Centre of Research Excellence in Speech and Language

Newsletter

About Us

The Centre of Research Excellence in Speech and Language is an international collaboration of experts in the fields of speech pathology, paediatric neurology, neuroscience, genetics, and bioinformatics.

Our vision is to transform speech pathology practice by identifying, understanding and targeting the underlying causes of developmental speech and language disorders.

Thank you!

Our team wishes to thank families and clinicians for their ongoing participation and support.

A sincere thanks to all the families who have taken part, without your help this research would not be possible.

Latest News

New genes added to Genetics of Speech Disorders website

We have recently added the *SETBP1* gene to our website. We have also updated the website with our latest *FOXP1* findings! The website provides families and clinicians with information about genes associated with speech and language disorders. It also provides information on speech and language growth after diagnosis, evidence based interventions and ongoing research.

Visit our website!

Childhood Apraxia of Speech: Pharmaceutical Trial

Our group is trialling a new treatment for children with childhood apraxia of speech

(CAS). This treatment is the drug methylphenidate (MPH), commonly known as Ritalin. We want to find out whether MPH can help improve the speech and language outcomes for children aged 6-12 years with CAS.

MPH is approved as a treatment for ADHD in children. We want to find out if it is effective for children with CAS.

To be eligible to participate in the trial, children must: have a confirmed diagnosis of CAS, be aged 6-12 years, be able to complete spoken tasks, live within 100km of Murdoch Children's Research Institute, have some inattentive and/or hyperactive symptoms, and not have epilepsy, congenital heart disease, severe Tourette Syndrome or severe intellectual disability.

Participation in the trial will last for 2 months. It will involve only one visit to RCH for an initial health and medical screening. It will also involve four Zoom calls with the study team for speech and language assessments.

Register here!

Learn more!

Genetics of stuttering: Almost 300 Kiwis signed-up!

In November 2020, we officially launched the Genetics of Stuttering Study in New Zealand. We were lucky enough to have the support of New Zealand's Stuttering Treatment and Research Trust, and to be featured on TVNZ's Seven Sharp show. The work is also supported by Professor Lynette Sadlier and Meghan Evans at the University of Otago. The response to the study has been fantastic, with almost 300 Kiwis signing up so far. We are about to receive the first genetic data from our New Zealand participants, and are excited to get started with our analyses!

We are still continuing to recruit participants for the study from Australia and New Zealand.

Social media

If you would like to keep up to date with what the CRE Speech and Language team is doing, please follow us on Twitter, at MCRI_SpeechLang. Alternatively, you can still find us on Facebook 'Genetics of Speech and Language Disorders'.

Awards, Conferences and Events

KAT6A and *KAT6B* Symposium

On September 1st 2021, CRE PhD student, Miya St John, was invited to speak at the first *KAT6A* and *KAT6B* Virtual Symposium. The symposium was an initiative led by the *KAT6A* Foundation and will hopefully be the first of many future events. The symposium was designed to support patients and their families living with *KAT6A* and *KAT6B*-

related disorders by providing a platform and open dialogue between current researchers and collaborators. Researchers from Australia, USA, UK, Canada, and Spain presented a number of flash talks regarding their respective research areas. An ongoing crowdsourcing initiative, undertaken by the *KAT6A* Foundation, found that *KAT6A* parents and families indicated speech and language research the highest priority for patient health and quality of life. It was timely then that Miya was able to present her research investigating the speech and language profile of individuals with *KAT6A* syndrome. We thank the *KAT6A* Foundation for putting on such a wonderful event.

Human Genetics Society of Australasia 44th Annual Scientific Meeting

On August 16th this year, CRE PhD student Miya St John and Prof Morgan presented at the Human Genetics Society of Australasia 44th Annual Scientific Meeting. The meeting was a 4-day event, with the theme: "From Promise to Precision: Beyond a 2020 Vision", covering various aspects of genetics and genomics research. Miya presented her current research on speech, communicative behaviours, and adaptive functioning in individuals with *KAT6A* variants. Prof Morgan also discussed the team's findings on speech and language in children with *SETBP1* haploinsufficiency disorder. It was a diverse and thought-provoking conference, and a wonderful opportunity to present speech and language content to a genetics audience.

NHMRC Research Excellence Award

Prof Angela Morgan was awarded the Elizabeth Blackburn Investigator Grant Award (Leadership in Clinical Medicine and Science) in June 2021 at the 2020 NHMRC Research Excellence Awards. The award recognised Prof Morgan as one of the leading researchers in the field and acknowledged her work in understanding the genetic causes of speech disorders in children.

Selected recent findings and publications

FOXP1-related disorder

Our team recently published a study on the speech and language features in 29 individuals with *FOXP1* gene changes. The results showed that individuals with *FOXP1* disorder have a range of speech and language features. Speech was characterised by dysarthria and features of apraxia. Expressive language skills were stronger than receptive language skills in most individuals. These findings help to inform our understanding of speech and language in individuals with *FOXP1* disorder and will help us improve


DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY ORIGINAL ARTICLE

Severe speech impairment is a distinguishing feature of *FOXP1*-related disorder

RUTH O BRADEN[1,2] | DAVID J AMOR[1,2,3,4] | SIMON E FISHER[5,6] | CRISTINA MEI[1,2] | CANDACE T MYERS[8] | HEATHER MEFFORD[9] | DEEPAK GILL[9] | SIDDHARTH SRIVASTAVA[10] | LINDSAY C SWANSON[10] | HIMANSHU GOEL[11] | INGRID E SCHEFFER[1,2,3,12,13] | ANGELA T MORGAN[1,2,3,4]

1 Murdoch Children's Research Institute, Parkville, VIC. 2 Department of Audiology and Speech Pathology and Department of Paediatrics, University of Melbourne, Parkville, VIC. 3 The Royal Children's Hospital, Parkville, VIC. 4 Victorian Clinical Genetics Service, Parkville, VIC, Australia. 5 Language and Genetics Department, Max Planck Institute for Psycholinguistics, Nijmegen. 6 Donders Institute for Brain, Cognition and Behaviour, Radboud University, Nijmegen, the Netherlands. 7 Orygen and Centre for Youth Mental Health, University of Melbourne, Parkville, VIC, Australia. 8 Department of Pediatrics, Division of Genetic Medicine, University of Washington, Seattle, WA, USA. 9 TY Nelson Department of Neurology, The Children's Hospital at Westmead, Sydney, NSW, Australia. 10 Department of Neurology, Boston Children's Hospital, Boston, MA, USA. 11 Human Genetics, John Hunter Hospital, New Lambton Heights, NSW. 12 Austin Health, Heidelberg, Melbourne, VIC. 13 Florey Institute of Neuroscience and Mental Health, Parkville, VIC, Australia.

Correspondence to Professor Angela T Morgan, Murdoch Children's Research Institute, 50 Flemington Rd., Parkville, VIC 3052, Australia. E-mail: angela.morgan@mcri.edu.au

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Accepted for publication 6th May 2021.
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ABBREVIATION
VABS Vineland Adaptive Behaviour Scales

AIM To delineate the speech and language phenotype of a cohort of individuals with *FOXP1*-related disorder.
METHOD We administered a standardized test battery to examine speech and oral motor function, receptive and expressive language, non-verbal cognition, and adaptive behaviour. Clinical history and cognitive assessments were analysed together with speech and language findings.
RESULTS Twenty-nine patients (17 females, 12 males; mean age 9y 6mo; median age 6y [range 2y 7mo–33y; SD 8y 5mo]) with pathogenic *FOXP1* variants (14 truncating, three missense, three splice site, one in-frame deletion, eight cytogenic deletions; 28 out of 29 were de novo variants) were studied. All had atypical speech, with 21 being verbal and eight minimally verbal. All verbal patients had dysarthric and apraxic features, with phonological deficits in most (14 out of 18). Language scores were low overall. In the 21 individuals who carried truncating or splice site variants and small deletions, expressive abilities were relatively preserved compared with comprehension.

Read the article here

therapy approaches.

Current Studies

Speech Tracker

Our team have been developing a new online study called “Speech Tracker”, set to launch this month. This study builds on our previous research showing that speech and language is a core developmental challenge for children with certain genetic conditions.

In this new study, we are examining speech & language over time. Participants will be invited to complete short (15 minutes) speech tasks from home three times per year for three years. This data will help us better understand prognoses for children and families, and will help us develop more targeted speech therapies.

We are currently inviting children and adults with *SETBP1* loss of function or missense variants to take part in Speech Tracker. We then aim to expand the use of Speech Tracker to further genetic conditions of interest in coming months, so we encourage you to watch this space for updates.

Register here!

Stuttering Sub-phenotyping Study

PhD student, Sarah Horton, has begun recruiting participants for her Stuttering Sub-phenotyping Study. This study aims to investigate sub-groups of people who stutter based on features such as family history of stuttering, speech characteristics, and other health and medical conditions. Participants will complete online surveys and take part in a videoconference meeting to provide speech samples. We hope that identifying sub-groups of people who stutter with similar features will help us to better understand differences in stuttering such as why some people recover naturally and some don’t, or why some people respond to treatments and others don’t.

This project is being undertaken in conjunction with the international Genetics of Stuttering Study, which is currently recruiting participants who stutter or who have stuttered at some point in their lifetime.

Learn more!

Thank you!

Our team wishes to thank families and clinicians for their ongoing participation and support.

In the past year, we have expanded our interstate participation to reach more families across Australia.

A sincere thanks to all the families who have taken part, without your help this research would not be possible.

The Centre of Research Excellence in Speech and Language

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Speech and Language Group

Murdoch Children's Research Institute

50 Flemington Rd, Parkville, Victoria, Australia, 3052