Utility of Genomic Sequencing in Community Care Contexts, IRB# 21.0548
PARENT Discussion Guide, 4/22/2022

1. Greeting

Thank you so much for talking with me today. Our research study is focused on trying to improve care for children who have a genetic condition. We're focusing on the genetic diagnosis you received for your child through genetic testing. Our goal is to learn from you and your experiences to help families across the country. As a reminder, with your permission we will record this discussion. If you don't feel comfortable answering a question, that is totally OK, just let me know.

- 1a. Just to start, could you tell me a little bit about your family and child with a genetic condition?

Thank you for that.

2. Background

Initial Perceptions of Condition

- 2a. Prior to getting the genetic diagnosis, what did you understand about NAME'S condition?

Care Prior to Diagnosis

- 2b. Before getting a genetic diagnosis, was NAME receiving care / treatment / services from medical professionals, therapists, or school professionals in relation to their condition?

  Probe: If yes, which types of professionals was NAME receiving care / treatment / services from?

Reason for Seeking Diagnosis

- 2c. Could you please tell me a little bit about what led you to seek genetic testing?

  Probe: Did a medical, therapy, or school professional recommend that you look into genetic testing for NAME?

  Why did they feel genetic testing might be helpful?

  What did you feel / think when they suggested this?

  What did you feel / think about genetic testing?

- 2d. What were you expecting as a result of getting a diagnosis? What changes or impact were you expecting?

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Probe: NAME'S medical care, therapy, or other care or services?

Probe: Plan / Prepare

Probe: Reoccurrence risk for self, NAME, siblings, other family members?

Getting a Diagnosis

- 2e. Did getting a genetic diagnosis change what you understood about NAME'S condition?

  Probe: If yes, what changed?

- 2f. After giving you the genetic diagnosis, what were the next steps the genetics provider recommended

  Probe: Did you follow all recommendations, or only some? Why?

3. Sharing Diagnosis

Sharing with Community Professionals

- 3a. Did you share the genetic diagnosis with NAME's therapists or other professionals, or did they learn about it through other sources, e.g., through the EHR?

- 3b. After receiving NAME'S genetic diagnosis, can you tell me about any discussions you had with their therapists or other professionals in relation to NAME'S genetic diagnosis and their care with that professional?

Goals for Sharing with Community Professionals

- 3c. Why was it important for you to share NAME'S genetic diagnosis with those professionals?

- 3d. How did the therapists or other professional respond to learning NAME has a genetic diagnosis?

  Probe: What did they say?

- 3e. What were you expecting would happen as a result of that conversation?

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- 3f. As far as you know, did your child's care or therapy plan change because of their genetic diagnosis?

  Probe: Why do you think that change occurred?

  or

  Probe: Why do you think no change occurred?

- 3g. Does having a genetic diagnosis for NAME change the way you think about the goals of their therapy or other needed care?

- 3h. Because of receiving a genetic diagnosis, what changes in care or therapy do you think would be helpful for NAME?

- 3i. Did getting a genetic diagnosis for NAME lead to seeing new professionals? [Mention some of the professionals listed in the footer]

  Probe: If so, which professionals were new?

  Probe: How did getting a genetic diagnosis lead to NAME seeing new professionals?

- 3j. Did getting a genetic diagnosis for NAME lead you to no longer see some professionals?

  Probe: If so, which professionals is NAME no longer seeing?

  Probe: How did getting a genetic diagnosis lead to NAME no longer seeing those professionals?

4. Sharing with School Professionals

- 4a. When you received the genetic diagnosis, who at the school did you reach out to first?

  Follow-up: Describe then what happened next, what action was taken as a result?

  How did the school professional respond to learning NAME has a genetic diagnosis? And what did they say? Did anyone else become involved as a result?

  Probe: Did anything change about the provision of services as a result of the genetic diagnosis? Describe what changed or explain why it didn't change.

  Probe: Looking back now, is there anything you would change or do differently regarding sharing the genetic diagnosis with your child's school? Is there anything you wish they had done differently?

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Probe: What were you hoping or expecting would happen as a result of that conversation?

- 4b. Since learning about NAME'S genetic diagnosis, has anything changed about the school services NAME receives?

- 4c. Since learning about NAME'S genetic diagnosis, has anything changed about the school professional he/she sees?

  Probe: Why do you think that change occurred?

  or

  Probe: Why do you think no change occurred?

- 4d. Does having a genetic diagnosis for NAME change the way you think about the goals of their school services?

- 4e. Because of receiving a genetic diagnosis, what changes in school services do you think would be helpful for NAME?

- 4f. Did sharing NAME'S 's genetic diagnosis with the school lead to any changes? If so, what were they?

  Probe: Helpful in some way?

  Probe: Make things harder in some way?

5. Role of Other Parents

Connecting with Other Parents

- 5a. Since receiving NAME'S genetic diagnosis, have you talked, either in person or online, with other parents of children with the same condition?

- 5b. What have those conversations been like?

- 5c. Is it helpful to talk with other parents of children with the same condition?

  Probe: If so, in what ways is it helpful?

- 5d. Are there drawbacks to talking with other parents of children with the same condition?

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Probe: If so, what are those drawbacks?

- 5e. Have you received advice or ideas from other parents about what kinds of therapy might be good for your child? If so, what was that advice? Was it helpful? If so how?

- 5f. Have you received advice or ideas from other parents about issues related to school? For example, how teachers should work with your child while at school, services that your child might qualify for, etc.?

6. ***Other Domains of Utility*** (to provide context for Utility in Community Contexts)

Clinical Utility

- 6a. Did you learn anything about NAME'S genetic diagnosis that has changed the medical care that he/she has received?

  Probe: What changes have occurred?

- 6b. Did the genetic diagnosis change how often you are in contact with your child's medical team?

Personal/Family Utility

- 6c. Did you learn anything about NAME'S genetic diagnosis that has been important or made a difference for you, NAME, or your family?

  Probe: What changes have you seen?

  Probe: Have you shared this diagnosis with members of your family? If so, who have you shared it with?

- 6d. Have there been changes in how other people see or treat NAME as a result of their genetic diagnosis?

  Probe: Have there been changes in how NAME sees him/herself?

- 6e. Did getting a genetic diagnosis change NAME's prognosis?

- 6f. Did getting a genetic diagnosis change how you are planning or preparing for NAME'S future?

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Probe: Or your future?

- 6g. Did getting a genetic diagnosis impact how you think about future pregnancies:
  - for yourself,
  - for Name,
  - for Name's siblings,
  - for extended family members?

- 6h. Has the genetic diagnosis led you to change anything about your lifestyle/work/support system?

7. Conclusion

- 7a. Are there is anything else I didn't ask about that you'd like for me to know?
- 7b. Do you have any questions for me?

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