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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: investigating speech and language in individuals with PURA syndrome

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 PURA syndrome.

PURA syndrome is a rare genetic condition, characterised by developmental delay, hypotonia and communication difficulties. However, the speech and language profile has not been described.

This study aims to investigate the speech and language skills in PURA syndrome, including understanding the use of alternative forms of communication such as sign language or communication devices.

👶 Eligibility:
🔍 Aged 6 months and older
🔍 Confirmed diagnosis of PURA syndrome by genetic test
🔍 Who are verbal or non-verbal/non-speaking

✅ What is involved:
✏️ Completing surveys about the individual's speech, language skills, health and medical history
✏️ An in-person or online Zoom session with a Speech

Pathologist for further assessment (1 hour), where able
✏️ Surveys are available in English, French, Dutch, German, Spanish, Portuguese, Italian, or Chinese

Interested families are encouraged to contact the study team at geneticsofspeech@mcri.edu.au or reach out to the lead investigator, Dr. Miya St John at miya.stjohn@mcri.edu.au.

We would like to sincerely thank the PURA Foundation Australia for their wonderful support and fundraising efforts to facilitate this important research.

Register your interest to participate

Research into speech change with methylphenidate (Ritalin) in children with apraxia of speech

Researchers from the Speech & Language team at MCRI are continuing to investigate if the medication methylphenidate (Ritalin) can improve speech and language outcomes for children with Childhood Apraxia of Speech (CAS).

👶 Eligibility:
🔍 Aged 6-12 years
🔍 Diagnosed with CAS
🔍 Within 250km of Royal Children's Hospital Melbourne

✅ What is involved:
✏️ Online meetings
✏️ Initial hospital visit
✏️ 2-month trial with medication and placebo phases
✏️ Speech assessments

Hear Lorraine O'Donnell discuss this study

Learn more about the trial and how to enrol!

NEW STUDY: investigating speech and language in individuals with Sanfilippo syndrome

Our Speech & Language team at MCRI has recently launched an international research study examining speech and language outcomes in individuals (aged 6 months and older) with Sanfilippo syndrome (MPS III).

Sanfilippo syndrome (MPS III) is a rare genetic condition that leads to childhood dementia. It is estimated that 1 in 70,000 children worldwide are born with this inherited condition. The first sign of the disease is often a delay or regression in speech and language, yet a comprehensive speech and language profile has not been described.

The aim of this study is to enhance our understanding of the speech and language profiles associated with this condition. We hope to facilitate earlier referrals for diagnosis, identify those in need of support, and develop targeted intervention strategies.

👶 Eligibility:
🔍 Aged 6 months and older
🔍 Confirmed diagnosis of Sanfilippo syndrome by genetic test
🔍 Who are verbal or non-verbal
🔍 At any stage of their progression

✅ What is involved:
✏️ Completing online surveys on the individual's speech and language skills, and health and medical history
✏️ An in-person or online Zoom session with a Speech Pathologist for further assessment (1 hour), where able

The study is being conducted by CRE PhD candidate Jen Wong, 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.

We extend our heartfelt gratitude to the Sanfilippo Children's Foundation and the Childhood Dementia initiative for their valuable support of this important research.

Hear more about the study in this video featuring PhD candidate Jen Wong

Awards, Conferences & Events

HAROLD MITCHELL FOUNDATION POSTDOCTORAL TRAVELLING FELLOWSHIP

CRE team member Dr. Miya St John was awarded a Harold Mitchell Foundation Postdoctoral Travelling Fellowship. Miya will use the funds from this award to present at the international Koolen-de Vries summit in July next year. She will also learn a long-established therapy for Childhood Apraxia of Speech with our American colleagues.

Dr. Miya St John.

AUSTRALIAN STUTTERING CONFERENCE

The University of Melbourne's Department of Audiology and Speech Pathology, in collaboration with Melbourne Speech Pathology Clinic, hosted its inaugural Australian Stuttering Conference in Melbourne (22 November). The conference aimed to bring together researchers and clinicians to highlight the latest advancements in the diagnosis and management of stuttering, both locally and internationally, and to discuss how these advances inform clinical practice.

CRE Director Prof. Angela Morgan and PhD Student Sarah Horton were invited speakers at this event. Prof. Morgan delivered a talk titled "Progress in understanding the genetics of stuttering," discussing the latest genetic research findings related to stuttering and their implications for clinical practice. Ms. Horton presented on "Variability of Stuttering: implications for clinical outcome measurement." This study forms part of her PhD, which focusses on sub-phenotyping in an international genome-wide association study of people who stutter.

We would like to thank the organisers, in particular Dr. Elaina Kefalianos, for this wonderful and engaging event.

Ms. Sarah Horton presenting at the conference.

NATIONAL CONFERENCE IN SPEECH THERAPY

CRE Director Prof. Angela Morgan was invited as plenary speaker at this national conference in Sweden (14-15 November).

The theme for this year's conference was "Opportunities and challenges within today's speech therapy." The conference was attended by speech therapists and researchers in speech therapy and related fields.

Prof. Morgan presented on "The genetic bases of developmental speech and language disorders," highlighting her team's research into the genetic causes of speech disorders. The CRE team has identified over 30 causative genes that had not previously been linked to severe speech disorders.

We would like to thank the organiser, the Sahlgrenska Academy from the University of Gothenburg, for their invitation to present.

Prof. Morgan delivering her plenary talk.

SOCIETY FOR NEUROBIOLOGY OF LANGUAGE (SNL)16th ANNUAL MEETING

Prof. Angela Morgan was an invited keynote speaker at the Society for Neurobiology of Language 16th Annual Meeting (Brisbane, 24-26 October). SNL is a National Institutes of Health (NIH)-funded non-profit organisation whose overarching goal is to foster progress in understanding the neurobiological basis for language via the interdisciplinary exchange of ideas.

Prof. Morgan presented on "Genetic architecture of childhood speech disorders" highlighting that in the last few years, her team have shown that childhood apraxia of speech (CAS) arises from a genetic cause in 1 in 3 children. A full description of Prof. Morgan's keynote lecture can be found here. For more information on CAS, see our website.

We would like to thank the Society for the Neurobiology of Language for hosting such a successful conference.

Prof. Morgan's keynote lecture.

INTERNATIONAL CONFERENCE ON STUTTERING

CRE PhD student Sarah Horton presented at the International Conference on Stuttering held in Rome, Italy (24-26 October). The hybrid conference centered on the theme “Expectations and needs of people who stutter: how researchers and clinicians answer the call.”

In her presentation, Sarah shared findings from an observational study investigating variability of stuttering in children and adults. Sarah discussed the clinical implications of stuttering variability, including the need to look beyond observable stuttered speech and to also consider the impact of stuttering on the individual.

The conference brought together clinicians, researchers, and people who stutter, creating a unique opportunity for attendees to engage in discussions about innovative tools and therapies designed to support those who stutter.

Some of the speakers and research collaborators at the conference with Ms. Horton (top left).

2ND ANNUAL LYSOSOMAL DISEASE SUMMIT

CRE PhD student Lottie Morison presented at the 2nd Annual Lysosomal Disease Summit (Melbourne, 18-20 October). The Lysosomal Disease Summit brought together clinicians and researchers from Australia and the Asia-Pacific region to discuss metabolic and

lysosomal diseases. Lottie received a student scholarship from the Lysosomal Disease Summit.

Lottie presented her research characterising speech, language, and non-verbal communication in CLN2 and CLN3 Batten disease. CLN2 and CLN3 are the most common types of Batten disease, a group of rare lysosomal storage disorders that cause childhood dementia.

We would like to sincerely thank all the families who participated in our research and our team of collaborators. We look forward to sharing the study results with you once these are published.

This research was support by BDSRA Australia's 2022 research grant.

Ms. Lottie Morison presenting.

SPEECH AND LANGUAGE DISORDERS: LESSONS FROM GENETICS (SYMPOSIUM)

The Max Planck Institute for Psycholinguistics held a 1-day Symposium in Nijmegen, the Netherlands (13 September), focusing on recent advances in language genetics. This event aimed to foster discussions among a diverse, multidisciplinary audience, bringing together leading experts from fields such as genomics, clinical genetics, and speech/language therapy. The symposium addressed various speech and language disorders, including stuttering, dyslexia, and childhood apraxia of speech (CAS). Prof. Morgan presented on the monogenic causes for childhood apraxia of speech.

Prof. Morgan also participated in the Max Planck Institute of Psycholinguistics Colloquium lecture on 10 September with a presentation titled "Unravelling the genetic bases of childhood communication disorder."

We extend our heartfelt thanks to the organising committee and hosts, Dr. Else Eising, Dr. Marscha Engelen, Drs. Marie-Christine Franken, and Prof. Simon Fisher for organising this impactful symposium.

Speakers at the Symposium with Prof. Morgan (far right).

STUDENT BURSARY AWARD

CRE PhD student Lottie Morison was awarded a student bursary to attend the International Dementia Conference 2024 (5-6 September) in Sydney.

The conference organisers we were particularly impressed by Lottie's passion and dedication to her research in Batten Disease, a form of childhood dementia.

Lottie's PhD research focusses on characterising speech and language in Batten Disease, the most common type of childhood dementia.

Ms. Lottie Morison.

Selected recent publications

Inherited PURA Pathogenic Variant Associated With a Mild Neurodevelopmental Disorder

CRE members Hildebrand, Morgan, Lauretta, Amor, Scheffer & other team members and collaborators, have published an article in Neurology Genetics: *Inherited PURA Pathogenic Variant Associated With a Mild Neurodevelopmental Disorder*.

Purine-rich element-binding protein alpha (PURA) regulates gene expression and is ubiquitously expressed with an enrichment in neural tissues. Pathogenic variants in *PURA* cause the neurodevelopmental disorder

PURA syndrome that has a variable phenotype but typically comprises moderate-to-severe global developmental delay, intellectual disability (ID), early-onset hypotonia and hypothermia, epilepsy, feeding difficulties, movement disorders, and subtle facial dysmorphism. Speech is reportedly absent in most, but the specific linguistic phenotype is not well described.

This study describes an Australian family comprising a mother-child duo with primary speech disorder (dysarthria), borderline intellect, oropharyngeal dysmotility (gastrointestinal disorder), and facial dysmorphism without other common or severe features of PURA syndrome.

The researchers used genome sequencing and identified a pathogenic gene variant as part of a study of children with severe primary speech disorder in the absence of moderate or severe ID. The study showed for the first time that a germline *PURA* pathogenic variant may be inherited and that variants in *PURA* should be considered in a broader spectrum of phenotypes including speech disorder and borderline intellect. Given the relatively small cohorts of individuals with PURA syndrome available, the research team expects the inclusion of such milder symptoms to increase diagnosis and improve management of PURA syndrome. Because the phenotypes of PURA syndrome have been reported as difficult to recognise in daily clinical practice, formal assessment of speech and language disorder and consideration of mild intellectual disability and family history are likely to enhance clinical diagnosis.

The team would like to thank Ms. Mel Anderson and the wider team of families from the PURA Foundation Australia for their support.

Neurol Genet. 2024 Aug 6;10(5):e200181. doi: 10.1212/NXG.0000000000200181

Inherited *PURA* Pathogenic Variant Associated With a Mild Neurodevelopmental Disorder

Michael S Hildebrand [1,✉], Ruth O Braden [1], Mariana L Lauretta [1], Antony Kaspi [1], Richard J Leventer [1], Melinda Anderson [1], Himanshu Goel [1], Melanie Bahlo [1], Ingrid E Scheffer [1], David J Amor [1], Robert Janowski [1], Dierk Niessing [1], Angela T Morgan [1,✉]

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

Speech and Language Group

Murdoch Children's Research Institute

50 Flemington Rd, Parkville, Victoria, Australia, 3052

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Speech and Language Group
Murdoch Children's Research Institute