THE PRU AND PARENT PRU

The Personal Utility and Parental Personal Utility Scales: Measures to assess personal utility of genomic results

User Guide

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INTRODUCTION AND BACKGROUND

1.1   PERSONAL UTILITY DEFINITION

The term "personal utility" pertains to the patient-centred aspects of genomic medicine, highlighting the nonmedical benefits that individuals derive from genomic testing, which encompasses their thoughts, emotions and behaviour's. Previous studies have established that personal utility involves nonmedical perceived benefits such as increased self-awareness, a deeper understanding of one's condition, acts of altruism and enhanced coping abilities. (Kohler et al, 2017a; Kohler et al, 2017b; Turbitt et al, 2023)

1.2   IMPORTANCE OF MEASURING PERSONAL UTILITY

Measuring personal utility in genomic medicine is essential for patient-centred care and promoting patient autonomy. The decision to pursue genomic testing is complex and multifaceted and therefore clinical and personal utility both play a critical role in the decision-making process for patients. Accurately measuring and assessing genomic medicine outcomes, including personal utility, enhances our holistic understanding of the outcomes experienced by patients and their families who undergo genomic testing (Kohler et al, 2017a; Kohler et al, 2017b; Turbitt et al, 2023).

DEVELOPMENT OF PRU/PARENT PRU

2.1   INITIAL DEVELOPMENT OF PRU

A systematic review was conducted to inform the preliminary scale, which included 35 items representing 15 elements of personal utility. Following a modified Delphi assessment, in which the 35 items were rated based on a Likert-type scale, 24 items representing personal utility remained. To assemble the PrU scale, the 24 items could be rated on a 7-point Likert-type scale with the stem "*Please indicate how useful you find the following outcomes of your test result*" (Appendix 5.1).

The 24-item scale was piloted with 24 adults who had undergone genome sequencing, including 19 over-the-phone enabling collection of more comprehensive verbal feedback, allowing for collection of open response data. Following data analysis from the piloting, and recommendations from The Clinical Sequencing Evidence Generating Research (CSER) investigators, items which represented overlapping concepts were removed, and 17 items of personal utility remained (Appendix 5.2)

2.2   VALIDATION OF PRU

The 17-tem scale was administered to CSER participants at follow-up (0-4 weeks following disclosure of genomic results), over two sites. Responses of 841 participants who had received genomic results were investigated using exploratory factor analysis, which resulted in a final 14-item scale with a 3-factor solution (self-knowledge, reproductive planning and practical benefits) (Appendix 5.3)

Details of the statistical analysis used for the validity of the final PrU scale can be found in the published paper (Turbitt et al, 2023).

2.3   INITIAL DEVELOPMENT OF PARENT PRU

The initial development of the PrU scale is described in 2.1; The scale was then adapted for parents of children who have undergone genomic testing by adjusting the wording of each item (i.e. "help with my life planning" converted to "help with my child's life planning"). Parent feedback was obtained via SurveyMonkey from 6 parents whose children had previously undergone genomic testing, enrolled via Stanford Center for Undiagnosed Diseases. The 17 items as outlined above, were then refined based on parent feedback, and feedback from the CSER investigators (Appendix 5.4)

2.4   VALIDATION OF PARENT PRU

The refined 17-item scale was administered to CSER paediatric cohorts at follow-up (0-4 weeks following disclosure of genomic results), over four sites. Responses from 755 participants whose child has undergone genomic sequencing were investigated using exploratory factor analysis, which resulted in a final 16-item scale with a 3-factor solution (child benefits, affective parent benefits, parent control) (Appendix 5.5)

Details of the statistical analysis used for the validity of the final Parent PrU scale can be found in the published paper (Turbitt et al, 2024).

2.5   LIMITATIONS OF PRU

There may be limitations in the generalisability of PrU as the sample population was more highly educated than the general population and was developed for the use in CSER consortium studies. Further work could explore addition or refinement of items that may be relevant to the specific context or time-points in which the scale is administered.

2.6   LIMITATIONS OF PARENT PRU

Parent PrU scale was created and validated for use in CSER consortium studies and therefore further work could explore addition or refinement of items that may be relevant to the specific context or time-points in which the scale is administered.

USING AND ADMINISTRATION OF PRU

3.1   THE PRU AND PARENT PRU MEASURE PERSONAL UTILITY

The PrU and Parent PrU measure the personal utility concepts that are most relevant to the end users of genomic testing. For the PrU, this includes items related to self-knowledge, practical benefits, and reproductive planning. For the Parent PrU this includes items related to child benefits, affective parent benefits and parent control.

3.2   ADMINISTERING THE SCALE

The PrU and Parent PrU are intended for adults or parents of children who have received genomic test results and are to be either self-administered (i.e. online/paper survey) or

administered by a clinician/researcher. The items should be randomised prior to administering to avoid order effects.

3.3   PRU SCORING

The PrU is scored on a 7-point Likert-type scale, where 1 is "not at all useful" and 7 is "extremely useful". If an item is not responded to, that item is removed from the scoring analysis and is not treated as a "zero". *Self-knowledge* is scored based on the average score of items 5,6,9,10,13,14. *Reproductive planning* is scored based on the average score of items 2 and 3. *Practical Benefits* is scored using average of score items 1,4,7,8,11 and 12. An overall score of personal utility could also be calculated, taking the average score of all items. The decision to use the overall score, or subscale scores depends on the user's research question or objective.

3.4   PARENT PRU SCORING

Parent PrU is scored on a 7-point Likert-type scale, where 1 is "not at all useful" and 7 is "extremely useful". If an item is not responded to, that item is removed from the scoring analysis, and is not treated as a "zero". *Child Benefits* is scored based on the average score of items 1, 2, 3, 4, 5, 6, 7, 12, 13. *Affective parent benefits* are scored based on the average score of items 10, 11, 14, 15, 16. *Parent control* is scored using an average of score items 8 and 9. An overall score of personal utility could also be calculated, taking the average score of all items. The decision to use the overall score, or subscale scores depends on the user's research question or objective.

3.5   INTERPRETATION

The average score for each subscale or the overall score follows the same Likert-type scoring system in that 1 is "not at all useful" and 7 is "extremely useful".

FREQUENTLY ASKED QUESTIONS

4.1   WHEN SHOULD I USE PRU VS. PARENT PRU

PrU is validated for adults (18+) who have received genomic test results. Parent PrU is validated in the paediatric genomic context, for parents of children aged 21 and younger who have received genomic test results.

4.2   WHY MEASURE PERSONAL UTILITY?

Recognising and measuring personal utility as an outcome of genomic testing promotes a more holistic and patient-centred framework to assessing a person's health.

4.3   WHAT IS THE DIFFERENCE BETWEEN CLINICAL AND PERSONAL UTILITY?

Broadly, clinical utility is the concept that genomic testing will lead to improved health outcomes (i.e. therapeutic management and prognosis) (ACMG, 2015). Personal utility is

a patient-centred construct in which patients derive non-medical (non-clinical) benefits from genomic test results (Kohler et al, 2017a; Kohler et al, 2017b). Refer to Clinician-reported Genetics testing Utility InDEx (C-GUIDE) (Hayeems et al, 2022) for validated resources to measure clinical utility of genomic testing.

4.4 WHAT ARE EXAMPLES OF APPLICATIONS FOR PRU/PARENT PRU?

The PrU and Parent PrU measures the personal utility of genomic testing. Outcome measures of PrU have potential applications in both a clinical and research setting, as outlined in the publications (Turbitt et al, 2023; Turbitt et al, 2024) and may include:

- Clinicians or researchers may administer the PrU or Parent PrU to anticipate what benefits their patients may experience from genomic testing and inform discussions around test decision making

- Researchers assessing the value of genomic medicine in a specific context or the effectiveness of novel interventions and protocols may use PrU as an outcome measure

- Parent PrU may be relevant for use in studies exploring parent-reported personal utility of genomic sequencing, such as newborn screening and the neonatal/paediatric intensive care unit.

4.5 DO I NEED TO RANDOMISE THE ORDER OF THE ITEMS?

Yes, to avoid order effects the person administering the survey should first randomise the order of the items. If using an online survey, you can typically request this happen automatically.

4.6 AM I ABLE TO MODIFY PRU/PARENT PRU OR MAKE CHANGES TO THE ITEMS?

Please do not modify items of the PrU or Parent PrU. If item wordings are changed or removed/added in the absence of validation, the legitimacy of the findings produced will be compromised. If you would like to validate PrU/Parent PrU for a different clinical population, please contact Dr Erin Turbitt (erin.turbitt@uts.edu.au)

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PERSONAL UTILITY SCALE (PrU) —Original version 24 items

Set survey to randomise items to avoid order effects

Please indicate how plausible you find the following outcomes of genome sequencing:

	Very implausible	implausible	Somewhat implausible	Neutral	Somewhat plausible	Plausible	Very plausible
Help one understand one's health condition better							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Help one cope with one's health risks							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Improve one's self-knowledge							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Make one feel good for contributing to research

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Help one or one's family mentally prepare for the future

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Make one feel good for providing knowledge to one's family

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Sequencing results are valuable simply because they provide information

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Improve one's understanding of one's family

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Inform one's decisions about having children

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Use for prenatal testing to ensure children do not have condition

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Allow one to organize long-term care

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Satisfy one's curiosity

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Help one feel more in control of oneself	1	2	3	4	5	6	7
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Make one nervous about discrimination; insurance, employment							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Motivate one to get one's affairs in order							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Spur increased communication with one's family members							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Sequencing results are valuable no matter what the results are							
How plausible is this outcome for you?	1	2	3	4	5	6	7

How plausible is this outcome for others	1	2	3	4	5	6	7

Spur feelings of responsibility for one's children's health risks

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Lead to greater support from one's friends and family

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Allow one to take advantage of social programs; advocacy

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Help one feel more in control of one's life situation

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

Help one live more fully

How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Inform one's plans for school or career							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7
Give one a false sense of security							
How plausible is this outcome for you?	1	2	3	4	5	6	7
How plausible is this outcome for others	1	2	3	4	5	6	7

PERSONAL UTILITY SCALE (PrU) — 17 items

Set survey to randomise items to avoid order effects

Please indicate how useful you find the following outcomes of your test result:

	Not at all useful	A little useful	Somewhat useful	Neutral	Useful	Very useful	Extremely useful
Help with life planning	1	2	3	4	5	6	7
Inform my plans for school or career	1	2	3	4	5	6	7
Inform my decisions about having children	1	2	3	4	5	6	7
Use for testing a future pregnancy, if appropriate	1	2	3	4	5	6	7
Help me or my family mentally prepare for the future	1	2	3	4	5	6	7
Help to better understand my health	1	2	3	4	5	6	7
Contribute to my self-knowledge	1	2	3	4	5	6	7
Help me cope with my health risks	1	2	3	4	5	6	7
Help me feel more in control of my health	1	2	3	4	5	6	7
Help me feel more in control of my life	1	2	3	4	5	6	7

Simply to provide information	1	2	3	4	5	6	7
Satisfy my curiosity	1	2	3	4	5	6	7
Help me to use social programs, like resources and services	1	2	3	4	5	6	7
Improve communication with my family members	1	2	3	4	5	6	7
Feel good about helping the medical community	1	2	3	4	5	6	7
Feel good about having information for my family members	1	2	3	4	5	6	7
Feel good about taking responsibility for my children's health	1	2	3	4	5	6	7

5.3　PRU VALIDATED 14-ITEM SCALE

PERSONAL UTILITY SCALE (PrU) —FINAL VERSION 14 items

Set survey to randomise items to avoid order effects

Please indicate how useful you find the following outcomes of your test result:

	Not at all useful	A little useful	Somewhat useful	Neutral	Useful	Very useful	Extremely useful
1. Help with life planning	1	2	3	4	5	6	7
2. Inform my decisions about having children	1	2	3	4	5	6	7
3. Use for testing a future pregnancy, if appropriate	1	2	3	4	5	6	7
4. Help me or my family mentally prepare for the future	1	2	3	4	5	6	7
5. Help to better understand my health	1	2	3	4	5	6	7
6. Contribute to my self-knowledge	1	2	3	4	5	6	7
7. Help me cope with my health risks	1	2	3	4	5	6	7
8. Help me feel more in control of my life	1	2	3	4	5	6	7
9. Simply to provide information	1	2	3	4	5	6	7
10. Satisfy my curiosity	1	2	3	4	5	6	7

11. Help me to use social programs, like resources and services	1	2	3	4	5	6	7
12. Improve communication with my family members	1	2	3	4	5	6	7
13. Feel good about helping the medical community	1	2	3	4	5	6	7
14. Feel good about having information for my family members	1	2	3	4	5	6	7

Scoring notes:

Subscale 1: <u>self-knowledge</u> – compute average score of items 5, 6, 9, 10, 13, 14

Subscale 2: <u>reproductive planning</u> – compute average score of items 2 and 3

Subscale 3: <u>practical benefits</u> – compute average score of items 1, 4, 7, 8, 11, 12

5.4 PARENT PRU VALIDATED 16-ITEM SCALE

Citation: Turbitt, E., Kohler, J.N., Brothers, K.B., Outram, S.M., Rini, C., Sahin-Hodoglugil, N., Leo, M.C., Biesecker, B.B. (2024) The Parent PrU: A measure to assess personal utility of pediatric genomic results. *Genetics in Medicine*, *16*(1), 100994. 10.1016/j.gim.2023.100994

PARENTAL PERSONAL UTILITY SCALE (PrU) —FINAL VERSION 16 items

Set survey to randomise items to avoid order effects

Please indicate how useful you find the following outcomes of your child's test result:

		Not at all useful	A little useful	Somewhat useful	Neutral	Useful	Very useful	Extremely useful
1	**Help with my child's life planning**	1	2	3	4	5	6	7
2	**Inform plans for my child's school or career**	1	2	3	4	5	6	7
3	**Inform my child's decisions about having children**	1	2	3	4	5	6	7
4	**Help me or our family mentally prepare for the future**	1	2	3	4	5	6	7
5	**Help to better understand my child's health**	1	2	3	4	5	6	7
6	**Contribute to my child's self-knowledge**	1	2	3	4	5	6	7
7	**Help me cope with my child's health risks**	1	2	3	4	5	6	7
8	**Help me feel more in control of my child's health**	1	2	3	4	5	6	7
9	**Help me feel more in control of my child's life**	1	2	3	4	5	6	7
10	**Simply to provide information**	1	2	3	4	5	6	7

11	**Satisfy my curiosity about my child**	1	2	3	4	5	6	7
12	**Help my child use social programs, like resources and services**	1	2	3	4	5	6	7
13	**Improve communication with my family members**	1	2	3	4	5	6	7
14	**Feel good about helping the medical community**	1	2	3	4	5	6	7
15	**Feel good about having information for family members**	1	2	3	4	5	6	7
16	**Feel good about taking responsibility for my child's health**	1	2	3	4	5	6	7

Scoring notes:

Subscale 1: <u>Child benefits</u> – compute average score of items 1, 2, 3, 4, 5, 6, 7, 12, 13

Subscale 2: <u>Affective parent benefits</u> – compute average score of items 10, 11, 14, 15, 16

Subscale 3: <u>Parent control</u> – compute average score of items 8, 9