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murdoch children's research institute

Translational Centre for Speech Disorders
Centre of Research Excellence

Newsletter

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

Special Blue Edition for Apraxia Awareness Month

May 14th was Apraxia Awareness Day

Childhood Apraxia of Speech (CAS) is a motor speech condition that makes it difficult to speak. People with CAS know what they want to say but the brain cannot plan and program the muscles and movements of their lips, jaw, and tongue to speak clearly and be understood.

This month our Speech & Language team dressed all in blue to celebrate **Apraxia Awareness Day.**

Thank you to all the amazing families we work with! We would not be able to do our research without your time, drive and dedication 💙

Please check out our Instagram or Facebook pages for more insights

Second Opinion CAS Clinic

An end...

Thank you so much to all the families that have received CAS assessments in our research clinic. We have now finished our research clinic and are no longer able to provide second opinion assessments for CAS.

We'd like to thank the families and referrers who have supported our research clinic and helped us learn more about CAS. We thoroughly enjoyed the working relationship and collaboration and meeting so many wonderful families.

And a beginning...

Second Opinion CAS assessments are now being conducted at the Melbourne Speech Pathology Clinic (Level 1, 723 Swanston St, Carlton 3053). Please note this is a fee based service. They are now taking appointment bookings for May 2025 onwards.

THE UNIVERSITY OF MELBOURNE

Contact Information

Website: https://speech.mthc.com.au/

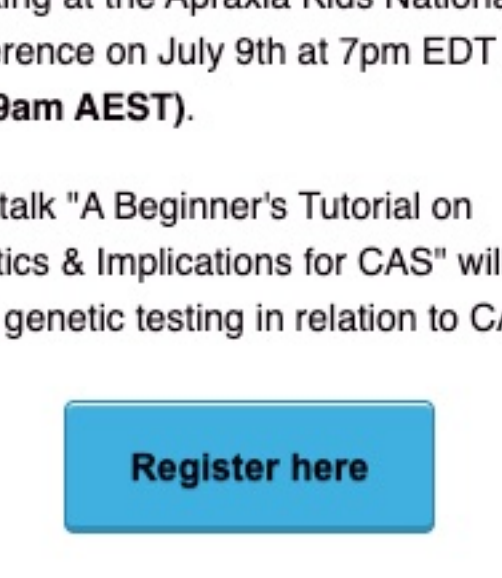
Email: speech-clinic@unimelb.edu.au

Phone: 03 9035 7199

Referral form: https://speech.mthc.com.au/contact/

Opening hours: Monday - Friday 8:30 am to 4.30 pm

Apraxia Kids Virtual National Conference (July 9-12)



2025 National Conference:
Creating Community Connections
Virtual
July 9-12th, 2025

Genetic counsellor Mariana Lauretta and Professor Angela Morgan will be speaking at the Apraxia Kids National Conference on July 9th at 7pm EDT **(July 10th 9am AEST)**.

Their talk "A Beginner's Tutorial on Genetics & Implications for CAS" will cover genetic testing in relation to CAS.

Register here

By the end of the session the participant will be able to:

- Explain the difference between a chromosomal condition and a single gene condition.
- List the different types of genetic testing relevant to a child with CAS.
- Describe 2 practical and emotional factors to consider as a parent or SLP if considering genetic testing for a child with CAS

More information about the conference can be found here

Parental attitudes and experiences in pursuing genetic testing for their child's motor speech disorder: Plain language Summary now available!

A couple months ago, genetic counsellors in our CRE team - Ms. Christy Atkinson, Ms. Mariana Lauretta and Mr. Quan Lee, published '*Parental attitudes and experiences in pursuing genetic testing for their child's motor speech disorder*'

We are now happy to share the plain language summary of the findings. See some excerpts below:

See the full version HERE

New Studies

Recruiting: Okur Chung neurodevelopmental syndrome

We are recruiting for a new study looking at speech and language in individuals with Okur-Chung neurodevelopmental syndrome (OCNDS) or also known as CSNK2A1-related neurodevelopmental syndrome in collaboration with CSNK2A1 Foundation

Eligibility:

🔍 Aged 6 months to adulthood

🔍 Diagnosed with a CSNK2A1 variant/Okur-Chung neurodevelopmental syndrome by genetic test

🔍 Verbal or non-verbal/non-speaking

✅ **What's Involved:**

✏️ Online or in-person meeting

✏️ Surveys about speech, language, and health

✏️ Surveys available in English, French, Dutch, German, Spanish, Portuguese, Italian, Polish, or Chinese

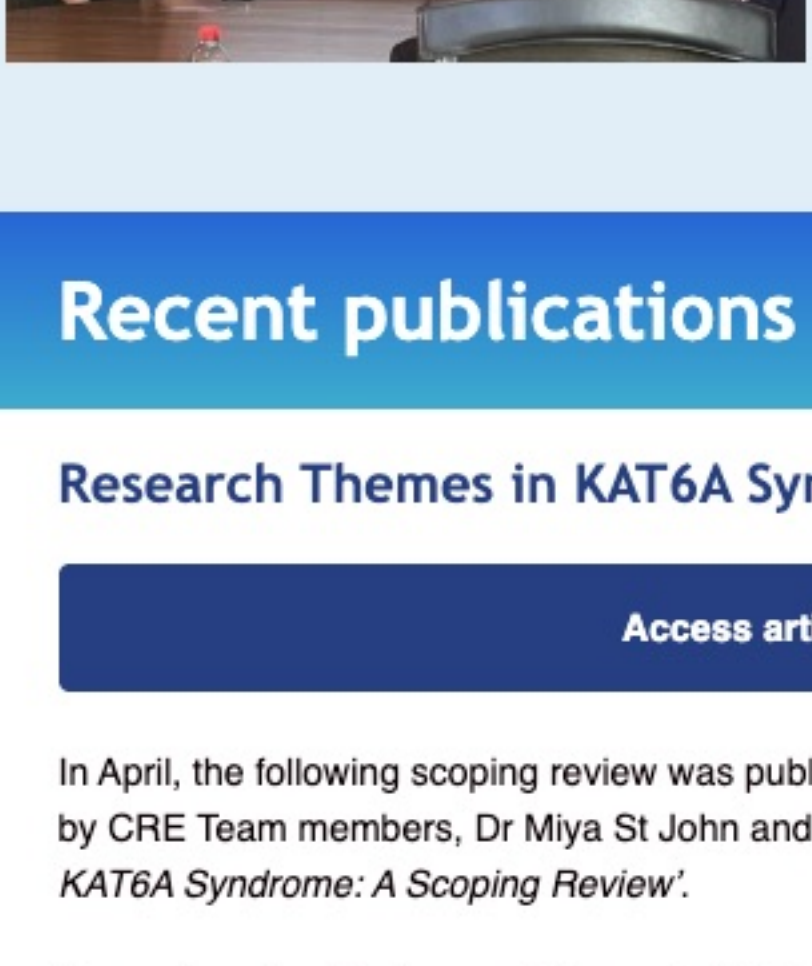
Register Interest here

Register interest or ask any questions at:

geneticsofspeech@mcri.edu.au

Conferences

GETA Family Conference



CRE team member and PhD student Alecka Garrett presented at the GETA (Genetic Epilepsy Team Australia) conference

This presentation focused on educating families of children with SYNGAP1-related disorder about the importance of detailed speech and language phenotyping to support families and bolster research in the advent of precision therapies. Alecka reported updates on the ongoing SYNGAP1 speech, language, and feeding study, addressed frequently asked questions, and outlined what families can expect if they sign up to the study.

Thank you to GETA for their invitation!

Recent publications

Research Themes in KAT6A Syndrome: A Scoping Review

Access article HERE

In April, the following scoping review was published by Dr Tanya Tripathi and co-authored by CRE Team members, Dr Miya St John and Prof David Amor: '*Research Themes in KAT6A Syndrome: A Scoping Review*'.

The review described current themes in KAT6A syndrome research, including (1) genotype-phenotype correlation, (2) the neurodevelopmental profile, (3) the epigenetic role of *KAT6A*, (4) molecular biomarkers, (5) drug discovery, and (6) the phenotypic overlap with other conditions like Rett syndrome.

This review emphasises the complexity of KAT6A syndrome and the need for a future research focus on longitudinal studies, the development of biomarkers, and precision medicine trials for individuals and their families.

Understanding speech and language in KIF1A-associated neurological disorder

Access article HERE

CRE PhD student and Speech Pathologist Lottie Morison is the lead author of the article 'Understanding speech and language in '*KIF1A-associated neurological disorder*'

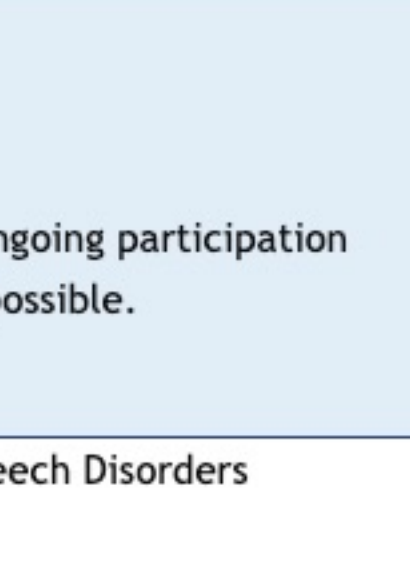
This research aimed to understand the speech and language features, support needs, and strengths in people with KIF1A-associated neurological disorder (KAND).

The researchers found there was a big range of speech and language ability in the group. Most people had delayed speech and language milestones and all people who were speaking had dysarthria. Those in the study had stronger expressive language than receptive language. Some individuals used augmentative and alternative communication (AAC) (e.g., speech generating devices, key word sign). Participants were very socially motivated.

This study improves our understanding of what therapies and supports might be helpful for people with KAND including capitalising on strengths like social motivation to support success in therapy, individualised speech and language therapies, and early and adaptable AAC.

Click the image for a Plain Language Summary

Click the image for a KAND Fact Sheet



Thank you to the individuals with KAND and their families for generously taking part in the research, KIF1A.org and KIF1A.au, and our collaborators.

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

Speech and Language Group

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