Clinical Sequence Evidence-Generating Research Consortium

**CSER Provider Measures – post-RoR Follow-up #1 (0-6 weeks post-RoR)**

Proposed by: multiple CSER Working Groups

Version 1.1, Dated 4/26/2018

# Survey Measures and Outcomes WG

***NOTE: For ALL CSER SMO WG Provider Measures***

***Audience:*** *Some subset (some fixed N (TBD) per result type (positive, negative, VUS) per site, e.g., N=50. don't anticipate that all providers will respond. don't anticipate that all providers will need to be asked)*

***Target:*** *the provider who will be managing the downstream patient care/using the results, not necessarily the same provider as the disclosing provider. Could be primary or specialty care.*

## [NECESSARY] Healthcare Provider Confidence

***Frequency:*** *per patient, could have a positive, negative or VUS primary result. could ask a provider to respond more than once for different patients.*

Provider Confidence

Before consulting resources other than those provided with the result, when you received the genomic result, how confident did you feel in your…….

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | Not at All Confident | A Little Confident | Somewhat Confident | Very Confident |
| … knowledge about the patient’s medical condition? |  |  |  |  |
| … ability to explain the result to your patient? |  |  |  |  |
| … ability to answer your patient’s questions about the result? |  |  |  |  |
| … ability to manage the patient’s care based on the result? |  |  |  |  |

## [NECESSARY] Healthcare Provider Perceived Utility

***Frequency:*** *per patient, could have a positive, negative or VUS primary result. could ask a provider to respond more than once for different patients.*

Regarding this patient’s genomic result, how much do you agree or disagree with the following statements?

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Strongly disagree | Somewhat disagree | Somewhat agree | Strongly agree | Don’t know |
| The genomic result is important to this patient's care. |  |  |  |  |  |
| The genomic result might lead to patient harm through unnecessary follow-up. |  |  |  |  |  |
| Using the genomic result was a burden to me in terms of time or additional work. |  |  |  |  |  |

How useful did you find the genomic result for managing this patient’s care?

* Not at all useful
* A little useful
* Somewhat useful
* Very useful

In what ways did you find the genomic result useful? Please check all that apply.

* + Changed current management of patient’s care
  + Supported the management of patient’s care already underway
  + Directed need for future disease surveillance or screening
  + Informed disease prevention
  + Identified at-risk family members

## [NECESSARY] Healthcare Provider Specialty

***Frequency:*** *only measured once per provider.*

What is your primary role as a health care provider?

* Physician (MD/DO)
* Nurse practitioner (APRN/MSN/DNP)
* Physician’s Assistant (PA-C)*.*
* Genetic counselor
* Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

What is your specialty? (select all that apply)

* General Internal Medicine
* Internal Medicine Subspecialty \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
* General Pediatrics
* Pediatrics Subspecialty \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
* Family practice
* General Med/Peds
* Neurology
* Med/Peds Subspecialty *\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_*
* Ob/Gyn
* General Surgery
* Surgery Subspecialty *\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_*
* Psychiatry
* Hem/Onc
* Medical Genetics
* Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

## [NECESSARY] Healthcare Provider Experience

***Frequency:*** *only measured once per provider.*

Have you received any of the following formal genetics education (not including college or professional degree)? Please check all that apply.

* Genetics residency or fellowship
* Genetics education course (online or in person)
* Residency rotation in genetics
* Graduate degree (in addition to your professional degree) focused on genetics
* Other
* I have had no formal genetics training.

How many years have you been in practice (since residency/training ended)?

* 0-5 years
* 6–10 years
* 11–15 years
* 16–20 years
* 21–25 years
* >25 years

# ELSI & Diversity WG

## [NECESSARY] Health Provider Self-Identified Race/Ethnicity

Please pick the category that best describes your race/ethnicity:

* American Indian or Alaska Native
* Asian
* Black or African American
* Hispanic or Latino
* Native Hawaiian or Other Pacific Islander
* White

# Clinical Utility, Health Economics, and Policy WG

## [NECESSARY] Recommended Actions Attributable to Genomic Testing

**PROVIDER for patients with primary and or secondary finding(s) –equivalent number of negatives:**

***Administer items soliciting data on recommended actions should be asked at the ROR FU#1 visit (0-6 weeks after disclosure) all positives (primary and secondary) and potentially an equal number of negatives.***

**This initial set is about provider opinions and not actions taken.**

1. Based on the information you have now (post study genetic test results), how confident are you that you have identified the primary causal etiology of the child’s condition? (You can check any box along the scale)  
[answers from “not at all” 0 to 6 “completely”] - primary only   
  
2. What do you think is the chance that the child (or patient, for adults) is a genetic condition? (You can check any box along the scale)  
[answers from “definitely not genetic” 0 to 6 “definitely genetic”

3. Were you able to articulate a clear next step to establish a diagnosis based on the genetic test results? (*Check one*)

[answers “no” and “yes”]

4. Were you able to give the patient a clear recommendations for management of symptoms based on genetic test results? (*Check one*)

[answers “no” and “yes”]

**The next set of questions are about actions recommended *after receiving the results of genetic testing associated with participation in the research study*.**

Which of the following did you order or recommend for the participant related to Primary Finding and/or Secondary Finding? [Note: The detailed list for each question will only be presented if the higher level question is answered Yes]. (*Check all that apply*)

1. A cytogenetic test Y/N
   1. Karyotype only
   2. Karyotype and microarray
   3. Microarray only
   4. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
2. An additional molecular genetic test Y/N
   1. Single gene test or small panel test (<10 genes)
   2. Large panel test (>10 genes)
   3. Exome sequencing
   4. Mitochondrial DNA testing
   5. Other (Please specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
3. ***Other types of laboratory testing (select all that apply)  Y/N***
   1. A metabolic lab test Y/N
      1. Plasma amino acids
      2. Urine organic acids
      3. Acylcarnitine panel
      4. Lactate
      5. Ammonia
      6. Metabolomic panel
      7. Transferrin isoelectric focusing
      8. Guanidinoacetate
      9. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   2. ***Endocrine***
      1. *Thyroid*
      2. *Diabetes related*
      3. *Adrenal axis*
      4. *Other (Please specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)*
   3. ***Lipids***
      1. *Cholesterol panel*
      2. *Other (Please specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)*
   4. ***Chromosome stability***
      * 1. *DEB breakage*
        2. *Telomere length*
        3. *Radiation sensitivity*
        4. *Other (Please specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)*
   5. ***Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)***
4. An imaging test Y/N
   1. MRI with contrast (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   2. MRI without contrast (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   3. CT with contrast (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   4. CT without contrast (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   5. Heart ultrasound/echocardiography (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   6. Ultrasound of other body parts (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   7. Plain films (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)

**Will anesthesia be required for any of the above?** [y/n]

If 4 is selected as yes. Was the imaging test ordered:

One time only

Recurring

1. A procedure to obtain a tissue sample for additional testing
   1. Muscle biopsy
   2. Lumbar puncture
   3. Skin biopsy
   4. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)

**Will anesthesia be required for any of the above?** [y/n]

1. Prophylactic Surgery to reduce disease risk? Y/N
   1. If yes – (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
2. Non-invasive electrophysiology Y/N
   1. EKG
   2. EEG
   3. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
3. Invasive electrophysiology Y/N
   1. EMG
   2. Nerve conduction
   3. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
4. Referral to another medical specialty for evaluation or management Y/N
   1. Cardiology
   2. Neurology
   3. Genetics and Metabolism
   4. Ophthalmology
   5. Nephrology
   6. Dermatology
   7. Pulmonology
   8. Immunology/Allergy
   9. Rheumatology
   10. Hematology
   11. Oncology
   12. Psychiatry
   13. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
5. Referral to a non-MD health professional
   1. Audiology
   2. Dental
   3. Genetic counselor
   4. Psychologist
   5. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
6. Referral for mental health support
   1. mental health
   2. social support
   3. palliative care
   4. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
7. Referral for therapeutic services
   1. Speech therapy
   2. Occupational therapy
   3. Physical therapy
   4. Other (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
8. Other changes to management
   1. Recommended a new medication (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   2. Recommended a change of dose of an existing medication (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   3. Recommended discontinuation of an existing medication (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   4. Changes to over the counter (OTC) medicines or supplements (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   5. Medical/metabolic diet (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   6. General dietary recommendations (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   7. Recommended change in exercise or level of activity (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   8. Other types of lifestyle changes (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)
   9. Any other recommendations not covered above (Please specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)