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Translational Centre for Speech Disorders
Centre of Research Excellence

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Newsletter

Translational Centre for Speech Disorders

The Centre of Research Excellence (CRE) - **Translational Centre for Speech Disorders** is an international collaboration of experts to study the neurobiology of speech disorders such as apraxia of speech to better understand the cause.

Our vision is to improve outcomes for children with speech disorders. Our novel approach to understanding the neurobiology of speech disorders will transform diagnosis, prognosis and enable precision therapies to improve long term outcomes.

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Please visit our updated Website which has been enhanced with a Donate button allowing donations to our Translational Centre for Speech Disorders.

Latest News

New Fact Sheets

New Fact Sheets on *SETBP1* Haploinsufficiency Disorder (*SETPB1*-HD), KAT6A syndrome and *FOXP2*-related speech and language disorder (*FOXP2*-SLD) have been added to our website.

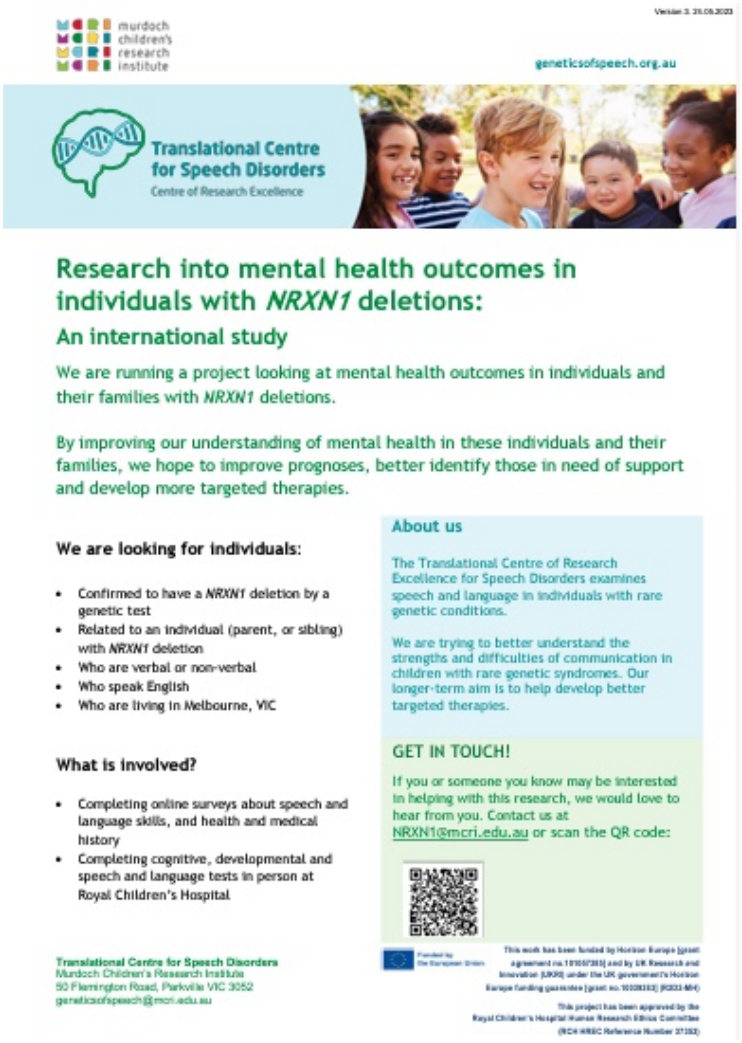
We would like to thank the SETPB1 Society, KAT6 Foundation and *FOXP2*-SLD community for their input into our Fact Sheets.

NRXN1(RaDiaNT) Study

The Speech and Language team have recently commenced a new study in collaboration with Thomas Bourgeron, Louise Gallagher and other Chief Investigators on the EU-Horizon grant on 'Risk and Resilience in Mental Health Conditions'. This international project is investigating factors that influence mental health outcomes in individuals and their families with *NRXN1* deletions.

Genetically, *NRXN1* deletion is known to play a significant role in neurodevelopment and disruptions of this gene can lead to conditions such as autism spectrum disorder, attention deficit/hyperactivity disorder, intellectual disability, language difficulties and subtle movement problems. Some individuals with *NRXN1* deletion are at a higher risk for adverse mental health outcomes.

Currently, little is understood regarding which factors contribute to positive versus more challenging mental health outcomes in individuals with *NRXN1* deletions. By improving our understanding of mental health in these individuals and their families, we hope to improve prognoses, better identify those in need of support and develop more targeted therapies.



Learn more!

Learn more about this study and how to enrol!

Stuttering sub-group study

Researchers from the Speech and Language team are investigating subgroups of people aged 5 years and older, who stutter (past or present), based on family history, speech characteristics, personal experience of stuttering, and other health and medical conditions.

Identifying subgroups of people who stutter with similar features will help us to better understand differences in stuttering such as why some people resolve naturally and some don't, or why some people respond to speech pathology treatments and others don't.

Participation in the study is free and easy. Participants are expected to complete online surveys (around 45 minutes) and attend an online Zoom session with a speech pathologist from our team (around 45 minutes). No formal diagnosis of stuttering is needed.

Find out about our current studies

Awards, Conferences and Events

Our team members have been attending and presenting at national and international conferences. We have highlighted a few of these conferences and the important research that was presented.

Symposium on Child Speech Disorders: translating cutting-edge science into clinical practice

The NHMRC Centre of Research Excellence (CRE) Translational Centre for Speech Disorders hosted their first in-person Symposium on **Monday 9 October 2023** at The Royal Children's Hospital in Melbourne.

The CRE Symposium brought together local and interstate guests from a range of disciplines including speech pathology, paediatric neurology, neuroscience and genetics to facilitate a day of stimulating presentations and networking opportunities. Prof Angela Morgan, renowned expert in speech genetics and

Director of the CRE Translational Centre for Speech Disorders, opened the day with a warm welcome to more than 200 guests.

The morning session "*Speech Pathology diagnosis, therapy and scope of practice in Genetics*" had a wide range of speakers and topics. Prof Kathryn North, Director of the Murdoch Children's Research Institute, opened the afternoon session "*Latest advances in Speech Pathology research*". Prof Angela Morgan's keynote presentation delved into genetics in speech pathology clinical practice followed by other speakers including an international presenter. As surprise act, Mr Josh Earl finished the day with a stand-up comedy act.

Prof Morgan also took the opportunity to officially thank Prof Sheena Reilly for her major contribution to the field of Speech Pathology.

Thank you to everyone who came to the Symposium and we look forward to seeing you at future events!

See full summary of the Symposium

FOXP1 International Conference

On 22 June 2023, team member Dr Ruth Braden presented virtually at the 2023 Family Day and Conference about *FOXP1*-related disorder "W*hat do we know about speech and language abilities*."

The conference was hosted by the International FOXP1 Foundation in New York, and was an opportunity for families and individuals with *FOXP1* disorder from all over the world to come together with researchers to learn about *FOXP1* disorder.

We would like to thank the organising committee and the FOXP1 Foundation board for inviting us to present, and for hosting such a successful inaugural event.

Batten Disease Support & Research Association (BDSRA) International Family Conference

Congratulations to team member and PhD candidate Lottie Morison, who came third in the NCL Researcher's Challenge short videos at the Batten Disease Support & Research Association (BDSRA) international Family Conference 14-16 July 2023. The topic of her video was "*Communication is connection: Understanding speech, language, and non-verbal communication in CLN2 and CLN3 disease*."

Earlier this year, Lottie was awarded a Batten Disease Research Grant from BDSRA Australia to help improve the quality of life of children with Batten Disease,

a genetic childhood dementia. Her research focuses on characterising speech and language in the CLN2 and CLN3 forms of Batten Disease.

Lottie also received a student scholarship from the Lysosomal Research Summit to attend their Summit on 27-29 October 2023 in Melbourne, Australia. The Lysosomal Disease Summit brought together clinicians and researchers from Australia and the Asia-Pacific region to discuss lysosomal disease awareness, resources, diagnosis, and treatment. Batten Disease is a type of lysosomal disease.

We would like to thank the BDSRA Foundation Australia for their ongoing support.

Learn more about our research into Batten Disease

Koolen-de Vries Syndrome (KdVS) Patient Advocacy Summit

On 19-21 July 2023, the KdVS Foundation hosted the fourth biennial KdVS Patient Advocacy Summit in Orlando, Florida. The summit was an opportunity for international researchers and clinicians to collaborate and share research with individuals with KdVS and their families.

Team members Dr Miya St John and Olivia van Reyk presented at the summit virtually from Melbourne. Miya presented on a feasiblity trial of a home-based speech

treatment for individuals with Koolen-de Vries syndrome. Olivia presented an update on our current Speech Tracker study tracking changes in speech and communication over time in individuals with KdVS.

Our sincere thanks to the KdVS Foundation for organising the Patient Advocacy Summit.

XXIIIrd International Congress of Genetics (ICG)

The ICG (16-21 July, Melbourne) brought together the international genetics community to share ideas and the latest research under the overarching theme: Genetics & Genomics: Linking Life & Society.

CRE investigator, Prof Michael Hildebrand, presented on the first study to link structural brain anomalies to developmental stuttering (*"Mutation of cyclophillin-40 molecular chaperone causes persistent stuttering"*). A fifth gene has been identified in severe developmental stuttering.

The results of this study have just been published (*see Selected findings and publications* for more information).

ABC interview with Prof Michael Hildebrand

PhD awarded to Dr Miya St John

Congratulations to team member, Dr Miya St John, who passed her PhD thesis "Speech and language in chromatin-related disorders" in July without corrections. Miya investigated communication skills across several chromatin-related neurodevelopmental disorders and published her findings in three key publications. In her first publication, she found that many individuals with KAT6A syndrome do not develop typical verbal/vocal communication, and alternative communication methods (like sign language or electronic communication devices) are common and recommended. In her second publication, her findings showed that many people with Koolen-de Vries syndrome (KdVS) develop verbal communication, although talking is delayed. Once talking commences, people with KdVS have complex speech diagnoses including apraxia, dysarthria, and fluctuating stuttering. Many people with KdVS have strengths in social skills and emotional control. Her final publication was a systematic review of the current evidence on speech and language in 70 chromatin disorders. Results showed that communication challenges are common across all chromatin disorders,

and specific types of chromatin genes (“writers” and “readers”) appear particularly important for motor speech development.

KAT6 Foundation grant

Recently, Dr Miya St John received a grant of $10,000USD from the KAT6 Foundation to support a research study working with individuals with *KAT6B*-related conditions. This research aims to characterize the speech and language abilities of individuals with *KAT6B*-related conditions, and in doing so will provide a clearer diagnosis, prognosis and treatment planning for families and clinicians.

DDX3X International Conference

Prof Angela Morgan was an invited keynote speaker at the first international conference on *DDX3X*-neurodevelopmental disorder (DDX3X syndrome) on 20 October in Paris, France followed by a family day. The conference brought together doctors, families, researchers and clinicians to share knowledge and best practices. Prof Morgan presented on the speech and language characteristics in individuals with *DDX3X*-neurodevelopmental disorder.

DDX3X-neurodevelopmental disorder causes neurodevelopmental conditions, such as intellectual disability and autism. The team is currently preparing a publication with results from their research on speech and language features in *DDX3X*-neurodevelopmental disorder.

The team would like to thank Association Xtraordinaire, DDX3X Foundation and GENIDA for their continued support.

Visit to the House of Commons, UK Parliament

Speech and language therapists, charities, parents and carers, and MPs and peers converged in the House of Commons (UK) on Thursday, 19 October in London, for a special parliamentary reception to increase awareness of childhood apraxia of speech and other speech and language difficulties.

Mikey Akers is a young adult who spoke to raise awareness of childhood apraxia of speech.

Prof Angela Morgan and others from the US contingent of Apraxia Kids were in attendance to support Mikey's wish to raise awareness.

Mikey Akers

Mikey Akers and family to the left standing amongst the US contingent from Apraxia Kids (Prof Morgan - far right first row).

Travers Reid Award

Congratulations to team member and PhD student Sarah Horton who has received the Travers Reid Award this year. The Travers Reid award is open to any student presenting at the 13th Oxford Dysfluency conference, and recognises the contribution of research to the lives of children and young people who stammer.

Although the 13th Oxford Dysfluency conference in September was cancelled due to unforeseen circumstances, an international sub-committee hosted an online session last week to allow students to present their work.

Sarah was successful in receiving the award for her presentation titled “Executive function, stuttering severity, and subjective impact of stuttering.”

Selected recent findings and publications

Barriers and enablers for Genetics in Speech-Language Pathology

Team member Mariana Lauretta, is the lead author on a published paper exploring the barriers and enablers to incorporating genetics in speech pathology clinical practice. Twelve paediatric speech pathologists were interviewed to determine their attitudes and beliefs associated with genetics as it relates to a case study of a child with childhood apraxia of speech. This study identified barriers including lack of confidence, knowledge, and belief about effects on the family. Enablers were also identified, including seeing value in genetic diagnosis and establishing relationships with genetics services. The findings from this study may assist with the development of strategies to support speech pathologists to incorporate genetics into future clinical practice.

PubMed

An Investigation of Barriers and Enablers for Genetics in Speech-Language Search

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J Speech Lang Hear Res. 2023 Sep 15;1-15. doi: 10.1044/2023_JSLHR-22-00714. Online ahead of print.

An Investigation of Barriers and Enablers for Genetics in Speech-Language Pathology Explored Through a Case Study of Childhood Apraxia of Speech

Mariana L Lauretta [1], Anna Jarmolowicz [1], David J Amor [1 2 3], Stephanie Best [4 5 6 7], Angela T Morgan [1 2 3 8]

Affiliations + expand

PMID: 37713535 DOI: 10.1044/2023_JSLHR-22-00714

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Abstract

Purpose: Advancements in genetic testing and analysis have allowed improved identification of the genetic basis of childhood apraxia of speech, a rare speech presentation. This study aimed to understand speech-language pathologists' (SLPs') consideration of incorporation of genetics in clinical practice using a theory-informed qualitative approach.

Method: Semistructured interviews were conducted with 12 pediatric SLPs using a behavior change theory (Theoretical Domains Framework [TDF]) within a case study describing a child with complex co-occurring features, including childhood apraxia of speech. Interviews focused on three stages of the patient journey (prereferral, referral, and postreferral). Interviews were analyzed to identify barriers and enablers to considering incorporation of genetics in current clinical practice. Barriers and enablers were grouped and mapped onto a contextually relevant TDF-coded analysis framework.

Results: Barriers were identified across several TDF domains, through all stages of the patient journey. Lack of confidence, relevance, and level of experience were most common prereferral, and connection to and awareness of genetics services and contextual factors were barriers in the referral stage. Perception of professional role, knowledge, and beliefs about effects on families were barriers postreferral. Associated enablers were also identified, including seeing value in genetic diagnosis, support from other health care professionals, supervision, and relationships with genetics services.

Conclusions: Results of this qualitative study highlight barriers and enablers to incorporating genetics into speech-language pathology clinical practice. These findings will assist in the development of theory-informed implementation strategies to support SLPs into the future.

Supplemental material: https://doi.org/10.23641/asha.24112800.

Read more about the article

A study of members of a four-generation Australian family has revealed a link between a new gene pathway and structural brain anomalies in some people who stutter into adulthood.

Read more about this publication

Gene linked to persistent stuttering into adulthood uncovered

A new study led by CRE investigators Professors Michael Hildebrand and Angela Morgan, has discovered a link between a new gene pathway and structural brain anomalies in some people who stutter into adulthood, opening up promising research avenues to enhance the understanding of persistent developmental stuttering.

Professor Hildebrand and Professor Morgan led a large team of international researchers across 18 institutions to implicate a fifth gene, called *PPID*, in severe developmental stuttering, linking a new 'chaperone pathway' to the disorder. Chaperones are proteins that shuttle other proteins to the correct part of a cell so they can complete their function. The researchers suspect the damaged gene will change movement and function of proteins during brain development, triggering neural changes that cause persistent stuttering.

Published in the leading international neuroscience and translational journal *Brain*, researchers studied 27 members of a four-generation Australian family, 13 of whom have stuttering.

Stuttering is a speech disorder affecting around five per cent of children and one per cent of adults worldwide. In over two thirds of cases of stuttering in childhood, the stuttering eventually resolves with therapy. However, in severe cases like most individuals in the family studied here, the disorder can persist into adulthood. Despite stuttering being commonly inherited, only four genes have previously been implicated in stuttering.

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 our research would not be possible.

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