The Parent PrU: a measure to assess personal utility of pediatric genomic results

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Abstract

Purpose: We aimed to adapt and validate an existing patient-reported outcome measure, the Personal Utility (PrU) scale, for use in the pediatric genomic context.

Methods: We adapted the adult version of the PrU and obtained feedback from six parents whose child had undergone sequencing. The resulting measure, the Parent PrU, was administered to parents of children in four pediatric cohorts of the Clinical Sequencing Evidence-Generating Research (CSER) consortium after they received their children's genomic results. We investigated the measure's structural validity and internal consistency.

Results: We conducted a principal-axis factor analysis with oblimin rotation on data from 755 participants to determine structural validity. These analyses yielded a 3-factor solution, accounting for 76% of the variance in the 16 items. We used Cronbach's *α* to assess the internal consistency of each factor: (1) child benefits (α = .95), (2) affective parent benefits (α = .90), and (3) parent control (α = .94).

Conclusions: Our evidence suggests that the Parent PrU scale has potential as a measure for assessing parent-reported personal utility of their children's genomic results. Additional research is needed to further validate the Parent PrU scale, including by comparing its findings with utility assessments reported by clinicians and children themselves.

Keywords: Health services evaluation; Patient-reported outcome measure; Perceived value; Psychometrics; Parent benefits

Introduction

Assessing the value of a new medical intervention is critical for integrating it into clinical care and making funding decisions.[1] The value or utility of clinical genomic sequencing has traditionally been assessed by evaluating impact on medical management or health outcomes (i.e. clinical utility).[2] However, there is growing recognition of the broadening concept of utility to include people's reports of the benefits of receiving results even in the absence of clinically useful information (personal utility).[3]

Consistent with this perspective, we developed the Personal Utility (PrU) scale to assess adults' perceptions of personal utility stemming from receiving their clinical genomic findings.[4] We conducted a systematic literature review in the first step of developing the PrU scale, which enabled us to produce a working definition of personal utility that included concepts such as self-knowledge, ability to plan for the future and mental preparation.[5] Our research to validate the PrU scale determined that for adult patients, personal utility is a multi-dimensional concept consisting of three factors: self-knowledge, reproductive planning, and practical benefits.[4] In the present study, we sought to extend measurement of personal utility to pediatric genomic healthcare.

Genomic technologies are particularly valuable in the pediatric rare disease context where 50-75% of rare diseases affect children, though many evade diagnosis through traditional genetic techniques.[6] Genomics offers a possible solution to diagnosing pediatric rare disease and the application of genomics in pediatrics is increasing in developed countries. Some governments with publicly funded healthcare such as Australia and the United Kingdom have allocated funds for genome or exome sequencing for children with suspected genetic conditions.[7, 8] This support reflects the potential impact of genomics on improving outcomes for children and their families. Metrics used to measure this impact should incorporate clinical dimensions of utility, as well as patient and family perspectives on the value of receiving this information.

Parents generally manage their children's clinical care and research participation,[9] and genomic results may have implications for both children and their biological parents. Furthermore, children are at a distinct life stage compared to adults, and genomic information may provide unique insights that guide tailored actions specific to these different life stages. Consequently, we posit that parents of children undergoing genome sequencing likely conceptualize personal utility differently from adults receiving their own genome sequencing information. Although parents are not likely to be able to accurately evaluate their children's perception of the utility of genomic results, they can consider and report their own perspectives on benefits in relation to themselves and their child's care.[10] Examples may include using genomic information to inform plans for children's education,[11] and potentially fulfilling the duty of being a responsible parent.[12]

The objective of this study was to adapt the adult PrU scale to the pediatric context and validate the scale among parents receiving their children's genomic results.

Methods

Initial Scale Development

We described the initial scale development in detail in a prior publication.[4] Briefly, elements of personal utility in genomics were identified through a systematic literature review.[5] These elements were refined through a modified Delphi assessment with adult participants of a genomic sequencing study.[13] We assembled the PrU scale by converting these refined elements to items. We presented the items as statements with the stem *"Please indicate how useful you find the following outcomes of your test result"*, and respondents assessed each item with a 7-point, Likert-type scale (1=Not at all Useful to 7=Extremely Useful).

Development and refinement of a parent version of the PrU scale

We converted the adult version of the PrU scale to a parent version, adapting the wording (e.g., “help with my life planning” converted to “help with my child’s life planning”). We obtained feedback on this parent version of the PrU scale. We presented the scale online using SurveyMonkey to six parents enrolled in the Stanford Center for Undiagnosed Diseases whose child had undergone genome sequencing. These six parents were a mix of those with and without a diagnosis for their child and had received genomic results or were awaiting results. Parents responded anonymously; therefore, the specific characteristics of these six parents are not available. However, they were from a wider pool of 13 parents who were white (n=11), Black (n=1) or Asian (n=1). Of these, most identified as Not Hispanic or Latino (n=8). All 13 reported English as their primary language. We sampled from this pool of 13 parents until no new changes or recommendations to modify items were uncovered.[14]

Parents first viewed the scale and read introductory instructions explaining that we were seeking their input about non-medical reasons that parents may value from their child’s genetic information (see supplemental information Box S1). Next, they rated each item, were asked to describe the item in their own words (“what do you think this item is asking?”), and indicated whether it should be included in a study of personal utility (response options: “yes” or “no”). We refined items as a result of this feedback. For example, the item “make me feel good for contributing to the community” was altered to “feel good about helping the medical community” as one parent was unsure of the meaning, and another parent suggested more specificity was required.

We partnered with the Clinical Sequencing Evidence-Generating Research (CSER) consortium pediatric cohorts to validate the scale. Upon sharing the scale with CSER investigators, further refinement occurred to produce the parent version of the PrU scale with 17 items administered for this study (Supplemental Table S1). Changes included altering wording of the stem or item and deleting some items. Further information about the changes and rationale is available in previous publications.[4, 15]

Sample and data collection for exploratory factor analysis

We administered the 17-item Parent PrU scale to parents of CSER participants 0–4 weeks after disclosure of genomic results. Item order was randomized. Data from four CSER sites were included (NYCKidSeq, P3EGS, SouthSeq, and NCGENES 2). The patient populations at these sites were comprised of children undergoing diagnostic sequencing with suspected genetic conditions. Participants received positive, uncertain or negative results. Where more than one parent completed the survey, only responses from the "primary parent" were included in this analysis. Each site had slightly different definitions of the primary parent, though in general this was the parent primarily responsible for their child's care and who attended all research visits.

We gathered descriptive variables including demographic characteristics (age, race and sex of child, parent education and race) and time (in weeks) between result return and scale completion. We also used responses from the 4-item positive feelings subscale of the FACToR (Feelings About genomiC Testing Results) scale to test convergent validity (see statistical analysis below). The positive feelings subscale asks respondents to report on the psychological impact of their child's genomic results,[16] including feeling happy about their child's genetic test result, feeling relieved about their child's genetic test result, feeling they understood clearly their child's choices for disease prevention or early detection, and feeling that the information they received from their child's genetic test result was helpful for planning for the future. Response options ranged from 4=not at all, to 0=a great deal. Note that FACToR items measure negative psychological impact, so we reverse scored the items to facilitate interpretation; higher scores indicate a more positive psychological impact.

Statistical analysis

We started by examining the descriptive statistics of the items to check for floor or ceiling effects or differential non-response. We then employed exploratory factor analysis to assess the

structural validity of the 17 items that purport to measure personal utility. We determined the suitability of the items for factor analysis by using the Kaiser–Meyer–Olkin (KMO) test, requiring a value of at least .70 to be considered adequate.[17] We extracted the factors using principal-axis factoring, which emphasizes the shared variance among the items, and we used direct oblimin rotation to make it easier to interpret the extracted factors while also taking into account the correlation among the factors using the pattern matrix. We decided on the number of factors to extract based on eigenvalues, scree plot, and theoretical soundness of potential factor solutions (i.e., seeking a solution that explained a substantial amount of variance and had a logical pattern with simple structure).[18] We followed the factor-loading rules that satisfactory variables load onto their primary factor above .40; load onto alternative factors below .30; and there is a difference of at least .20 between primary and alternative factor loadings.[19] We examined any item with a communality score of less than .40 to consider why it was included and whether the item should be dropped.[20] We named the factors based on the items with the highest loadings (particularly any items over .90) on the factor.[18]

Next, we determined the internal consistency of each factor using Cronbach's alpha. Finally, we evaluated the correlation of the positive feelings subscale of the FACToR with PrU subscale scores to assess convergent validity using Pearson's correlation. We used the positive feelings subscale given the similarity of the subscale with personal utility (e.g., one FACToR item asks about feeling that the genetic test result is helpful for planning for the future). We expected that high personal utility scores would be positively correlated with high positive feelings scores. Other FACToR subscales (insurance coverage, uncertainty, negative emotions) were not appropriate for convergent validity testing.

Results

Respondent characteristics

The Parent PrU scale was completed by 781 participants in the four CSER studies. There were 26 participants who did not provide responses to all PrU items and were removed from the analysis, leaving 755 valid responses (Table 1). On average, there were 5 missing responses per item (5/781; 0.6%), with a range of 1 to 11. Item 13 ("help my child to use social programs, like resources and services") had the highest number of missing responses.

The children of the parents who completed the scale were mostly male (*n*=438, 58.0%), aged 7 years and 3 months on average (*SD*=4 years and 4 months), and described by parents as White (*n*=212, 32.0%), Hispanic/Latino (*n*=230, 24.7%) and/or (could select all that apply) Black (*n*=140, 21.1%). Most parent responders were educated to some post-high school or beyond (*n*=627, 83.0%). Responses were provided an average of 1.1 weeks after receiving genomic results (*SD*=4.1), ranging from 0 weeks to 55 weeks.

Exploratory factor analysis results

We examined item distributions (Supplemental Table S2) and found no evidence of strong ceiling or floor effects. The KMO test across the items was .96, indicating that the items share a great deal of common variance and would be appropriate for factor analysis.

We investigated the feasibility of a two and three factor solution based on the criteria described above (also see Figure S1 of supplemental information). We did this by examining the pattern matrix loadings in addition to the theoretical rationale of factors. The two-factor solution indicated one factor mostly concerning benefits for the child and one factor centered on aspects of utility for the parent. However, two items that we considered parent benefits loaded with factor one. These items were PrU9 “Help me feel more in control of my child's health” and PrU10 “Help me feel more in control of my child's life”. In the three-factor solution, items PrU9 and PrU10 loaded separately onto a third factor. We settled on the three-factor solution that accounted for 76% of the variance in the items.

The first factor accounted for 64% of the variance and the nine items that clearly loaded on this factor were centered around benefits to the child. We named this factor **child benefits**. The second factor accounted for an additional 5% of the variance and consisted of five items that focused on affective benefits to the parent (**affective parent benefits**). The third factor accounted for an additional 3% of the variance and consisted of two items related to **parent control** (Table 2). One item cross-loaded: PRU16 “Feel good about having information for family members”. We placed this item in factor two “affective parent benefits” for three reasons: the factor-loading value (.34) for factor one was close to our cut-off (.30); the item loaded highly on factor two (.68 – a difference greater than .20 between primary and alternative factor loadings); and the factor conceptually fit as an affective parent benefit.

We removed item PrU4 (“Use for testing a future pregnancy, if appropriate”) which had low communality (.3). We determined the low communality may be due to the wording being unclear whether the responder was required to consider the parent’s or child’s future pregnancy. The child benefits factor correlated with the affective parent benefits factor (r=.75) and parent control factor (r=.76), and the affective parent benefits factor correlated with the parent control factor (r=.68).

Overall score, internal and external consistency

The Cronbach’s α for all 16 PrU items was .96. We used the mean of the 16 items to generate a total score, and for this sample the mean was 5.2 (SD=1.5; response range was from 1 to 7), representing slightly higher than the response option “useful”. The Cronbach’s α and descriptive statistics of the PrU subscales (the mean of the items that comprised each factor) were: child benefits, α=.95, $mean$=4.95, SD=1.7; affective parent benefits, α=.90, $mean$=5.62, SD=1.4; parent control, α=.94, $mean$=4.94, SD=1.96.

There was a positive association between the FACToR positive feelings subscale and all PrU subscales; child benefits (r= .59, p<.001), affective parent benefits (r= .60, p<.001), and

parent control (r= .56, p<.001). Individuals with higher PrU subscale scores reported a greater level of positive feelings towards their genetic test results.

Discussion

Our findings provide initial validation evidence for measuring parent-reported personal utility from their children's genomic test result. The structural validity results from our study suggest that parent personal utility consists of three factors: child benefits, affective parent benefits, and parent control.

Theorized models of utility suggest more complex structures including domains such as affective, cognitive, behavioral, and social.[5, 21] While we included items across these domains, our results suggest parents' conceptualization of the personal utility of their children's genomic test is more holistic in nature, denoted according to the beneficiary (self or child) of the utility.

The extent to which parent-reported personal utility is correlated with clinical utility is unknown as validated assessments of personal utility of genomic results have been lacking. We suggest our Parent PrU scale could be used alongside more traditional and tangible assessments of utility, allowing for comparisons between personal and clinical utility. It may be possible to measure items related to medical management through parent-reported clinical utility,[21] as well as more objectively such as tracking onward referrals or by clinician-reported utility.[22]

Another commonly used outcome scale in genomics is the Genomic Outcome Scale (GOS).[23] This is six-item scale was developed from the Genetic Counselling Outcome Scale (GCOS). Both the GOS and GCOS focus on the theoretical construct of empowerment. Empowerment and utility are distinct but related concepts. Empowerment focuses on the process of giving individuals the knowledge, skills and resources they need to take control of their health. Two items of the GOS are analogous to items in our personal utility scales – "I am able to make plans for the future" and "I can make decisions about the condition that may

change my future or my child(ren)'s future". Further work could test the convergent validity of the Parent PrU using the GOS. The GOS was not a harmonized measure administered to all CSER participants.

Further efforts should develop child or adolescent self-assessment measures of utility. While parents are generally the key decision makers in their children's care, the requirement for adolescent assent and the importance of including children in decisions about their own healthcare is well established.[9] Parents generally lack the lived experience of their child's condition, particularly in the context of rare genetic conditions. Emerging work in the conceptualization of severity is one such example that highlights the importance of understanding the reality of those living with genetic conditions.[24] Such work indicates that individuals living with early-onset genetic conditions often see their condition as part of their identity and report experiencing good health/wellbeing.

Limitations and Future Work

Our scale was created for use in CSER consortium studies, and as such, some practical decisions were taken during its development to make it suitable for that specific context. These decisions included removal of certain items to decrease burden on participants. Future work could explore addition or refinement of items that may be relevant to the specific context in which the scale is administered. This could also include further cognitive testing involving a broader range of people, incorporating diversity in race/ethnicity, geographic origin, and literacy levels.

Our intention was to measure experienced utility, though the 4-week response window may have resulted in too short a timeframe for some items to be experienced. This is unlikely to impact our validation analysis. However, additional testing of the scale in various contexts and time-points is needed to ensure that it captures the complete range of personal utility and to assess changes in personal utility over time.

We present the first of many required steps to develop a psychometrically valid scale. Future work should involve exploring the interpretability of the scale, including the minimum important change or difference in personal utility, as well as assessing test-retest reliability and responsiveness by examining longitudinal changes.[25]

The Parent PrU may be relevant for use in studies exploring parent-reported personal utility of genomic sequencing in contexts where this technology is gaining momentum such as newborn screening and the neonatal/pediatric intensive care unit (NICU/PICU). For example, parents have qualitatively reported similar dimensions of personal utility from ultrarapid genomic testing for critically unwell children.[26, 27] Validation of our scale in the rapid or ultrarapid context is recommended given the potential for some context specific factors to influence perceptions of personal utility such as heightened emotions and time pressures.

Finally, future research could use our scale to explore associations between parent-reported personal utility and health outcomes for children and families. Such evidence could provide support for development of interventions to improve perceptions of personal utility or other clinical applications.

Conclusions

We found initial validation evidence that supports a measure of parent-reported personal utility from their children's genomic test. We labeled the subscales: child benefits, affective parent benefits, and parent control. Our data suggest that parents' conceptualization of personal utility is holistic, focused on the beneficiary (self or child) of the utility. Future work is needed to continue validation efforts of the Parent PrU scale and compare with clinician reported and child self-assessments of utility.

Data availability: Data are available on request.

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Ethics Declaration: The study protocols to collect data analyzed in this report were approved by the Institutional Review Boards of Icahn School of Medicine at Mount Sinai, University of California San Francisco, University of Alabama at Birmingham and University of North Carolina, Chapel Hill. Informed consent was obtained from all participants.

*Conflict of Interest***:** The authors declare no conflict of interest.

Tables

Table 1. PrU parent descriptive statistics *N*=755

Variable		Frequency	Proportion (%)
Cohort name (primary site)			
	NYCKidSeq (Icahn School of Medicine at Mount Sinai)	457	60.5
	P3EGS (University of California San Francisco)	171	22.6
	SouthSeq (Hudson-Alpha Institute for Biotechnology)	103	13.6
	NCGENES 2 (University of North Carolina)	24	3.2
Age	*Mean*=7 years & 3 months, *SD*=4 years & 4 months, *Min*=0 months *Max*=21 years		
Child's sex			
	Male	438	58.0
	Female	317	42.0
Highest level of parent's education			
	Less than high school (less than 9th grade)	31	4.1
	Some high school (9th to 12th grade), no diploma	82	10.9
	High school graduate (diploma or GED equivalent	162	21.5
	Some post-high school training (college or occupational, technical, or vocational training), no degree or certificate	123	16.3
	Associate (2-year) college degree, or completed occupational, technical, or vocational program and received degree or certificate	91	12.1
	Bachelor's degree (for example: BA, AB, BS)	138	18.3
	Graduate or professional degree (for example: MA, MBA, JD, MD, PhD)	113	15.0

Unknown	15	1.9
Race of child		
White or European American	212	32.0
Hispanic/Latino(a)	230	24.7
Black or African American	140	21.1
Asian	46	6.9
American Indian, Native American, Alaska Native	11	1.7
Prefer not to answer	9	1.4
Unknown/none of these fully describe my child	8	1.2
Middle Eastern of North African/Mediterranean	5	0.8
Native Hawaiian/Pacific Islander	1	0.2
Race of Parent 1		
Hispanic/Latino(a)	288	36.8
White or European American	258	33.0
Black or African American	125	16.0
Asian	62	7.9
American Indian, Native American, Alaska Native	18	2.3
Middle Eastern of North African/Mediterranean	10	1.3
Prefer not to answer	9	1.1
Unknown/none of these fully describe me	8	1.0
Native Hawaiian/Pacific Islander	5	0.6
Race of Parent 2		
Hispanic/Latino(a)	272	43.0
White or European American	174	27.5
Black or African American	91	14.4

Asian	48	7.6
American Indian, Native American, Alaska Native	15	2.4
Unknown/none of these fully describe me	11	1.7
Middle Eastern of North African/Mediterranean	11	1.7
Prefer not to answer	7	1.1
Native Hawaiian/Pacific Islander	4	0.6

Weeks post return of results scale completed: *Mean*=1.1, *SD*=4.1, *min*=0*, max*=55

Participants could select more than one Race category.

Table 2. Factor structure and factor loadings

PrU items	Factors		
	1 Child benefits	2 Affective parent benefits	3 Parent control
PRU2 Inform plans for my child's school or career	.96		
PRU1 Help with my child's life planning	.89		
PRU5 Help me or our family mentally prepare for the future	.79		
PRU13 Help my child to use social programs, like resources and services	.68		
PRU3 Inform my child's decisions about having children	.63		
PRU6 Help to better understand my child's health	.62		
PRU7 Contribute to my child's self-knowledge	.61		
PRU14 Improve communication with my family members	.59		
PRU8 Help me cope with my child's health risks	.59		
PRU15 Feel good about helping the medical community		.82	
PRU17 Feel good about taking responsibility for my child's health		.74	
PRU11 Simply to provide information		.72	
PRU16 Feel good about having information for family members	.34	.68	
PRU12 Satisfy my curiosity about my child		.61	
PRU9 Help me feel more in control of my child's health			.93
PRU10 Help me feel more in control of my child's life			.76

Factor loadings above .30 are reported.

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