



Translational Centre for Speech Disorders

Centre of Research Excellence



Speech and language in individuals with CACNA1C-related disorders including Timothy syndrome and long QT type 8

An international study

We are running a project looking at speech and language abilities in individuals with variants in the *CACNA1C* gene. By improving our understanding of speech and language profiles in *CACNA1C*-related disorders, we hope to improve prognosis, better identify those in need of support and develop more targeted strategies.

We are looking for individuals:

- Aged 6 months and older
- Have a confirmed variant in *CACNA1C*
- Who are speaking or non-speaking

What is involved?

- Completing surveys about communication, 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, Chinese, or Polish

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.

Contact us

If you are interested in helping with this research or have questions email us at:

geneticsofspeech@mcri.edu.au

or scan the **QR code**

