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Newsletter

We are excited to share the latest news with you from our NHMRC Translational Centre for Speech Disorders!

Research studies

NEW STUDY: speech and language in individuals with SYNGAP1-DEE

Our Speech & Language team at the Murdoch Children's Research Institute (MCRI) has recently launched an international research study examining speech and language outcomes in individuals with SYNGAP1-developmental and epileptic encephalopathy.

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

👶 Eligibility:
🔍 Aged 6 months and older
🔍 Diagnosed with SYNGAP1-DEE by genetic test
🔍 Verbal or non-verbal/non-speaking

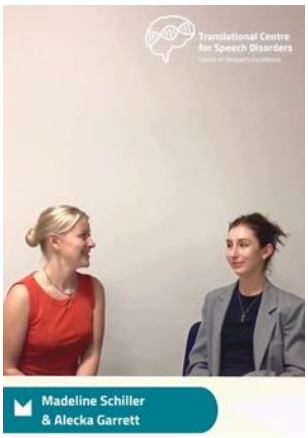
✅ What is involved:
✏️ Online or in-person meeting
✏️ Surveys about speech, language and health
✏️ Surveys are available in English, French, Dutch, German, Spanish, Portuguese, Italian, or Chinese

The study is being conducted by CRE PhD candidate Alecka Garrett under the supervision of CRE Director

Professor Angela Morgan and Chief Investigator Laureate Professor Ingrid Scheffer AO.

Interested families are encouraged to contact the study team at geneticsofspeech@mcri.edu.au or register their interest via this link.

This study is supported by a Syngap Research Fund Australia research grant. We sincerely thank Syngap Research Fund Australia for their wonderful support and collaboration.



Hear Alecka Garrett discuss this study

STUDY UPDATE: sensory & motor profile in childhood apraxia of speech (CAS)

CRE team member A/Professor Şermin Tükel Akay is conducting a study on the sensory and motor difficulties experienced by children with childhood apraxia of speech (CAS).

Many children with CAS have issues with fine and gross motor skills and sensory functions, which may interfere with academic performance and daily activities. By improving our understanding of the sensory and motor profiles of children with CAS, we hope to enhance

prognoses, identify those who need support, and develop more personalised treatment strategies.

A/Prof. Tükel Akay noted that since the study began in May 2024, she has had the privilege of meeting 55 children and their families who have been part of this research.

Participation is free and easy. The study involves some pleasant motor tasks and measurements for children conducted at the Murdoch Children's Research Institute, which usually takes about 1.5-2 hours to complete. Additionally, parents will be required to fill out some online surveys, which will take approximately 15-20 minutes to complete.

Interested families are welcome to contact the study team by registering their interest via email: speechclinic@mcri.edu.au.

We would like to express our sincere gratitude to the families who have already taken part in this study. Your involvement and support are greatly appreciated.

A/Prof. Akay in clinic assessing sensory-motor activities

Awards, Conferences & Events

RARE DISEASE DAY 2025

Tomorrow, Friday February 28, is International Rare Disease Day.

In honour of Rare Disease Day, we have been sharing informative posts this month highlighting key facts about

speech, language, and rare diseases. Please check out our social media channels Instagram or Facebook for more insights.

Additionally, CRE Director Prof. Angela Morgan was recently featured in a MCRI's 'Meet our Brilliant Minds' post, where she discussed the ongoing efforts to identify genetic causes of speech disorders in children. This important research aims to improve outcomes for those living with rare genetic diseases that impact their speech and language.

The Speech & Language team is showing their support for Rare Disease Day 2025.

Access 'Meet our Brilliant Minds' Instagram post

Selected recent publications

Parental attitudes and experiences in pursuing genetic testing for their child's motor speech disorder

Genetic counsellors in our CRE team - Ms. Christy Atkinson, Ms. Mariana Lauretta and Mr. Quan Lee, have published an article in the European Journal of Human Genetics: *Parental attitudes and experiences in pursuing genetic testing for their child's motor speech disorder.*

We conducted interviews with parents about their views and experiences in pursuing genetic testing for their child's motor speech disorder (Childhood Apraxia of Speech and/or Dysarthria). The genetic basis of these rare and typically severe speech disorders is well documented with approximately 30% of children who undergo genomic testing, receiving an explanatory genetic diagnosis.

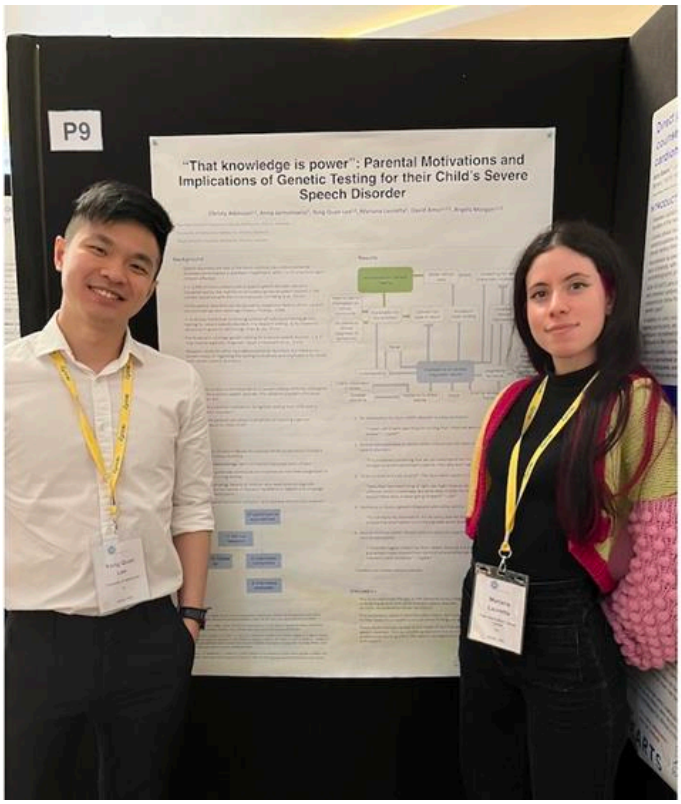
Our research shows that parents had largely positive attitudes towards genetic testing, regardless of their

child's testing outcome. We also found that having a multidisciplinary team (consisting of speech pathologists, clinical geneticists, genetic counsellors, and developmental paediatricians) is critical in supporting families throughout their genetic testing journey.

These findings are an important step to better understand the benefits of genetic testing for families affected by motor speech disorders across Australia.

At present, clinical genomic testing for children with motor speech disorders is not available via a government-funded clinical genetics service or medical practitioner. Our Speech Apraxia and Genetics Clinic which is run through the Royal Children's Hospital (Melbourne), has been the only pathway via research funding.

We would like to thank all the families and referrers who have supported our research clinic and helped us learn more about CAS.



Ms. Mariana Lauretta and Mr. Quan Lee presented some of the preliminary results at the Human Genetics Society of Australasia (HGSA) 47th Annual Scientific Meeting in August 2024.

Read more about this article

Adaptive Functioning in Children and Young People with Monogenic Neurodevelopmental Disorders

Dr. Emma Baker, an autism specialist in the Speech & Language team, is the lead author of a newly published article in Developmental Medicine & Child Neurology: *Adaptive Functioning in Children and Young People with Monogenic Neurodevelopmental Disorders*.

Emma's paper explores the adaptive behaviour profiles of children and young adults with one of 8 monogenic neurodevelopmental disorders (CDK13, DYRK1A, FOXP2, KAT6A, KANSL1, SETBP1, BRPF1, and DDX3X). Adaptive behaviours are the skills that are required for independent living and include communication, daily living, and social skills. Research shows that there is much variability in these skills within and across specific neurodevelopmental disorders.

The findings in this study highlight the need for transdiagnostic frameworks for intervention across these conditions to enable targeted person-centred approaches to maximise each individual's potential.


DMCN Developmental Medicine & Child Neurology

ORIGINAL ARTICLE | Open Access

Adaptive functioning in children and young adults with monogenic neurodevelopmental disorders

Emma K. Baker, Miya St John, Ruth Braden, Lottie D. Morison, Elana J. Forbes, Fatma Lelik, Stephen J. C. Hearps, David J. Amor, Angela T. Morgan

First published: 23 January 2025 | https://doi.org/10.1111/dmcn.16227

Access this publication here

Speech, language, and non-verbal communication in CLN2 and CLN3 Batten disease.

CRE PhD student and Speech Pathologist Lottie Morison, has published her second PhD paper in The Journal of Inherited Metabolic Disease: *Speech, language, and non-verbal communication in CLN2 and CLN3 Batten disease*. Batten disease is a group of genetic conditions which, collectively, are the most common cause of childhood dementia. CLN2 and CLN3 disease are the most common types of Batten disease.

The paper provides a novel contribution to understanding speech and language in CLN2 and CLN3 Batten disease. Speech and language disorders are often the first signs of Batten disease, with the nature and severity of these disorders changing across the course of the disease. This research helps us better understand the speech and language features of Batten disease and how clinicians can best support young people with Batten disease and their families. A Plain Language Summary and CLN2 and CLN3 Disease Fact Sheets are available on our website.

The team would like to sincerely thank the children, adolescents and young people with Batten disease and their families. This research was generously supported by the Batten Disease Support and Research Association (BDSRA) Australia's 2022 Research Grant. Thank you also to BDSRA United States, CLN2 disease Canada, and Batten Disease Family Association United Kingdom.

JIMD JOURNAL OF INHERITED METABOLIC DISEASE SSIEM

ORIGINAL ARTICLE | Open Access

Speech, Language and Non-verbal Communication in CLN2 and CLN3 Batten Disease

Lottie D. Morison, Ineka T. Whiteman, Adam P. Vogel, Lisa Tilbrook, Michael C. Fahey, Ruth Braden, Joanna Bredebusch, Michael S. Hildebrand, Ingrid E. Scheffer, Angela T. Morgan

First published: 16 January 2025 | https://doi.org/10.1002/jimd.12838

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Ingrid E. Scheffer and Angela T. Morgan are joint senior authors.

Read more about this article

Thank you!

Our team wishes to thank families and clinicians for their ongoing participation and support. Without your help our research would not be possible.

Centre of Research Excellence - Translational Centre for Speech Disorders

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