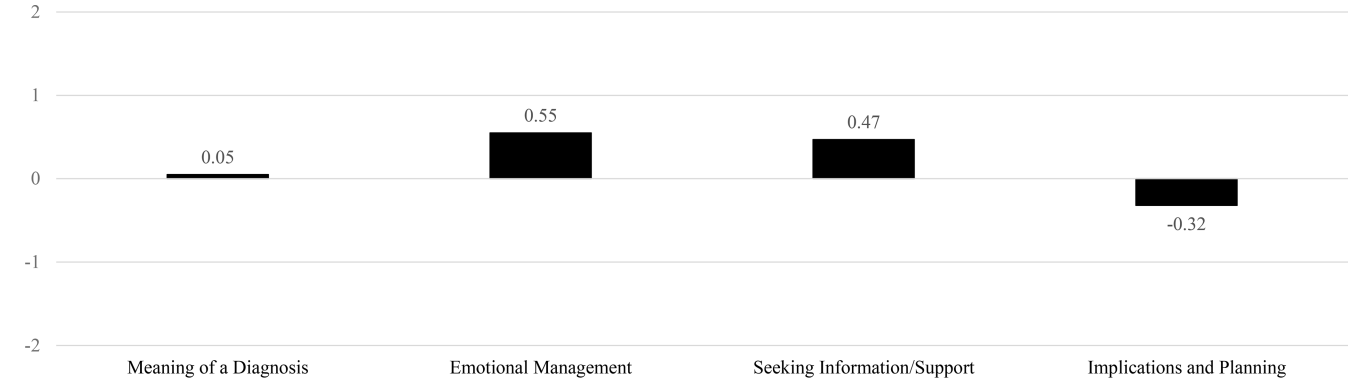
What is known about this topic

While exome and genome sequencing are changing the landscape of the diagnosis of previously undiagnosable rare and ultra rare disorders; genetic counselors face the challenges of facilitating parents of children and adult probands undergoing this testing, understanding, emotional management of the process, and utilization of the results. The GEmS is a newly developed measure designed to assess genome healthcare empowerment across four scales, Meaning of a Diagnosis, Emotional Management of the Process, Seeking Information and Support, and Implications and Planning.

What this paper adds to the topic

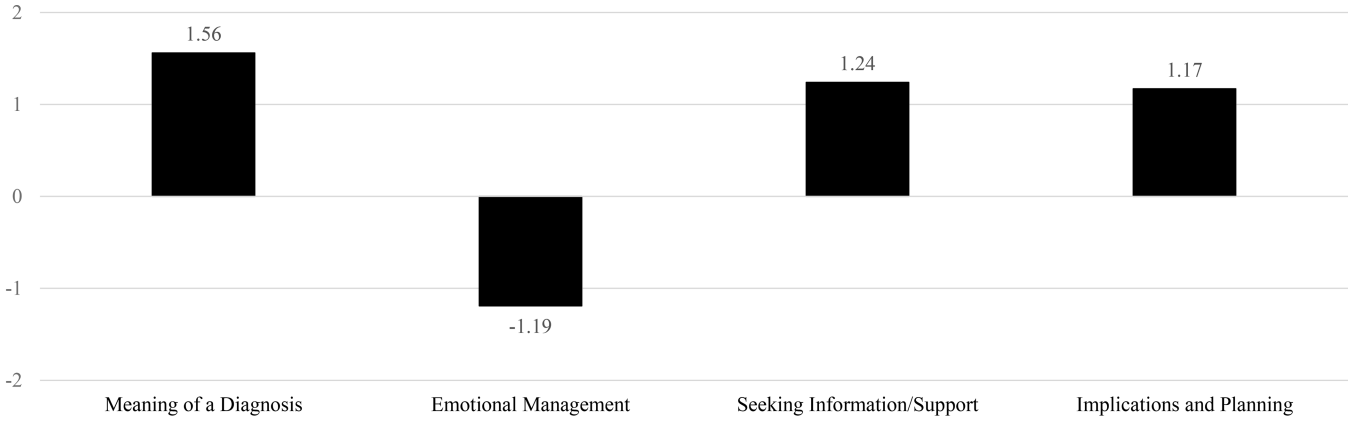
The GEmS was designed to identify aspects of Genomic Empowerment, in which extreme scores (high or low) on one or more of the scales, may identify areas of need amenable to targeted genetic counseling. The purpose of this paper is to present a strategy for the interpretation of the GEmS, illustrated by case examples.



Individual Item Scores (range 1-7) Higher scores reflect a more positive rating

4 likelihood of a dx	5 Feeling about no dx	7 find assistance	1 reproductive self
7 partner/genetics team	4*emotionally difficult	3 find families	1 reproductive family
7 therapy/education	4*worried/primary dx	7 learn families	7 reproductive others
7 medical decisions	4*worried/ 2nd findings	5 use information	7 primary medical provider
5 importance of a dx	6* uncertain dx	6 find information	7 partner/plan for child
4 severity	5 confident	6 understand	
5 improve child's life	4*little information	4 importance families	
6 life planning	6*physically difficult		
*reversed scored			

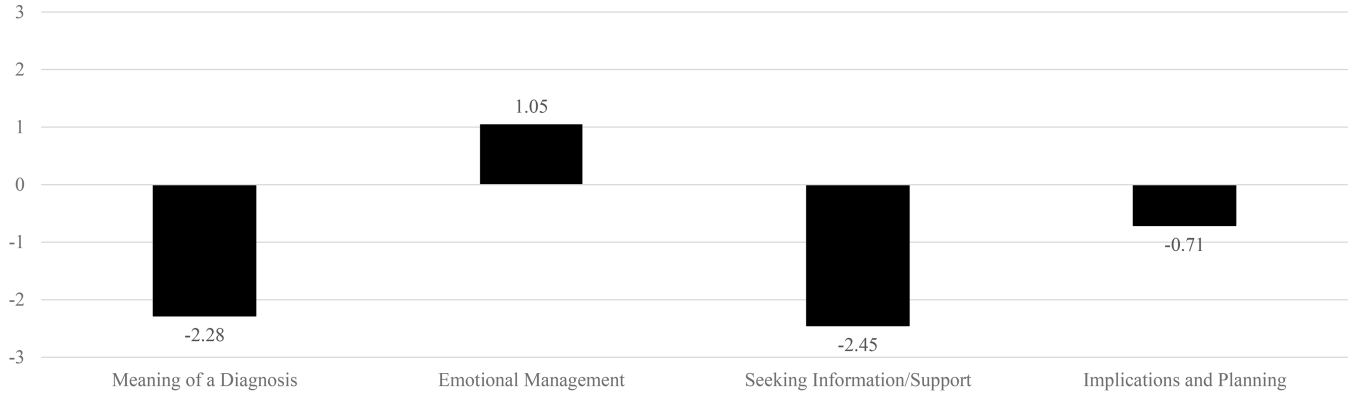
Figure 1:
Confident Realist



Individual Item Scores (range 1-7) Higher scores reflect a more positive rating

7 likelihood of a diagnosis	3 Feeling about no dx	4 find assistance	4 reproductive planning self
7 partner with genetics team	1*emotionally difficult	4 find families	7 reproductive planning family
7 therapy/education	5*worried/primary dx	7 learn families	7 reproductive planning others
7 medical decisions	4*worried/2nd findings	7 use information	7 primary medical provider
7 importance of a dx	1* uncertain dx	7 find new information	7 partner/plan for child
7 severity	7 confident	7 understand	
7 improve child's life	2*little information	7 importance families	
7 life planning	1*physically difficult		
*reversed scored			

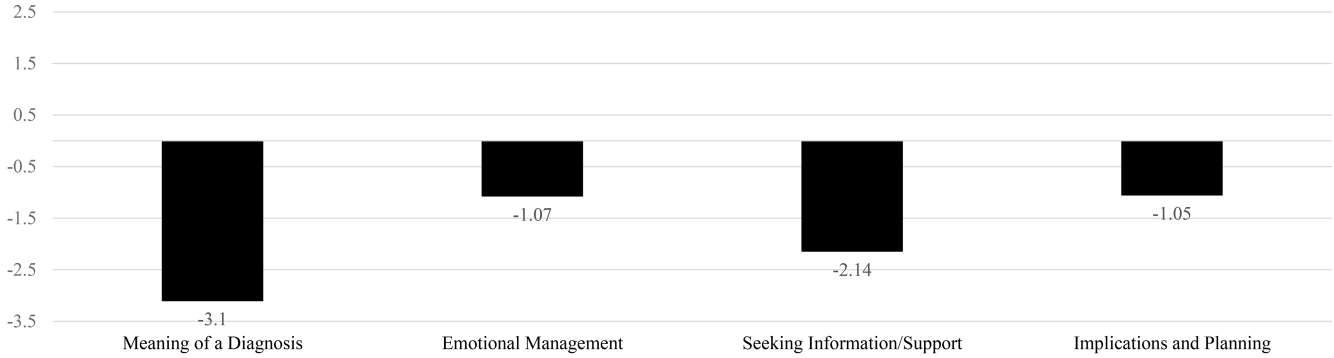
Figure 2:
Engaged But Worried Planner



Individual Item Scores (range 1-7) Higher scores reflect a more positive rating

2 likelihood of a diagnosis	4 Feeling about no dx	5 find assistance	1 reproductive self
6 partner with genetics team	4*emotionally difficult	2 find families	3 reproductive family
4 therapy/education	6*worried/primary dx	3 learn families	5 reproductive others
2 medical decisions	6*worried/2nd findings	2 use information	7 primary medical provider
2 importance of a dx	6* uncertain dx	2 find new information	5 partner/plan for child
3 severity	6 confident	3 understand	
4 improve child's life	6*little information	2 importance families	
5 life planning	5*physically difficult		
*reversed scored			

Figure 3:
Resigned Acceptor



Individual Item Scores (range 1-7) Higher scores reflect a more positive rating

1 likelihood of a diagnosis	4 Feeling about no dx	2 find assistance	4 reproductive self
6 partner with genetics team	2*emotionally difficult	2 find families	1 reproductive family
2 therapy/education	3*worried/primary dx	3 learn families	4 reproductive others
2 medical decisions	3*worried/ 2nd findings	2 use information	4 primary medical provider
6 importance of a dx	3* uncertain dx	6 find new information	6 partner/plan for child
1 severity	6 confident	2 understand	
2 improve child's life	2*little information	4 importance families	
2 life planning	2*physically difficult		

*reversed scored

Figure 4:
Disengaged or Overwhelmed

Table 1

Genome Empowerment Scale and Relationship to Empowerment

GEmS Scales		Relationship to Empowerment
Emotion Focused		
Meaning of a Diagnosis (8 items)	• Likelihood that the sequencing will result in a diagnosis • Importance of a diagnosis for informing management decisions • Planning for the future • Perception of how a diagnosis may improve his/her child's life • Perception of severity of his/her child's condition • Open-ended question to describe how the information gained from the sequencing may in the future improve his/her child's life.	• Knowledge building • Education regarding risk benefits, and limitations • Expectations of health outcome • Engagement in healthcare
Emotional Management (8 items)	• Level of worry about what might be learned from the sequencing regarding their child's diagnosis • Diagnostic uncertainty • Feelings related to not receiving a diagnosis for their child • Tolerance for limited knowledge known if a rare or ultra-rare diagnosis is made • Open-ended question to allow the parent to describe worries about what might be learned from the sequencing.	• Emotional management strategies • Tolerance for health uncertainty • Self-efficacy • Management of health challenges
Action-Oriented		
Seeking Information and Support (7 items)	• Confidence in ability to understand ES/GS findings • Confidence in ability to seek new information on their own • Find assistance for medical/developmental management for his/her child • Importance and ability to find, learn, secure support from other families whose children have the same or similar diagnosis	• Connect to similar others • Social support • Ability to seek information • Identify and use available resources • Understanding healthcare options
Implications and Planning (5 items)	• Importance of a diagnosis for reproductive decision making for self, family members, and other families • Ability to partner with local health providers to develop management plans based on ES/GS result. Note: For this scale, it is important to consider these two aspects of information and planning that are assessed.	• Partner with health providers • Identify barriers to access • Understand limitations of available health information.

Table 2.

Range of GEmS Raw Total Score to Z scores categorized into 0 (Average) and ≥ or ≤1 SD (High or Low) N= 178

	≤−1 SD (Low)	**0 (Average)**	**≥+1 SD (High)**
Meaning of a Diagnosis (8 items)	≤37 (n=27)	38–51 (n=123)	52–56 (n=28)
Emotional Management (8 items)	≤25 (n=28)	26–42 (n=119)	43–56 (n=31)
Seeking Information and Support (7 items)	≤28 (n=35)	29–41 (n=111)	42–49 (n=32)
Implications and Planning (5 items)	≤19 (n=31)	20–30 (n=108)	31–35 (n=39)