

CSER SouthSeq Discussion Guide

Greeting

1. [Diagnosis Experiences]

➤ **First, I would like to talk with you about your experiences in trying to find the cause for NAME's medical condition.**

- When did you first become concerned about NAME?
 - What caused you to be concerned?
 - What did you do about your concerns?
- Can you tell me the story of how you and your family have searched for a cause of NAME's condition?
- What has been the *hardest* part so far in searching for the cause of his/her condition?
- What has been the *best* part so far in searching for the cause of his/her condition?

2. [Interest in and Reasons for a Diagnosis]

➤ **Next I would like to talk a little bit about your interest and reasoning in finding the cause for NAME's condition.**

- Think about how you felt right before you started this study.
Did you feel it was important to know the cause of NAME's condition?
 - Why was it important (or not important) to know?
 - What were the one or two most important reasons for wanting to know the cause of NAME's condition?
- Were there any reasons that made you want to *avoid* knowing the cause?
[Probe]
- Had your interest in knowing the cause changed since the time you first learned that NAME had this condition? [Probe]

3. [Expectations]

➤ **Let's talk now about your expectations regarding NAME's genetic testing in this study.**

- What were your thoughts when you first heard about this study?

Next I am going to ask you about your *expectations* for this study and then I will ask about your *hopes* for this study.

- What did you expect to learn from these tests?
 - In what way were your expectations met?
 - In what way were your expectations *not* met?
- What you *expected* to learn from these tests, was that different from what you had *hoped* to learn from the tests?
 - Can you tell me about that? [PROBE for differences between 'hoped for' and 'expected.']

4. [Parental Emotions re Child]

➤ For some parents, the search for a cause to their child's condition can be emotional.

Learned Prenatally:

If parents knew prenatally their child might have a medical problem,

Ask:

- **During your pregnancy**, how would you describe your emotional state in relation to his/her medical problem?
PROBE for emotional states:
 - Positive feelings/states, such as hope, acceptance
 - Negative feelings/states, such as despair, anxiety, denial
 - Conflicting simultaneous positive and negative feelings.
- Did NAME'S condition affect how you bonded with him/her?

Learned at Birth:

If parents learned at birth their child might have a medical problem,

If parents learned later their child might have a medical problem,

Ask:

- **When you learned NAME had a medical problem**, how would you describe your emotional state in relation to that problem?
PROBE for emotional states:
 - Positive feelings/states, such as hope, acceptance
 - Negative feelings/states, such as despair, anxiety, denial
 - Conflicting simultaneous positive and negative feelings.
- Did NAME'S condition affect how you bonded with him/her?

Learned Later:

- Some families have expressed feeling guilt regarding their child's condition.
Did you at any time experience a feeling of guilt?
 - **If yes**, can you tell me about those feelings?
- **While you waited** for the genetic test results for NAME, what were you feeling?
PROBE for emotional states:
 - Positive feelings /states such as hope, acceptance
 - Negative feelings such as despair, anxiety, denial
 - Conflicting simultaneous positive and negative feelings
- Tell me about your experience when you received NAME'S test results.
 - Who gave you the genetic results?
 - Where were you when they gave you the results?
- **When you got the test results** for NAME, what did you feel?

PROBE for emotional states:

- Positive feelings/states, such as hope, acceptance
- Negative feelings/states, such as despair, anxiety, denial
- Conflicting simultaneous positive and negative feelings.
- Did knowing the genetic results affect / change the way you bonded with NAME?
- Would you say the genetic results from this study for NAME:
 - Caused you emotional distress?
 - [If Yes] In what ways?
 - [If No] Why do you think you felt no distress?
 - Provided emotional relief?
 - [If Yes] In what ways?
 - [If No] Why do you think you feel no relief?
- What was good about the way you received NAME'S genetic results?
- What could have been better?
- Would you say you received the genetic results too early in NAME'S life, too late, or at about the right time?

5. [Parents' Results]

➤ **Let's set aside the genetic results for NAME's condition for a minute and talk about any results you may have gotten about yourself.**

- Did you get any genetic results about yourself or for OTHER PARENT?
 - [If yes] Can you tell me what you learned?
PROBE:
 - Emotions
 - Meaning to person, family, extended family
 - [If yes] Do you/OTHER PARENT have a medical problem that is explained by your/OTHER PARENT'S genetic result?
 - As far as you know, are there any medical conditions in your/OTHER PARENT'S family history that can be explained by your/OTHER PARENT'S genetic result?

6. [Personal Utility/Meaning]

➤ **Thinking about the genetic test for your child (and for yourself/OTHER PARENT), we want to learn if and how the results impacted your lives and the lives of other family members.**

- Taken as a whole, would you say that you were emotionally better before or after you learned the genetic test results from the study—or did you feel no emotional change? [Probe]
- Did you learn anything about your child's genes, your genes, or his/her OTHER PARENT'S genes that distressed you?
 - [If Yes], Can you tell me more about what you learned?
- Did you learn anything about your child's genes that were important or made a difference for him/her?
 - [If Yes], Probe:
 - Changes in medical care
 - Changes in therapy (speech, physical, etc)

- Have you done anything differently in your life since learning something about all your genes?
 - [If Yes] Probe.
- Did you learn anything about your child's gene, your genes, or OTHER PARENT'S genes that you think matters for other people in your family?
 - If yes, have you told them about this?
 - Do you think this was important or made a difference for you or for them?
 - [If Yes] Probe.

[If applicable]

- As far as you know, have any family members done anything differently since learning something about their genes from this study?
 - [If Yes] Probe.

7. [Reproductive Decisions]

➤ **The next topic I would like to discuss with you is about future pregnancies.**

- Did you learn anything from the genetic test in this study that has changed the way you think about having more children? [Explore]
 - [If Yes] How has your thinking about having more children changed?
 - [If No] Why do you think the test results did **not** change how you feel about having more children?
- In thinking about having more children, would you say that you are better off now than you were before you received the genetic test results?

8. [Results and Educational Materials]

➤ **The last topic I would like to discuss with you is about your experience with the written genetic test report you received from the study**

- Were you able to understand the report?
 - Yes: Are there specific things about the report that made it easy to understand?
 - No: Are there specific things about the report that made it difficult to understand?
- Is there any information you wished was on the report that was not there?
- Have you used any of the links and resources that were mentioned on the report?
- Have you shared the report with anyone?
 - Yes: Who have you shared the report with?
 - No: Are you planning to share the report with anyone?

We would also like to ask you about your experience with the Genome Gateway website.

- Did you have difficulty accessing or navigating the Genome Gateway site?
 - Yes: Probe for specific difficulties
 - Logging in/remembering website URL/password
 - Unable to read on particular type of device
 - Finding educational information
 - Finding questionnaires
 - Playing videos
 - Downloading files

- Messaging study staff
 - No: Is there anything specific that made using Genome Gateway easy?
- At what point(s) during your participation in the study did you use Genome Gateway?
 - Right after enrolled
 - While waiting on results
 - After receiving results
 - Combination of 1+ of above
- Thinking specifically about the educational information in Genome Gateway, did you find the material helpful?
 - Yes: What specific topics do you recall being most helpful
 - No: Are there specific topics that you recall being unhelpful or confusing?
 - No: Are there other topics you wish had been covered, but were not?

Did you use any other tools, websites, resources, or online patient support groups to learn more about your/your child's results?

- Yes: What resources did you use? Did you find them helpful?
- No: Probe for why
 - Wanted information that is not available
 - Satisfied with information already received
 - Overwhelmed and not looking for more information
 - Negative result, no reason to look anything up