



Translational Centre for Speech Disorders

Centre of Research Excellence



Speech and Language in individuals with Schinzel Giedion Syndrome (*SETBP1* gain of function)

An international study

We are running a project looking at speech and language outcomes in individuals aged 6 months and older with Schinzel Giedion Syndrome.

By improving our understanding of speech and language in this condition, we hope to improve prognoses, better identify those in need of support and develop more targeted strategies.

We are looking for individuals:

- Aged 6 months and older
- Confirmed to have Schinzel Giedion Syndrome by genetic test
- Who are verbal or non-verbal
- Who speak English, Dutch, French, German, Portuguese, Spanish or Italian

What is involved?

- Completing online surveys about speech and language skills, and health and medical history
- An online Zoom session with a Speech Pathologist for further assessment (1-2 hours)

About us

The Translational Centre of Research Excellence for Speech Disorders examines speech and language in individuals with rare genetic conditions.

We are trying to better understand the strengths and difficulties of communication in children with rare genetic syndromes. Our longer-term aim is to help develop better targeted therapies.

GET IN TOUCH!

If you or someone you know may be interested in helping with this research, we would love to hear from you. Contact us at:

geneticsofspeech@mcri.edu.au