

## **Supplemental Material S2. Interview guide.**

*The following questions were used to guide the semi-structured interviews as part of this study. Questions have been framed based on domains of the Theoretical Domains Framework (Cane et al., 2012; Michie et al., 2005), and a clinical vignette has been included to facilitate exploration of clinical reasoning in a specific clinical context (Jackson et al., 2015; Rousseau et al., 2020).*

### **Consent**

Participants were sent the Participant Information Sheet for the study prior to the interview. The project will be discussed, and if the participant agrees, verbal consent will be obtained.

**Verbal consent:** Before we begin this interview, can I confirm that you have read the information statement "Participant Information Sheet - Supporting speech-language pathologists in the genomic era: A Theoretical Domains Framework approach"?

Do you have any questions about the interview?

I am now going to switch on the audio and video recorder to record your consent. Is that okay?

*The audio and video recorder (hosted through Zoom (Zoom Video Communications, 2021)) will then be switched on with the participant's permission, and the participant will be asked the following:*

- Do you consent to participate in this project to explore speech-language pathologists' experience and views as they relate to genomic healthcare, as outlined in the Participant Information Sheet?
- Do you consent to the interview being audio and video recorded?

### **Questions**

**Opening:** Thank you so much for your time today. As you will be aware, genetics and genomics are becoming more integrated across the healthcare system. Speech-language pathologists play an important role in providing assessment and therapy for children with a range of diagnoses, including those with suspected and confirmed genetic conditions. Yet little research has been done studying speech-language pathologists' perceptions, beliefs, attitudes, and values as they relate to genomic healthcare, and how speech-language pathologists can be supported in this space.

The aim of these interviews is to learn from speech-language pathologists, as we realize speech-language pathologists have a key role in supporting communication in children with genetic conditions. I will be sharing a case study for us to discuss, and I'll be very keen to hear your thoughts.

I am not looking to test your knowledge about genetics and genomics, but am hoping to explore and understand your thoughts about the use of genetics and genomics in speech-language pathology practice and what relevance it might have. This will include exploring your current practice, and what might support you to incorporate the use of genetics and genomics in clinical practice. If you have any questions,

please save them until the end of the interview—I'm very happy to answer them then, or find someone who can assist.

## Interview structure

We're going to explore the patient journey, as it relates to children with suspected or diagnosed genetic conditions. This will be framed in three stages of the patient journey, and there will be a clinical example to think through at each stage (Fedorenko et al, 2016; Mei et al., 2018). The first stage is referred to as the *pre-referral stage*, which might occur when first meeting a family. Throughout this interview, we are going to focus on Luke, a 5-year-old boy who has been referred to your speech-language pathology service. His parents, Emma and Brian, are concerned about Luke's speech and language development.

- What would be your first steps when meeting a family such as Luke's for the first time?
- What would your first session look like?

Pre-referral stage	Domain
<p><i>I will now provide some more information about Luke.</i></p> <p><i>Luke is a 5-year-old boy who has been referred to your service. His parents, Emma and Brian, tell you that Luke's language seems to be behind other children his age, and he had delayed developmental milestones. Upon formal language assessment, you find he has expressive and receptive language impairment, across semantics, syntax, and morphology. Luke is also found to have speech sound impairment upon formal speech assessment, characterized by phonological disorder and childhood apraxia of speech.</i></p> <p><i>Emma and Brian are also concerned about Luke's learning, as his teachers have reported he has difficulty with reading and writing at school. Luke's hearing has been tested, and is within the normal range for speech and language development.</i></p>	
<p>Aside from formal and informal speech and language assessments, what other assessments, investigations or referrals would you consider for this child?</p> <p><i>If genetics is not raised:</i></p> <p>Do you usually consider the possible causes of speech impairment in your initial assessment? <b>(Do you usually consider genetic factors as a possible cause?)</b></p> <p><i>(Is it that genetics isn't something you think might be relevant for Luke, or not knowing the process that means you might not consider genetics?)</i></p> <p><b>What processes</b>, if any, do you have in place to determine if a child may be suitable for referral to genetics services? This could be a direct referral from yourself, or a referral via a pediatrician/as part of a developmental team. Have these been of help?</p> <p>If not, what processes could be helpful? <b>(How have these been of help? If not, what processes would help you?)</b></p>	<p>Memory, attention, and decision making; Behavioral regulation</p>

<p>If needed, who would you go to for support about Luke's kind of case?</p> <p>Has anyone in your professional support network suggested or discussed the consideration of genetics in your clinical practice? How have they done so? (<i>In what way? Or if not: Why do you think this might be the case?</i>)</p> <p>If you were to seek out further information about making a referral, which other healthcare professionals do you think you would go to?</p>	Social influences
<p>How would you feel about taking a family history for this family? (<i>What would the family history involve? Could you tell me a little more about why?</i>)</p> <p>If Luke's parents had questions about Luke having a genetic condition, how would you feel about discussing genetic concepts? For example, things like how a genetic condition occurs or whether speech and language impairments are linked to genetic conditions. (<i>Could you tell me a little more about why?</i>)</p>	Emotion; Beliefs about capabilities
<p>Do you feel genetic testing would be of value in Luke's case? (<i>If so, what value do you feel genetic testing may have?</i>)</p> <p>How do you feel genetic testing might affect Luke's outcomes in terms of his speech and language?</p> <p>Now thinking about the impact that considering genetic testing for Luke may have on you, how do you think this might impact your day-to-day practice?</p> <p>(<i>Might follow up to probe resources and skills?</i>)</p>	Beliefs about consequences
<p><b>Referral stage</b></p> <p><i>The next stage is the referral stage, in which a referral could be made to a clinical genetics service. A clinical genetics service is a service that provides assessment, information, testing and diagnosis for individuals with genetic conditions (diagnosed and suspected). It is often staffed by clinical geneticists and genetic counselors, who provide support and management options for these individuals.</i></p>	Domain
<p>How confident do you feel about identifying whether Luke could be referred to a clinical genetics service? (<i>What would help you in being more confident? OR What has helped you to be confident?</i>)</p>	Skills; Optimism
<p><i>This is the next part of Luke's case.</i></p> <p><i>Through engagement with an external supervisor, the supervisor has suggested that Luke's presentation would warrant further investigation through a clinical genetics service. Luke's parents are both open to this.</i></p>	
<p>Whose role do you think it would be to refer a child with speech and language concerns to genetics services?</p> <p>Where do you think speech-language pathologists might fit into this?</p>	Professional identity

<p>For Luke, if you were thinking about genetic testing, how comfortable are you in contacting a genetics service or pediatrician? (<b><i>What would help you to be more comfortable? OR What has helped you to be comfortable?</i></b>)</p> <p>For Luke, if you were thinking about genetic testing, how comfortable are you in making a referral to a genetics service or pediatrician (with the aim of them further investigating concerns around genetics)? (<b><i>What would help you to be more comfortable? OR What has helped you to be comfortable?</i></b>)</p> <p>What do you feel would be the hardest part of making a referral or engaging with a genetics service or pediatrician? (<b><i>What would help you with this part?</i></b>)</p>	<p>Emotion; Beliefs about capabilities</p>
<p>Thinking about your day-to-day practice, what impact would a referral make to your clinical practice, if any? (<b><i>might probe resources and skills etc.</i></b>)</p>	<p>Beliefs about consequences</p>
<p><b>Post-referral stage</b></p> <p><i>The final stage is termed the post-referral stage, and refers when a client has undergone genetic testing and what happens afterward. When a client has genetic testing, there are two main outcomes: 1) A genetic condition is identified, or 2) No genetic condition can be identified at this stage.</i></p>	<p><b>Domain</b></p>
<p><i>This is the final part of Luke's case.</i></p> <p><i>After Luke was referred to a clinical genetics service, the genetics healthcare professionals recommended that Luke have genetic testing. Two months passed, and Luke's family had another appointment with the genetics service to receive his testing results. Luke was found to have a genetic change called 16p11.2 deletion. This refers to there being a missing part of genetic information, or DNA, of chromosome 16.</i></p> <p><i>Luke's family continues to attend your service for speech pathology support. Emma and Brian feel concerned about Luke's diagnosis, and express their concerns to you—they are worried about his prognosis, and what his diagnosis means for their family.</i></p>	
<p>Whose role do you think it would be to discuss prognosis and impacts on the family?</p> <p>Where do you think speech-language pathologists might fit into this? (Or, how do you think speech-language pathologists would fit into this?)</p> <p>Who would you seek support from to address Emma and Brian's concerns?</p> <p>Where do you think genetic counselors might fit into this?</p>	<p>Professional identity</p>
<p>How do you feel about discussing the speech and language prognosis of a genetic condition such as Luke's? (<b><i>Why do you think this is? What is the hardest part of discussing his prognosis?</i></b>)</p> <p>How do you feel about providing counseling to Luke's family? (<b><i>Why do you think this is? What is the hardest part of discussing providing counseling?</i></b>)</p> <p>How would you feel if these aspects were to be incorporated into the role of a speech-language pathologist <b><i>in the future?</i></b></p>	<p>Beliefs about capabilities; Emotion</p>

What value do you perceive children such as Luke receiving a genetic diagnosis might have in terms of their speech and language therapy and outcomes? How do you think this might impact Luke's therapy outcomes?	Beliefs about consequences; Reinforcement
<i>If Luke had already received a diagnosis of 16p11.2 deletion and was then referred to your speech-language pathology service:</i> How would his diagnosis shape your therapy planning? ( <b><i>What would be helpful in planning his assessment and/or therapy?</i></b> ) Are there any specific resources, professionals, or tools that you would find helpful? <b><i>What are they?</i></b>	Beliefs about consequences; Environmental context and resources
<i>In a more general sense:</i> How do you feel about playing a role in the mainstreaming of genetics and genomics? ( <b><i>Why do you think this is?</i></b> ) Overall, what sort of role do you envisage as a speech-language pathologist?	Emotion; Professional identity

Are there any other relevant areas, that we haven't discussed, that you would like to share?

Thank you so much for participating in this interview. Please feel free to get in touch if you have any further insights to share.