****Clinical Sequence Evidence-Generating Research Consortium

CSER Parental Patient Measures – post-ROR Follow-up #2 (5 - 7 months post-RoR)

Proposed by: multiple CSER Working Groups

Version 1.4.1, Dated 8/10/2018

## Feelings about Genomic Testing Results (FACToR) – Parent

*Citation: Meng Li, et al. "The Feelings About genomiC Testing Results (FACToR) Questionnaire: Preliminary Development and Validation" (2017) Manuscript in submission*

The following questions ask about how you, as a parent, felt after receiving your child’s genetic test results. Please indicate how much you had each specific feeling in the past week by circling the one answer for each question: *not at all, a little, somewhat, a good deal*, or *a great deal*.

1. How upset did you feel about your child’s genetic test result?
2. How happy did you feel about your child’s genetic test result?
3. How anxious or nervous did you feel about your child’s genetic test result?
4. How relieved did you feel about your child’s genetic test result?
5. How sad did you feel about your child’s genetic test result?
6. How frustrated did you feel about recommendations for your child's care based on the genetic test?
7. How uncertain did you feel about what your child’s genetic test result means for your child?
8. How uncertain did you feel about what your child’s genetic test result means for other family members’ risk of disease?
9. How much did you feel that you understood clearly your child’s choices for care based on the genetic test result?
10. How concerned did you feel that your child’s genetic test result would affect his or her ability to get or keep health insurance?
11. How helpful was the information you received from your genetic test result in planning for your child’s future?
12. How concerned did you feel that your child’s genetic test result might make it hard for them to get or keep a job?
13. How guilty did you feel about your child’s genetic test result?
14. How much loss of control over your child’s life did you feel because of your child’s genetic test result?

## Perceptions of Uncertainties in Genomic Sequencing (PUGS) (PARENT)

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| *Citation: Biesecker BB, Woolford SW, Klein WMP, Brothers KB, Umstead KL, Lewis KL, Biesecker LG, Han PKJ. PUGS: A novel scale to assess perceptions of uncertainties in genome sequencing. Clin Genet. 2017 Aug;92(2):172–179. PMCID: PMC5462880*  *Parental Perceptions of Uncertainties in Genome Sequencing (PUGS)* | | | | | | | |
| Rate how certain you feel about the following aspects of your child’s sequence results: | | | | | | | |
|  | Very  Uncertain | |  | | | Very  Certain | |
| 1. What my child’s test results mean for his/her health | 1 | 2 | | 3 | 4 | | 5 |
| 2. What actions I need to take based on my child’s test results | 1 | 2 | | 3 | 4 | | 5 |
| 3. How my child’s doctor may use the results to improve my child’s health | 1 | 2 | | 3 | 4 | | 5 |
| 4. Whether I am worried or concerned about my child’s test results | 1 | 2 | | 3 | 4 | | 5 |
| 5. Whether my child’s test results revealed something alarming | 1 | 2 | | 3 | 4 | | 5 |
| 6. Whether my child’s test results disrupted my life | 1 | 2 | | 3 | 4 | | 5 |
| 7. Whether I can trust my child’s test results | 1 | 2 | | 3 | 4 | | 5 |
| 8. Whether my child’s test results are accurate | 1 | 2 | | 3 | 4 | | 5 |

## Patient Reported Utility (PrU)

*Citation: Kohler, Jennefer N., et al. "Defining personal utility in genomics: A Delphi study." Clinical genetics 92.3 (2017): 290-297*

PARENTAL PERSONAL UTILITY SCALE (PrU) —FINAL VERSION 17 items

*Set survey to randomize 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 |
| Help with my child’s life planning | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Inform plans for my child’s school or career | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Inform my child’s 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 our family mentally prepare for the future | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Help to better understand my child’s health | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Contribute to my child’s self-knowledge | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Help me cope with my child’s health risks | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Help me feel more in control of my child’s health | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Help me feel more in control of my child’s life | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Simply to provide information | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Satisfy my curiosity about my child | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Help my child 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 family members | 1 | 2 | 3 | 4 | 5 | 6 | 7 |
| Feel good about taking responsibility for my child’s health | 1 | 2 | 3 | 4 | 5 | 6 | 7 |

## Understanding (novel)

How well do you understand your child’s test results?

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Not at all   | A little bit   | Moderately   | Quite a bit   | Extremely   |

## Information seeking V2

*Citation: Read CY, Perry DJ, Duffy ME. Design and psychometric evaluation of the Psychological Adaptation to Genetic Information Scale. J Nurs Scholarsh. 2005;37(3):203–208. PMID: 16235859*

INSTRUCTIONS: Please administer at follow-up 2, 5-7 months post ROR. Please administer this survey to all patients/parents who received diagnostic or uncertain (VUS) findings with regards to the primary indication for testing, as well as to participants who received secondary findings.)

Information Sources for Follow-up 2

Which of the following sources, if any, did you use to find more information about the genetic test results you received at your last visit? Please rate the usefulness of the sources you used.

1=not useful at all, 5=very useful

Family or friends 1 2 3 4 5

Facebook 1 2 3 4 5

Support groups 1 2 3 4 5

My/my child’s other doctors 1 2 3 4 5

Internet Search, i.e. Google, Pub Med, etc. 1 2 3 4 5

Books and other print media 1 2 3 4 5

Information provided by the doctor who 1 2 3 4 5

ordered my/ my child’s genetic test

Other (please specify)\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ 1 2 3 4 5

None

If you used the internet to search for information about the results, please list any web sites you found helpful.

Please rate your level of agreement or disagreement with the following statements.

1=strongly disagree, 6=strongly agree

I understand how I and/or my child came to have this gene change.

1 2 3 4 5 6

I understand the health risks my relatives face because of this gene change.

1 2 3 4 5 6

I understand the chances I have of passing this gene change on to my children.

1 2 3 4 5 6

I feel that I can explain to other people what having this gene change means.

1 2 3 4 5 6

## Family communication

*Notes: Family Communication is to be administered for both positive and negative results. We feel that participants will still find it important to communicate to other family members that their genetic work-up was negative.*

1. Since receiving your/your child’s study results, have you shared the information with any biological family members (blood relatives)?

* Yes
* I didn’t share this information with anyone [PRG: if selected, skip to Q5]
* I haven’t shared this information yet, but plan to in the future [PRG: if selected, skip to Q3]
* I don’t have blood relatives to share this information with [PRG: if selected, skip all remaining questions]

1. Since receiving your/your child’s study results, have you shared the information with any of the following blood relatives?

|  |  |  |  |
| --- | --- | --- | --- |
|  | Yes | No | N/A |
| My child’s other biologic parent [PRG: only show for parents of pediatric patients] | ⭘ | ⭘ | ⭘ |
| My child(ren) | ⭘ | ⭘ | ⭘ |
| My siblings | ⭘ | ⭘ | ⭘ |
| My parents | ⭘ | ⭘ | ⭘ |
| My other biological family members  [If select “Yes”:] Please specify who: [PRG: FREE TEXT] | ⭘ | ⭘ | ⭘ |

1. On a scale of 1 to 5, how important were each of the following reasons for sharing your/your child’s genetic test results with blood relatives?

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Not at all important (1) | 2 | 3 | 4 | Very Important (5) |
| To give my blood relatives information about their genetic risk | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| To encourage my blood relatives to have genetic testing | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| The doctor/genetic counselor encouraged me to share the information with blood relatives | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| So my relatives could make family planning decisions | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| To share the information I learned because I thought it was interesting | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| To share my feelings about my genetic test results | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| So I could get help from blood relatives with coordinating and planning for things like appointments and other health-related responsibilities (for example, going to doctors’ appointments, getting child care, getting transportation, etc.) | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |

1. Are there any other reasons that influenced your decision to share the results with blood relatives? [PRG: FREE TEXT]
2. On a scale of 1 to 5, how important were each of the following reasons for not sharing your/your child’s genetic test results with blood relatives?

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Not at all important (1) | 2 | 3 | 4 | Very Important (5) |
| I don’t want to worry or upset them | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I would have to talk to a blood relative I’m not close to/prefer not to talk to | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I don’t have contact information for my blood relatives | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I have privacy concerns about sharing this information with my relatives | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I don’t know how to explain the genetic results to my relatives | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I don’t think this information is useful for my relatives | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I’m having trouble coping with my/my child’s results | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I’m overwhelmed with my/my child’s health | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |
| I’m worried that my relatives will treat me/my child differently | ⭘ | ⭘ | ⭘ | ⭘ | ⭘ |

1. Are there any other reasons that influenced your decision not to share the results with blood relatives? [PRG: FREE TEXT]
2. What type of information did you share with blood relatives? Please check all that apply.

* General information about my study results
* Detailed information about the genes they tested
* My relative’s risk of having a condition
* Information about the possibility of being treated unfairly based on the study results
* Recommendations of ways to prevent illness
* Recommendations for more screening and testing
* Feelings about my study results
* Other, please specify: [PRG: FREE TEXT]

1. How did you share information about your genetic test results with your blood relatives? Please check all that apply.

* In person
* By phone
* By letter
* By email
* Through social media
* Other, please specify: [PRG: FREE TEX]

## Recommended Medical Actions and Follow Through on Recommendations Attributable to Genomic Testing - For patients with a positive primary or secondary finding and an equal number of negatives

Parental Patient survey: Administer 5-7 months post-ROR

*For patients with a positive finding:*

*You will now be asked about what you did after you received your child’s genetic test results, including whether you shared the results with other health care providers.*

*For patients with a negative finding:*

*Your child’s genetic test results from participating in this study were negative. However, we would still like to know whether you talked to doctors or other healthcare providers about your child’s test result and anything you did after you received their negative result.*

1. Did you discuss your genetic test results with your/your child’s doctors or health care providers?
2. Yes
3. Not yet but I plan to
4. No and I don’t plan to

1a. If yes, please indicate which doctors or health care providers you have shared the results with.

1. Primary care provider/pediatrician
2. Oncologist
3. Cardiologist
4. Neurologist
5. Other specialist(s) \_\_\_\_\_\_\_\_\_

1b. If “no and I don’t plan to,” why not?\_\_\_\_\_\_\_\_\_\_\_

1c. If yes, did the doctor or health care provider make any recommendations based on the test result?

1. Yes
2. No
3. I don’t know/don’t remember

1c1. If yes, what were the recommendations?

1. Medication
   1. Start
   2. Stop
   3. Change (*e.g*., stop taking one medication and start another one or increase or decrease the dose or frequency)
2. Additional non-genomic medical tests for screening, monitoring, or diagnosis (*e.g*., blood test, imaging such as x-ray, MRI, etc)
   1. Start
   2. Stop
   3. Change (*e.g*., increase or decrease the frequency)
3. Referrals to consult with other doctors or specialist
   1. Yes
      1. If yes, please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
   2. No
   3. Stop seeing other doctors or specialists (Please specify:\_\_\_\_\_\_\_\_\_\_)
4. Referral to a non-MD health professional
   1. New consultation with one or more of the following (please check all that applies):
      1. Audiology
      2. Dental
      3. Genetic counselor
      4. Psychologist
      5. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   2. Stop seeing other non-MD health professional
5. Referral for mental health support
   1. Mental health
   2. Social support
   3. Palliative care
6. Referral for therapeutic services
   1. Speech therapy
   2. Occupational therapy
   3. Physical therapy

Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)

1. Lifestyle changes
   1. Change diet
   2. Change exercise
   3. Start taking vitamins and supplements
   4. Change alcohol consumption
   5. Stop smoking
   6. Other (Please specify: \_\_\_\_\_\_\_\_\_\_)

1c2. Have you followed the recommendations?

1. Yes

2. No but I plan to

3. No and I do not plan to

* + - * + 1c2a. If yes, which ones?\_\_\_\_\_\_\_\_\_\_\_\_

[PRG: Drop-down menu for Medication, Medical, Other, Lifestyle, with associated response line(s) as indicated below each response category]

**Medication**

Please specify which medication(s): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Medical**

New consultation with a medical specialist

* 1. Please specify which specialty(ies): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

New consultation with a non-MD health professional

* 1. Please specify which non-MD health professional(s): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

New consultation for therapeutic service

* 1. Please specify which therapeutic service(s): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Any additional laboratory testing?

* 1. Please specify which type of lab test(s):

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

An imaging test (such as x-ray, MRI, etc.)

* 1. Please specify which type of imaging test(s):

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

* 1. If yes, what is the frequency?

One time only

Recurring

**Other**

**Lifestyle**

1c2b. If “No and I do not plan to”, Why not? \_\_\_\_\_\_\_\_\_\_\_

1. Since the genetic diagnosis was made, have you received counseling from your ob/gyn, reproductive genetic counselor, or primary care provider to discuss how your/your child’s diagnosis might affect future pregnancies?
2. Yes
3. Not yet but I plan to
4. No, and I don’t plan to
5. Not applicable

2a. If “No and I do not plan to”, Why not? \_\_\_\_\_\_\_\_\_\_\_

## Patient-Initiated Actions Attributable to Genomic Testing FOR patients with a positive primary or secondary finding and an equal number of negatives

Patient-Initiated Actions Attributable to Genetic Testing

1. Have you made any changes in your/your child’s health care or lifestyle, not based on medical recommendations made by your doctor or health care provider?
   1. Yes
   2. No

1a. If yes, what kind of changes did you make on your own?

* 1. Changed diet
  2. Changed exercise
  3. Started taking vitamins and supplements
  4. Changed alcohol consumption
  5. Stopped smoking
  6. Stopped seeking diagnostic testing
  7. Stopped medication
     + - What medication did you stop on your own? \_\_\_\_\_\_\_\_\_\_\_
  8. Other \_\_\_\_\_\_\_\_\_\_\_\_

2. Did you change your/your child’s insurance based on the results of the genetic testing?

1. Yes
2. No

2a. If yes, what kind of change?

* + 1. Buying new or buying more life insurance
    2. Buying new or buying more disability insurance
    3. Buying new or buying more long term care insurance

1. Have you made any other changes to your lifestyle based on the results of the genetic testing?
2. Yes
3. No

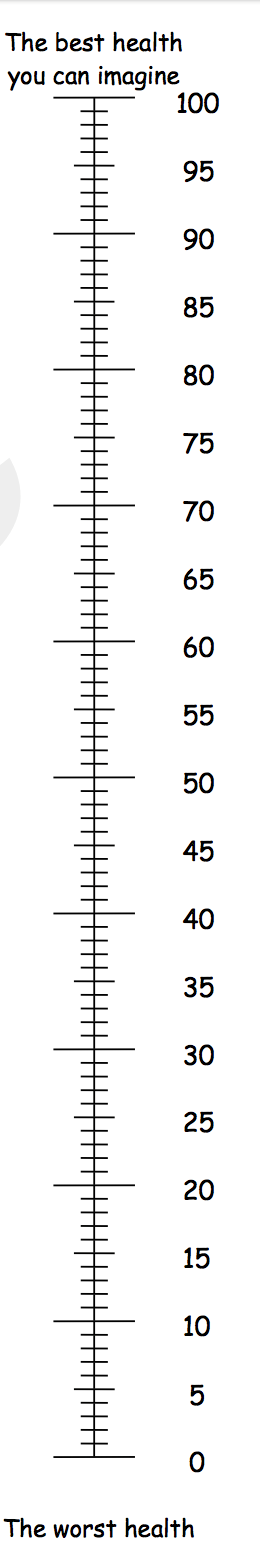
3a. If yes, what kind of change?

1. Changed job
2. Reduced time or quit job
3. Moved closer to hospital

## Quality of Life Ascertainment – Visual Analog Scale

FOR SUBSET OF PATIENTS WITH PRIMARY AND/OR SECONDARY FINDINGS

*See Scale on next page*

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* We would like know how good or bad your child’s health is TODAY.
* This line is numbered 0 to 100.
* 100 means the best health you can imagine.

0 means the worst health you can imagine.

* Mark an X on the scale to indicate how your child’s health is TODAY
* Now please write the number you marked on the scale in the box below

YOUR CHILD’S HEALTH TODAY=

How good is your child’s health TODAY

## Quality of Life Ascertainment - PedsQL

Due to survey licensure, we are unable to share copies of PedsQL surveys we are using, but will include the domain areas used in CSER (see below table).

More information about the PedsQL can be found on their website @ <http://www.pedsql.org/>. Some sample versions of these surveys can be found @ <http://www.pedsql.org/pedsql13.html>.

|  |  |  |
| --- | --- | --- |
| CSER Use Case | English | Spanish |
| Infant (1 - 12 months) | Parent Proxy: PedsQL™ Infant (1-12 months) | Parent Proxy: [SPANISH] PedsQL™ Infant Spanish version contains both Infant (1-12 months) and Infant (13 - 24 months) |
| Infant (13 - 24 months) | Parent Proxy: PedsQL™ Infant (13 - 24 months) |
| Generic Core Scale, Toddlers (age 2 - 4 years) | Parent Proxy: PedsQL Parent Report for Toddlers (ages 2-4) | Both Parent Proxy and Child Self Reports (if applicable): [SPANISH] PedsQL™ Generic Core Scale Please note this Spanish version contains all age groups |
| Generic Core Scale, Young Child (5 - 7 years) | Parent Proxy: PedsQL Parent Report for Young Child (ages 5-7) Child Self Report: PedsQL Young Child Report (ages 5-7) |
| Generic Core Scale, Child (8 - 12 years) | Parent Proxy: PedsQL Parent Report for Child (ages 8-12) Child Self Report: PedsQL Child Report (ages 8-12) |
| Generic Core Scale, Teenagers (13 - 18 years) | Parent Proxy: PedsQL Parent Report for Teen (ages 13-18) Teen Self Report: PedsQL Teen Report (ages 13-18) |
| Cancer Module, Young Child (5 - 7 years) | Parent Proxy: PedsQL Cancer Parent Report for Young Child (ages 5-7) | Parent Proxy: [SPANISH] PedsQL™ Cancer Module Please note this Spanish version contains all age groups |
| Cancer Module, Child (8 - 12 years) | Parent Proxy: PedsQL Cancer Parent Report for Child (ages 8-12) |
| Cancer Module, Teen (13 - 18 years) | Parent Proxy: PedsQL English Cancer Parent Report for Teens (13-18) |