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A Standardized Measurement and Valuation Scale of Genomic Utility for Policy Decisions: The GUV Scale

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ABSTRACT

Objectives: The multifaceted ways in which genomics can be valuable to clinicians, patients, families, and society are important for informing prioritization decisions by policy makers. This study aims to develop a standardized, cumulative, and preference-weighted genomic utility valuation (GUV) on a scale of 0% to 100%.

Methods: A multicriteria decision analysis was conducted with experts involved in policy, clinical, research, and consumer advocacy leadership in Australia for the valuation of policy priority indicators of genomic utility. The use of the GUV scale to support policy decisions is illustrated through a stylized example, and benchmark scoring thresholds of genomic utility were identified by mapping evidence from real-world health technology assessments leading to the public reimbursement of genomic testing in Australia onto the GUV scale.

Results: In total, 33 (73%) invited experts participated in the study. Clinical utility had the highest priority, followed by societal, diagnostic, economic, and family utilities. Improving health outcomes had the highest preference value (29.5%), followed by improving equity (22.6%), Having high diagnostic yield (22.2%), improving symptom management (15.5%), being cost saving (14.3%), having average diagnostic yield (13.1%), enabling access to clinical trials (12.3%), and enabling reproductive family planning (11.5%). Genomic testing scores from real-world health technology assessments ranged from 46% for syndromic and nonsyndromic intellectual disability to about 60% for mitochondrial conditions and genetic kidney diseases.

Conclusions: Comparisons of genomic utility across different clinical contexts may seem difficult because of the multiple criteria required to be weighted to support policy decisions. This comparison is now facilitated in a standardized manner with the GUV scale.

Keywords: genomics, preferences, priorities, rare diseases, utility.

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Highlights

- We developed the genomic utility valuation scale to enable a standardized measurement and scoring of genomic utility on a 0% to 100% scale based on 5 key policy priority indicators (clinical, diagnostic, economic, societal, and family utility).
- Improving health outcomes, having high diagnostic yield, being cost saving, improving equity, and enabling reproductive planning were the most preferred levels of the 5 indicators.
- Our work enables consistency in reporting and benchmarking of different genomic test indications, facilitating evidence-based research and policy decisions.

Introduction

Genomic utility is a multidimensional construct comprising different ways in which genomics can be useful to patients, families, clinicians, and society.[1] Within the medical literature, diagnostic utility, namely the establishment of an aetiologic diagnosis, has been the most-reported dimension of genomic utility,[2] reflecting the value of knowing the cause of the condition. Significant progress has been made in measuring and reporting the clinical and personal utility of genomics.[3,4] Clinical utility has traditionally described the clinical usefulness of genomics in improving patient health outcomes or the process of medical care, with personal utility describing the nonclinical usefulness of genomics to individuals and families, such as knowing how the condition is likely to progress or accessing peer support networks.[1] Although several variations of these definitions exist,[5] such as considering personal utility within a broader definition of clinical utility[1] or describing the overall importance of the different outcomes of genomics to individuals,[6,7] the multifaceted ways in which genomics can be valuable to patients, families, and society is widely recognized.[1]

Acknowledging the broad spectrum of health and nonhealth outcomes associated with genomic information and the potential of biasing prioritization and reimbursement decisions when these are not holistically considered,[7] ways of quantifying the value of genomics[6,8-12] and incorporating it into health economic evaluations to directly inform decision making[13-18] have been developed. However, decision making requires standardized evaluation criteria, and to this extent, efforts have been made to standardize the measurement of genomic utility. Hayeems et al[19] developed the Clinician-reported Genetic Testing Utility Index to assess the broader clinical utility of genetic and genomic testing from the perspective of clinicians. This has been validated in pediatric and adult rare disease populations.[20] Smith et al,[21-23] on the other hand have developed and validated the genetic utility scale to measure genomic utility from the perspective of parents of pediatric patients who have undergone clinically indicated genomic testing for diagnostic purposes and the perspective of adults undergoing risk-

based or population-based genomic screening. Similarly, Turbitt et al[24] have developed and validated the personal utility scale to measure the personal utility of genomic results.

Although these measures can provide valuable information about genomic utility, as reported by clinicians or perceived by patients and families, there are broader aspects of genomic utility required to support system-level decision making, including equity and cost-effectiveness, and information is further needed about the relative importance of the different components of utility. A recent commentary[25] proposed a wider framework to conceptualize genomic utility across multiple dimensions of relevance to patients, families, clinicians, health systems, and economies based on the guiding principle of a cumulative and preference-based valuation of utility.[25] This guiding principle was proposed to enable direct cardinal comparisons of different permutations of genomic utility that are likely to be generated across contexts. Currently, none of the existing measures of utility allows inferences about how better (or worse) one level of utility is from another, and none of the measures provide insights about the broader implications of genomics to society and the economy and how these could potentially be traded off against different levels of utility to facilitate policy making. Such frameworks are important to enable decision makers to consistently, objectively, and transparently answer questions such as "Is genomic utility of context 1 preferred to the genomic utility of context 2 and how much more (or less) is preferred?," "Is a lower level of clinical utility that improves equity preferred to a higher level of utility that exacerbates inequity of access to diagnosis?," and "How much genomic utility could be sufficient to support public reimbursement of genomics?"

This study reports on a multicriteria decision analysis (MCDA) conducted after a Delphi study to develop the genomic utility valuation (GUV) scale. In contrast to existing measures, the GUV scale enables a standardized measurement and valuation of genomic utility and its components that are required to support genomic research and policy through the cumulative and preference-based scoring of genomic utility on a 0 to 100 scale. The scale can be used by researchers in rare disease genomics research, including wider genomic screening, such as genomic newborn screening, to design evaluations for capturing the key outcomes that are important for implementation decisions. The scale can also be used by health technology assessment (HTA) bodies to support policy decisions based on standardized and quantifiable evidence of genomic utility. How the GUV scale can be used to support policy decisions is illustrated through a stylized example. We further map genomic utility evidence from different publicly funded rare disease genomic tests in Australia onto the GUV scale to indicate acceptable thresholds of genomic utility to support prioritization decisions.

Methods

A Delphi study was initially conducted to obtain consensus on indicators of utility among experts involved in policy, clinical, research, and patient/consumer advocacy leadership in Australia.[26] Experts (n = 45) were purposively identified through the Australian Genomics network, consisting of >100 organizations. We selected participants on the basis of experiences (ie, their leadership within a health service, state or federal government health department, HTA, patient/consumer advocacy, research, or their executive management of research). They were invited to participate by an invitation email. No further characteristics were noted. Activities included (1) 2 online survey rounds, programmed in REDCap, aiming to obtain expert consensus on different utility indicators that are considered a priority in determining the value of genomic testing for rare conditions in Australia and (2) a third round involving a preference valuation exercise aiming to derive the relative weight of the indicators considered important for informing health policy decisions. More information about the conceptualization of this work and the first 2 Delphi rounds can be found in the accompanying article.[26] This study reports on the third survey round of the experts. Ethical approval was granted by the Royal Children's Hospital Melbourne, Human Research Ethics Committee (HREC/89720/RCHM-2022).

The Survey

The first 2 Delphi rounds concluded 20 priority indicators of genomic utility included within the domains of test performance, clinical, prognostic, individual and family, economic, and societal.[26] To select the indicators relevant to policy decision making for inclusion in the MCDA (Table 1), the following decisions were made:

(1) The test performance domain from the Delphi study included clinical accreditation and diagnostic yield. Given that all tests introduced in the health system need to be clinically accredited, diagnostic yield was the only one retained. To ensure alignment with the concepts of utility and broader literature in the field, the indicator was renamed to diagnostic utility, which represented the first attribute in the MCDA (Table 1). The levels <10%, 30%, and >50% were selected to represent low, average, and high diagnostic yield.
(2) The prognostic domain in the Delphi study included 1 indicator (prognostic information). Given that prognostic information is a form of clinical utility, the indicator was included within the clinical utility attribute. The other indicators of the clinical domain were retained as levels of the clinical utility attribute apart from timely results, avoiding further investigations, and diagnostic purpose indicators. These indicators were dropped

Table 1. Genomic utility attributes and levels.

Attributes	Levels
Diagnostic utility	Low, <10% yield Average, 30% yield High, >50% yield
Clinical utility	Prognostic information only Access to nonclinical support, eg, disability care package Access to clinical trial(s) Improved symptom management only, ie, does not alter clinical outcomes Improved health outcomes, ie, mortality and morbidity
Family utility	Informs practical life planning or self-knowledge Cascade testing available to relatives Informs reproductive planning
Economic utility	Not cost-effective Cost-effective Cost saving
Societal utility	Likely to exacerbate inequity of access to diagnosis Unlikely to improve equity of access to diagnosis Likely to improve equity of access to diagnosis

as they are a function of the cost-effectiveness of the test, which was another attribute in the MCDA.
(3) The societal domain was retained but framed as a societal utility attribute with 3 levels (likely to exacerbate inequity, unlikely to improve equity, and likely to improve equity).
(4) The economic domain from the Delphi study included the cost to healthcare and cost to individuals/family indicators. Given that policy decisions are based on cost-effectiveness considerations, the attribute cost-effectiveness was included, which had 3 levels (not cost-effective, cost-effective, and cost saving).
(5) The family domain was framed as a family utility attribute and included the indicators of cascade testing and reproductive planning as attribute levels. The knowledge and practical outcomes indicators were merged into life planning and self-knowledge, given that they are both information-related outcomes.

Survey data were collected from August 30 to September 18, 2023. Before proceeding to the preference valuation exercise, participants were asked about the geographic location of their primary work, gender, age, area of expertise, and years of professional experience. The MCDA was performed using 1000Minds to elicit relative preference weights for genomic utility attributes and levels.[27] The 1000Minds software uses a potentially all pairwise rankings of possible alternatives method for creating and ranking dichotomous choice sets, each containing 2 attributes with levels differing between the pairs.[28] Choice tasks are adaptively presented, assuming preference transitivity, to minimize the number of choices presented while pairwise ranking all potential choice sets. Each choice task includes an option to report whether the 2 options are of equal value to the respondent. A choice task example is provided in Figure 1.

Analysis

The pairwise rankings of possible alternatives method generate preference values for each attribute level per respondent. The value of the least preferred attribute level is anchored at 0%, and the value of the most preferred attribute level represents the relative importance of the attribute. The relative importance of all attributes sums up to 100%. The preference values can then be compared across attributes and levels, representing the relative values of the levels to each other. This provides an insight into the priority of each attribute and the value gained when moving from one attribute level to another. In our analysis, preference values, attribute relative importance, and priority were reported across all experts and by the type of decision-making expertise (ie, policy, clinical, research, and consumer advocacy). Through a stylized example representing a scoring of 2 illustrative case studies of genomic testing for rare disease diagnosis, we demonstrated how genomic utility could be scored on a 0% to 100% scale to support health policy decision making. A benchmark threshold range is then estimated by mapping publicly funded items of genomic testing for heritable kidney diseases,[29] mitochondrial disorders,[30] and childhood syndromes[31] in Australia onto the GUV scale. The information about the level of genomic utility achieved in each of these settings across the GUV scale attributes and levels were extracted from publicly available summary documents from approved submissions to the Medical Services Advisory Committee or related peer-reviewed publications.[17,18,32]

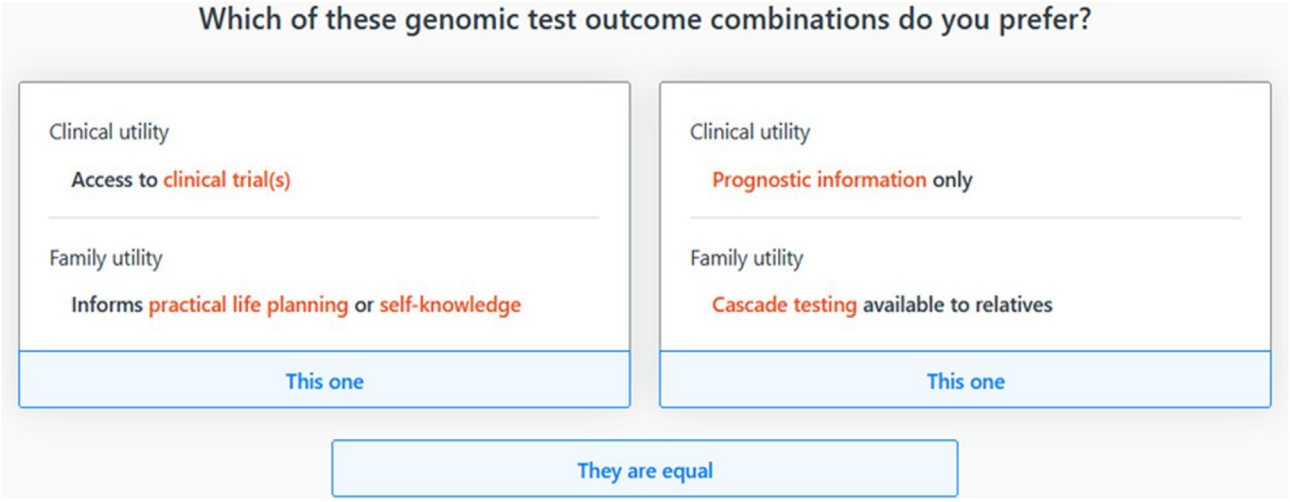
Results

In total, 33 (73%) invited experts participated in the MCDA survey. Respondents were primarily clinical experts (51.5%), female (66.7%), and aged between 41 to 60 years. Experts from research and policy equally comprised 36.4% of respondents, followed by consumer advocacy experts (12.1%). All experts had at least 6 years of experience in their field, with 39.4% of respondents having work experience of over 21 years. A detailed description of respondent characteristics can be found in Table 2.

As shown in Table 3, clinical utility was the attribute of highest priority, with a relative importance score of 29.5%, followed by societal utility (22.6%), diagnostic utility (22.2%), economic utility (14.3%), and family utility (11.5%). This means that clinical utility is approximately 3 times more valued than family utility, 2 times more valued than economic utility, and 1.3 times more valued than societal and diagnostic utilities. Improving equity was 2.4 times more valued than cost-effectiveness.

In terms of clinical utility, improving health outcomes, improving symptom management, and accessing clinical trials were the most preferred levels, with a preference value score of 29.5%, 15.5%, and 12.3%, respectively (Table 3). Improving equity (22.6%) or not exacerbating inequality (10.1%), having high (22.2%) or average (13.1%) diagnostic yield, being cost saving (14.3%) or

Figure 1. Choice task example.



cost-effective (9.3%), and informing reproductive planning (11.5%) were some of the valued levels of genomic utility. When disaggregating attribute relative importance scores by type of expertise (see Appendix Table 1 in Supplemental Materials found at https://doi.org/10.1016/j.jval.2024.11.014), a noticeable difference exists in the importance that consumer advocacy experts place on diagnostic utility, which is their most preferred aspect of genomic utility with a relative importance score of 33.4%. Consumer advocacy experts were found to value diagnostic utility almost twice as much compared with policy experts.

The preference values of Table 3 can be used to derive a cumulative and preference-based valuation of genomic utility. It is presumed that there are 2 scenarios in which genomic testing can be used to establish a diagnosis for individuals suspected of having a rare condition. The distribution of participants in the 2 scenarios receiving different aspects of clinical and family utility is listed in Appendix Table 2 (see Appendix Table 2 in Supplemental Materials found at https://doi.org/10.1016/j.jval.2024.11.014). We presume that individuals fall within only 1 level of these attributes. In scenario 1, we are 50% confident that equity will improve, there is an average diagnostic yield, and genomic testing is cost-effective. In scenario 2, we are 100% confident that equity will improve, there is high diagnostic yield, and genomic testing is cost saving. By weighting the preference value of each level with the outcomes achieved in the 2 scenarios, we can estimate a total score. In this case, scenario 1 had a score of 52.3% and scenario 2 a score of 87.2% (Fig. 2). When mapping genomic utility evidence from different publicly funded rare disease genomic tests in Australia onto the GUV scale, we see that genomic testing was prioritized with scores ranging from 46% for syndromic and nonsyndromic intellectual disability to approximately 60% for mitochondrial conditions and genetic kidney disease (Table 4), with the relevant supporting information provided in Appendix Table 3 (see Appendix Table 3 in Supplemental Materials found at https://doi.org/10.1016/j.jval.2024.11.014).

Table 2. Respondent characteristics.

Characteristics	Categories	n (%)
State or territory of primary work	Australia Capital Territory	1 (3.0)
	New South Wales	9 (27.3)
	Northern Territory	1 (3.0)
	Queensland	4 (12.1)
	South Australia	0 (0.0)
	Tasmania	1 (3.0)
	Victoria	15 (45.5)
	Western Australia	2 (6.1)
Gender*	Females	22 (66.7)
	Males	11 (33.3)
Age range†	31-40	3 (9.1)
	41-50	14 (42.4)
	51-60	11 (33.3)
	61-70	5 (15.2)
Type of expertise	Clinical	17 (51.5)
	Policy	6 (18.2)
	Consumer advocacy	4 (12.1)
	Research	6 (18.2)
Years of experience	0-5	0 (0.0)
	6-10	8 (24.2)
	11-20	12 (36.4)
	21+	13 (39.4)

*Nonbinary, Other, and 'Prefer not to say' were offered as options but were not selected.
†20-30, 71+, and "Prefer not to say" were offered as options but were not selected.

Table 3. Results.

Genomic utility	Preference value, %	SD, %	Relative importance, %	Priority ranking
Clinical utility				
Prognostic information	0.0	0.0	29.5	1
Nonclinical support	8.2	5.3		
Access to clinical trails	12.3	5.9		
Improved symptom management	15.5	7.3		
Improved health outcomes	29.5	6.1		
Societal utility				
Likely to exacerbate inequity	0.0	0.0	22.6	2
Unlikely to improve equity	10.1	7.0		
Likely to improve equity	22.6	7.9		
Diagnostic utility				
<10% yield	0.0	0.0	22.2	3
30% yield	13.1	6.6		
>50% yield	22.2	8.4		
Economic utility				
Not cost-effective	0.0	0.0	14.3	4
Cost-effective	9.3	4.9		
Cost saving	14.3	5.4		
Family utility				
Life planning and self-knowledge	0.0	0.0	11.5	5
Cascade testing	6.6	4.9		
Reproductive planning	11.5	5.3		

Discussion

This study aimed to develop a standardized framework for measuring and valuing the outcomes of genomic medicine (GUV scale). The GUV scale enables the systematic collection and weighting of the key outcomes of genomics to facilitate research and healthcare decision-making priorities and reimbursement decisions. In contrast to other measures of genomic utility from a clinical or patient perspective,[19,23] the GUV scale adopts a system-level decision-making perspective and enables a cumulative and preference-based valuation of genomic utility. Using an MCDA, we elicited expert preference values for the diagnostic, clinical, family, economic, and societal utility components of genomic utility, which enabled a scoring system of genomic utility on a scale of 0% to 100%. Experts prioritized the clinical utility of genomics and demonstrated a preference toward improving equity, high diagnostic yield, cost savings for the health system, and reproductive family planning.

A recent review and narrative synthesis of information extracted from formal HTAs of genetic testing for heritable conditions in Australia[33] highlighted key methodological and policy

Figure 2. Graphical representation of the genomic utility valuation scale scoring outcomes.

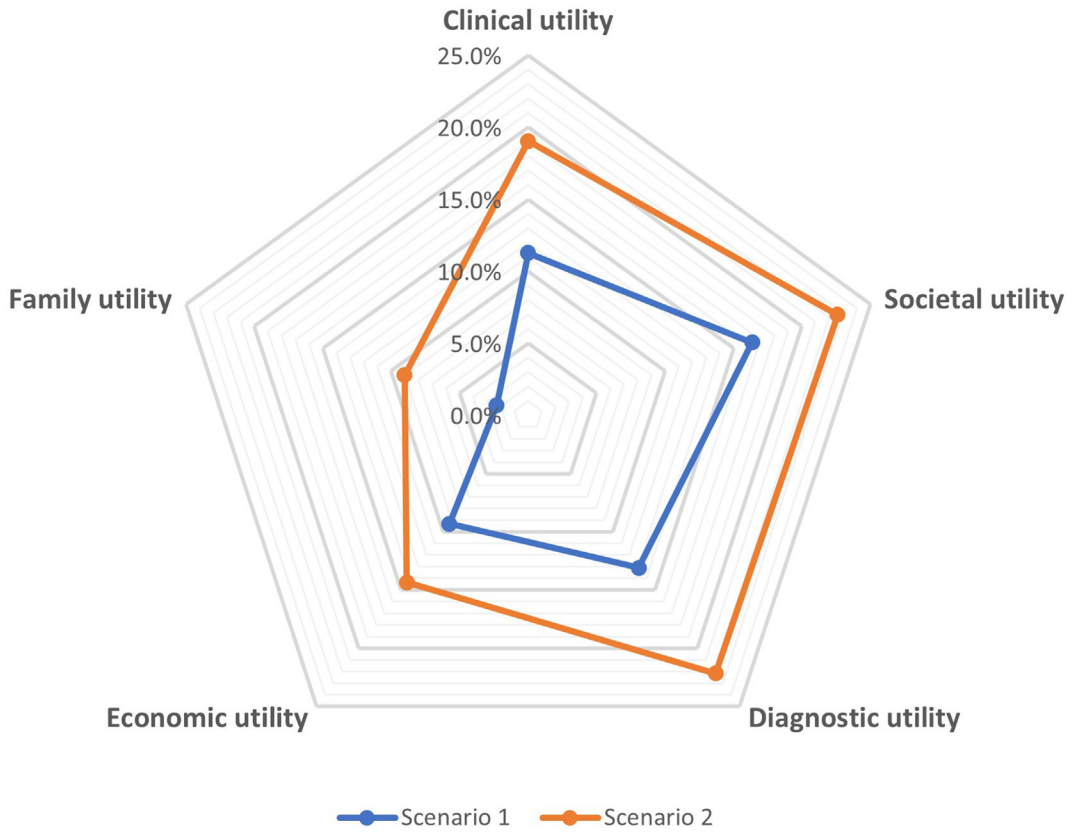


Table 4. Scoring of implemented cases of genomic testing in Australia.

Clinical utility	Preference value	Genetic kidney disease	Mitochondrial disorders	Childhood syndromes
Prognostic information	0.0	97	82	68
Nonclinical support	8.2	0	4	2
Access to clinical trials	12.3	0	0	0
Improved symptom management	15.5	0	2	30
Improved health outcomes	29.5	3	12	0
Societal utility				
Likely to exacerbate inequity	0.0	0	0	50
Unlikely to improve equity	10.1	0	0	50
Likely to improve equity	22.6	100	100	0
Diagnostic utility				
<10% yield	0.0	0	0	0
30% yield	13.1	72	12	0
>50% yield	22.2	28	89	100
Economic utility				
Not cost-effective	0.0	72	0	0
Cost-effective	9.3	0	100	100
Cost saving	14.3	28	0	0
Family utility				
Life planning and self-knowledge	0.0	52	51	45
Cascade testing	6.6	28	49	35
Reproductive planning	11.5	20	0	20
Total preference value		61	60	46

Note. All values are given in %.

evaluation challenges, including (1) incorporating preferences for the health and nonhealth outcomes of genomic testing, (2) measuring family utility, (3) ensuring equity of access to a geographically dispersed population, and (4) the role of patient and community needs in influencing the evidence thresholds for the prioritization decisions. The study concluded that these concepts should be considered for incorporation within the value assessment frameworks used by HTA agencies around the world.[33] The GUV scale enables these dimensions to be formally measured and weighted to support policy decisions. Heterogeneity of preferences was identified in experts involved in consumer advocacy who demonstrated a strong preference for knowing the genetic cause of conditions. This information aligns with the findings of preference-elicitation studies, in which the diagnostic outcomes in genomic medicine have also been demonstrated to be highly valued by patients and members of the public.[6,7,9,11,12,34,35] This information may merit further consideration from those involved in prioritization and reimbursement decisions.

The GUV scale provides a framework to support standardized policy decisions on the prioritization of genomic testing through the identification of genomic utility priority indicators. This was formally undertaken through a Delphi study followed by a preference valuation exercise among leading experts involved in policy, clinical, research, and consumer advocacy leadership in Australia through an MCDA. There are, however, limitations to our work. The sampling strategy using the national Australian Genomics network aimed at ensuring the representation of experts across different layers of decision-making expertise. Although experts were highly engaged and actively contributed to the Delphi and valuation studies, the modest sample size and composition of the final sample may mean that further work is required to validate the generalizability of the preference values in Australia and other jurisdictions. Further research is required to elicit societal preferences and to establish a broader understanding of the preference weighting of genomic utility by jurisdiction and expertise. Participating experts also considered a broad application of genomics, including both diagnostic and screening applications. However, the relative importance of the different genomic utility components may be different between diagnostic and screening applications of genomics. MCDA studies also involve a sequence of complex trade-offs to elicit preference weights. In our study, experts completed 15 to 43 choice tasks (27 choice tasks on average) because of the number of genomic utility indicators identified and the MCDA process required to establish preference weights. Indicatively, experts with less clear preference rankings completed more trade-offs for the software to establish preference values. Although these choice tasks were completed by expert decision makers in this field, it is unclear how the complexity and number of trade-offs may have affected the internal validity of the scale; therefore, further validation is required. Finally, preference values were generated by experts in Australia, and assessments of utility may be dependent upon health system structures or national prioritization processes. Further work from international experts is needed to support the application of the GUV scale to other countries.

Conclusions

In summary, we provide a standardized scale for measuring genomic test utility across multiple domains, allowing for consistency in reporting and benchmarking of different genomic test indications and facilitating evidence-based policy decisions. The GUV scale can become a valuable tool for providing evidence of genomic utility in a standardized manner to support research priorities and national HTA processes and recommendations.

Author Disclosures

Author disclosure forms can be accessed below in the Supplemental Material section.

Supplemental Material

Supplementary data associated with this article can be found in the online version at https://doi.org/10.1016/j.jval.2024.11.014.

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