**CSER HARMONIZED MEASURE REPOSITORY: Follow Through on Medical Actions Attributable to Genomic Testing**

| **Template topic** | **Definition/Note** |
| --- | --- |
| **Measure name and acronym** | Follow Through on Medical Actions Attributable to Genomic Testing |
| **Source citation for original measure** | Novel (developed through iterative process of CUHEP working group leadership) |
| **Category of assessment (construct/ CSER framework location)** | Clinical utility/Patient-centered utility |
| **Description of measure** | Asks whether parents followed up on actions recommended by the physician after receipt of genetic testing results. This is a survey rather than a psychometric measure and thus not appropriate for psychometric validation. However, some of these items may be useful as outcome variables to use in predictive validity analyses. |
| **Operational definition of construct** | NA |
| **Summary of changes made to measure for CSER (“CSER-adapted scale”), if any** | NA |
| **Time to administer** | estimated 00:01:30 plus free text |
| **Target Respondent(s)** | Adult patient and parent of pediatric patient versions |
| **Age range(s) or respondents** | NA |
| **Number of items** | Original scale: NA |
| CSER adapted |
| **Subscales and items per subscale** | Original scale: NA |
| CSER adapted |
| **Response scale (including anchor labels)** | Original scale: NA |
| CSER adapted |
| **Scoring instructions** | Original Scale: NA |
| CSER adapted |
| **Validated cutoff scores, if any** | NA |
| **Norms (if available)** | Original scale: NA |
| **Contact for permission to use/adapt (associated cost)** | NA |
|  |  |
| **Validated administration modes** | Examples: Paper and pencil, computer adaptive test, interview |
| **Original measure languages available** | NA |
| **Evidence for reliability (provide type and values)** | NA |
| **Evidence for validity (provide type and values if available)** | NA |
| **Evidence for sensitivity to change** | NA |
| **Relevant references in genetics or genomics** | NA |

**Paste original scale below**

**Paste CSER adaptation below**

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 to change your current care 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?

Please check one of the following:

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**

1. 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? \_\_\_\_\_\_\_\_\_\_\_

**Paste or list CSER site-specific adaptation/deviation below**

**ClinSeq:** “Alternative” version according to harmonized measures Google sheet, but surveys not available

**SouthSeq:**

Recommended Medical Actions and Follow Through on Recommendations Attributable to Genomic Testing- *(Necessary Time point: (4 months+ ROR))*

1. Did you discuss your child’s genetic test results with your child’s doctor(s) or health care provider(s)?

* Yes
* Not yet, but I plan to
* No and I don’t plan to (**Skip to Question 23**)

1. **If you answered yes to question 21**. With whom did you discuss your child’s results? Please indicate which doctor(s) or health care provider(s) you shared results with.

* Primary care doctor (pediatrician, family doctor) or nurse practitioner
* NICU doctor or nurse practitioner
* Neurologist
* Another specialist \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

1. If you answered “no and I don’t plan to”, why not? \_\_\_\_\_\_\_\_\_\_\_\_\_\_
2. **If you answered yes to question 21**. Did your child’s doctor(s) or health care provider(s) make any recommendations based on the test result?

* Yes
* No
* I don’t know / don’t remember

1. **If you answered yes to question 24.** If yes, what were the recommendations (check all that apply)?
   1. Start a medicine
   2. Stop a medicine
   3. Change a medicine (change a dose of medicine)
   4. Start therapy (physical therapy, speech therapy, etc.)
   5. Stop therapy (physical therapy, speech therapy, etc.)
   6. Make changes to my child’s therapy (physical therapy, speech therapy, etc.)
   7. Order a medical test (x-ray, CAT scan, heart test, etc.)
   8. Cancel a medical test (x-ray, CAT scan, heart test, etc.)
   9. Refer my child to another healthcare provider
   10. Cancel a referral to a healthcare provider
   11. Change my child’s diet
   12. Start my child on vitamins or supplements
2. **If you checked i.** What kind of healthcare provider(s) did your child’s doctor or nurse practitioner refer him/her to? (Check all that apply)
   1. Cardiologist (heart doctor)
   2. A neurologist (brain doctor)
   3. A geneticist (genetics doctor)
   4. A nephrologist (kidney doctor)
   5. A dermatologist (skin doctor)
   6. A pulmonologist (lung doctor)
   7. An immunologist (immune system doctor)
   8. A hematologist (blood doctor)
   9. An oncologist (cancer doctor)
   10. An audiologist (hearing doctor)
   11. An optometrist or ophthalmologist (eye doctor)
   12. A dentist
   13. A social worker
   14. A hospice or palliative care program
   15. Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
3. Sometimes children qualify for clinical trials because they have received certain genetic test results. Was your child’s doctor able to find a clinical trial for your child?

* Yes
* No
* I don’t remember
* I don’t know

1. **If answered no to 27.** Sometimes families find out their child doesn’t qualify for a certain clinical trial because they have received certain genetic test results. Did this happen to your child because of his/her genetic test results?

* Yes
* No
* I don’t remember
* I don’t know

The last few questions have been asking you about what your child’s doctors or healthcare providers have recommended for your child. Now we want to know which of these things you’ve been able to do.

1. **The following options are only presented if they were selected in question 25.** Which of the following have you and your child been able to do so far as a result of your child’s genetic results? (check all that apply)
2. Start a medicine
3. Stop a medicine
4. Change a medicine
5. Start a kind of therapy (physical therapy, speech therapy, etc.)
6. Stop a kind of therapy (physical therapy, speech therapy, etc.)
7. Make changes to a therapy (physical therapy, speech therapy, etc.)
8. Get a medical test (x-ray, CAT scan, heart test, etc.)
9. Avoid getting a medical test (x-ray, CAT scan, heart test, etc.)

1. See another healthcare provider
2. Avoid having to see another healthcare provider
3. Change my child’s diet
4. Start my child on vitamins or another supplement
5. **If you checked g on Question 29.** What kind of medical test(s) has your child had as a result of the genetic test results? (Check all that apply)
   1. Another genetic test
   2. Another type of blood test
   3. An X-ray
   4. An MRI
   5. A CAT scan
   6. A heart ultrasound or ECHO
   7. An ultrasound of another part of the body
   8. A test that required a surgery (such as a spinal tap or biopsy)
   9. Another kind of surgery (getting a g-tube or trach)
   10. A test of brain electricity (EEG)
   11. A test of heart electricity (EKG)
   12. A test of muscle electricity (EMG)
   13. A test of nerve electricity (nerve conduction test)
   14. Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
6. **If you checked i on Question 25.** What kind of other healthcare provider(s) has your child seen as a result of his/her genetic test results? (Check all that apply)
   1. A cardiologist (heart doctor)
   2. A neurologist (brain doctor)
   3. A geneticist (genetics doctor)
   4. A nephrologist (kidney doctor)
   5. A dermatologist (skin doctor)
   6. A pulmonologist (lung doctor)
   7. An immunologist (immune system doctor)
   8. A hematologist (blood doctor)
   9. An oncologist (cancer doctor)
   10. An audiologist (hearing doctor)
   11. An optometrist or ophthalmologist (eye doctor)
   12. A dentist
   13. A social worker
   14. A hospice or palliative care program
   15. Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
7. **If at least one item was checked on 25, but no items were checked on 29.** It looks like your child’s doctor or nurse practitioner recommended some things, but you haven’t been able to do those things. Why is that?\_\_\_\_
8. **If at least one item was checked on 25, but no items were checked on 29.** Do you still plan on doing some of these things?

* Yes
* No

1. **If you responded “no” on 33.** You said you don’t plan to do the things your child’s doctor(s) or health care provider(s) proposed. Why is that?\_\_\_\_
2. Since you got your child’s genetic test results, have you been told by a doctor or other healthcare provider that your child’s diagnosis might affect future pregnancies in your family?
   * Yes
   * No
   * Not applicable (I wasn’t planning on having more kids)