





Speech Apraxia and Genetics Clinic

What does our clinic do?

Our clinic provides a speech diagnostic assessment for children with severe speech disorder where childhood apraxia of speech, or other diagnoses such as dysarthria, are suspected. This includes assessment to confirm or rule out childhood apraxia of speech. The clinic involves a one-hour speech assessment. We generate a report including the child's diagnosis and brief treatment recommendations. Our clinic does not provide treatment but works with the child's treating therapist on a treatment plan.

Our clinic also includes a one-hour paediatric genetic assessment. For children who are eligible for genetic testing, we begin with a chromosomal microarray which detects small extra or missing sections of chromosome material. If no changes are found, the next step is whole exome sequencing, which looks at hundreds of genes that may be associated with neurodevelopmental conditions including speech disorders.

Who do we see?

We see children over 3 years of age with a primary developmental concern of speech disorder. Children may have other mild co-occurring developmental features such as learning difficulties, gross or fine motor issues or attentional issues, but we do not see children who have moderate hearing loss. Children with a formal diagnosis of autism spectrum disorder may be seen on a case-by-case basis.

The Speech Apraxia and Genetics Clinic incorporates both research and clinical care and is funded by the Royal Children's Hospital Foundation. We are also supported by the Royal Children's Hospital Speech Pathology Department.

When: Friday afternoon, twice a month

Where: The Royal Children's Hospital, 50 Flemington Rd, Parkville VIC 3052 (or via telehealth)





Timing and outcomes

Our clinical reports are generated within 2-3 weeks of your original appointment. Chromosomal microarray analysis takes about two months, and if the child progresses to whole exome sequencing to look at single gene changes, results can take between three and four months.

We contact families when results become available from the chromosomal microarray or whole exome sequencing.

How to refer

A referral to the Speech Apraxia and Genetics Clinic can be obtained through a general practitioner. This referral letter needs to be addressed to Prof. Angela Morgan, Speech Apraxia and Genetics Clinic, Speech Pathology Department, The Royal Children's Hospital and mention the referral is for a speech assessment.

A child's treating speech pathologist may also provide a referral through the <u>Speech Pathology Second Opinion Referral Form</u>, available from The Royal Children's Hospital website. This must be accompanied by a recent speech pathology assessment report.

Surveys

This clinic exists for just a 2-year period, funded by the RCH Foundation. Medicare do not currently recognise the genetics of speech disorder. Our grant pays for the \$4500 cost per family for genetic (exome) testing. Without the grant, the clinic would not exist. We need to show the health and cost viability of the clinic. This is done via critical surveys that we will send you before genetic testing, and at two time points after genetic testing. These 3 surveys are critical to helping us show the importance of the clinic, by using results to lobby to Medicare at the end of our grant. This is important so that future children could also benefit from a clinic.

Genetic counselling

For families who receive a genetic diagnosis, Prof. David Amor will deliver the genetic result. We are able to arrange **genetic counselling** for families where required.

A common question for families is how will a genetic diagnosis change my child's care? Obtaining a genetic diagnosis will help provide the answer to "why" does a child have this speech presentation. Receiving a genetic diagnosis can also help provide information to support applications to the National Disability Insurance Scheme. A genetic diagnosis is unlikely to change your current speech therapy management because current best practice is still focused on managing the speech symptoms. Although the associated speech diagnostic assessment may help to better target your current therapy plan.

For more information

Please contact the Speech and Language team with any queries via email at: speechclinic@mcri.edu.au

Working in partnership with:



