



**Translational Centre  
for Speech Disorders**  
Centre of Research Excellence



# Speech and Language in individuals with *SYNGAP1*-DEE

## An international study

We are running a project looking at speech and language outcomes in individuals aged 6 months and older with *SYNGAP1*-developmental and epileptic encephalopathy.

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 speech therapy practices.

### We are looking for individuals:

- Aged 6 months and older
- Diagnosed with a pathogenic or likely pathogenic *SYNGAP1* variant by genetic test
- All communication abilities (verbal or minimally-verbal)

### What is involved?

- Completing online surveys about speech and language skills, health & medical history, quality of life, and epilepsy diagnosis
- A 1 hr appointment with a Speech Pathologist for further speech assessment (in-person or online)

### 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](mailto:geneticsofspeech@mcri.edu.au)

