

1. Greeting

Thank you so much for talking with me today. This research study is focused on trying to improve care for children who have a genetic condition, so our goal is to learn from you and your experience 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.

2. Background

- 2a. To start, could you tell me a little bit about your work as a therapist? How long have you been a physical therapist? How long have you been working with children? What is your primary practice setting? What diagnoses in children do you typically see?
- 2b. How frequently do you care for children with genetic conditions?
- 2c. As a physical therapist, have you received training or education about genetic conditions and genetic testing? If so, what was that training?

Probe: When in your education or career did you build that knowledge? Was this education or training formal or 'on the job'? Was this training you sought out as part of receiving CEUs or training you received as part of your initial degree? What led you to seek this training?

3. Initial Perceptions of Condition and Involvement in Search for Diagnosis

- 3a. Were you seeing NAME prior to him/her getting a genetic diagnosis?
If so:
- 3b. Prior to getting the genetic diagnosis, why was NAME referred to you? What was his/her medical diagnosis and what was your treatment diagnosis?
- 3c. Did you suspect the condition may have a genetic cause?

Probe: If so, where did you take your thought that NAME might have a genetic condition? With whom did you discuss this? What happened as a result?

- 3d. In this case, do you think you played a role in the process of NAME getting a genetic diagnosis? For example, did you suggest a particular condition or recommend genetic testing?

4. Getting the Diagnosis

- 4a. How did you find out about NAME'S genetic diagnosis? Who provided the information?
- 4b. Did getting a genetic diagnosis change what you understood about NAME's presentation? If yes, what changed?
- 4c. After you learned about NAME's genetic diagnosis, what next steps did you take? Did you talk with other professionals to get ideas?
- 4d. Did you do research or seek additional training or resources for the genetic condition? If so, what were your sources? Which one was the most helpful?
- 4e. At the time that you learned about NAME's genetic diagnosis, what recommendations or education did you receive about this diagnosis, if any? What was the source of those recommendations?
- 4f. Did you find the results of the genetic testing relevant for your decision making and treatment planning as a physical therapist? Did your treatment plan change?

Probe: If so, how?

Probe: If not, why not?

Probe for changes to:

- Goals of treatment
 - Frequency and intensity of therapy
 - Prognosis
 - Treatment and assessment approach.
- 4g. How did the genetic diagnosis impact your view of the ICF (International Classification of Functioning, Disability and Health) model for this child?

5. Perceived Goals of Parent/Caregiver:

- 5a. How did the genetic diagnosis impact your relationship with NAME and his/her family, if at all?
- 5b. As far as you know, did the parent/caregiver's view of the role of PT in their child's treatment change as a result of the genetic diagnosis, and if so, how?
- 5c. As far as you know, did the parent/caregiver's participation or expectations of physical therapy change following the genetic diagnosis and if so, how?

Probe: Did it change their goals for therapy?

Probe: Did it change their expectations for what their child might be able to attain through therapy?

6. Other Services:

- 6a. As a physical therapist, considering the genetic diagnosis, did you recommend any additional services or programming for the child and family? If so, what did you recommend? How did the family respond to those recommendations?
- 6b. (If physical therapist works in the school context): How did you find out about the genetic diagnosis of the student you were working with? How did you utilize that information?

Probe: Follow-up: How was this information used by the IEP team?

Probe: Did the team require or request additional information and if so, what additional information was provided and by whom?

Probe: Did the genetic diagnosis change anything about the service provision? If so, how and if not, why not?

- 6c. (If physical therapist works in a school context) Who officially communicated the genetic diagnosis to your colleagues in the school system? What were you hoping or expecting would happen as a result?
- 6d. (If physical therapist works in a school context) How did the genetic diagnosis impact the provision of services that the child received in school, if at all?
- 6e. (If physical therapist works in early intervention) How did the genetic diagnosis impact the provision of services that the child received in early intervention, if at all?

Follow-up: How was this information utilized by the team? Did it change the child's IFSP (Individualized Family Service Plan)?

Probe: Did the team require or request additional information and if so what additional information was provided and by whom?

Probe: Did the genetic diagnosis change anything about the service provision? If so, how and if not, why not?

- 6f. (If physical therapist works in outpatient therapy) How did the genetic diagnosis impact the provision of services that the child received in outpatient therapy, if at all?

- 6g. As far as you know, did this impact insurance coverage for services for NAME?
- 6h. As far as you know, did this impact NAME'S ability to access federally funded services, like early intervention, social security, etc.?

7. Utility and disutility in the classroom:

- 7a. (If physical therapist works in a school context) Did knowing the genetic diagnosis change anything about the way you interacted with the child in the classroom? Whether you decided to 'pull out' or 'push in' for PT services in the classroom?
- 7b. (If physical therapist works in a school context) As far as you know did the genetic diagnosis change the service plan for the child in the classroom, if so how? (i.e. require changes, reevaluation for 504 or IEP?)
- 7c. (If physical therapist works in a school context) As far as you know, did the genetic diagnosis impact the provision of:
 - Supplemental aids and services?
 - Supportive services (ie PT, OT, SLP)?
 - Educational environment?
- 7d. (If physical therapist works in a school context) As far as you know, did the genetic diagnosis have an impact on the child's academic achievement, classroom access or engagement? If so, how?

8. Role of Experience with Other Children

- 8a. How comfortable do you feel with being able to recognize a possible genetic cause for a child's presentation. How do you decide whether to communicate that insight to the family?
- 8b. As a physical therapist, what signs or symptoms or presentation typically signal to you a possible genetic component to the presentation?
- 8c. How has experience with genetic diagnoses impacted your practice as a physical therapist?
- 8d. Has your experience with similar genetic diagnoses led to any changes in treatment approach? If so, how do you think these changes occurred?

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- 8e. Has your experience in working with children with genetic diagnoses led you to explore additional educational opportunities or other collaborative opportunities? If so, how and what additional training did you pursue?

9. Conclusion

- 9a. Are there is anything else I didn't ask about that you'd like for me to know?
- 9b. Do you have any questions for me?

10. Additional Possible Questions:

- 10a. As far as you know, did the child's treatment team change as a result of the genetic diagnosis? Were there new people who helped take care of NAME, or were there people who were no longer involved with taking care of NAME?
- 10b. If there were changes, how did this impact your role in the child's treatment team?