



Translational Centre for Speech Disorders

Centre of Research Excellence



Speech and Language in individuals with PURA syndrome

An international study

We are running a project looking at speech and language outcomes in individuals aged 6 months and older with pathogenic or likely pathogenic variants in the *PURA* gene (also known as PURA syndrome).

By improving our understanding of speech and language profiles 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
- Who have a likely pathogenic or pathogenic variant in *PURA* confirmed by genetic test
- Who are verbal or non-verbal/non-speaking

What is involved?

- Completing surveys about speech and language skills, and health and medical history
- An optional in-person or online Zoom session with a Speech Pathologist for further assessment (~1 hour)
- Surveys are available in English, French, Dutch, German, Spanish, Portuguese, Italian, or Chinese

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