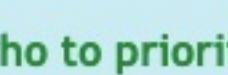




## Newsletter

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



### Recent publications

#### Childhood motor speech disorders: who to prioritise for genetic testing

[Access article HERE](#)

New paper from our team outlining genomic findings in a cohort ascertained for childhood motor speech disorder is now freely available to read in the European Journal of Human Genetics.

This study aimed to understand which clinical variables in children with a speech motor disorder (CAS/dysarthria) were predictive of a genetic diagnosis. This work helps guide recommendation and preparation for integrating of genetic testing in childhood motor speech disorder.

##### Key findings:

- 44/153 (29%) children had a genetic diagnosis
- This is similar to disorders where clinical genetic testing is routine (epilepsy, intellectual disability and cerebral palsy)

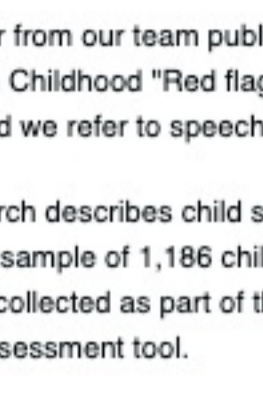
##### Clinical variables associated with a genetic diagnosis:

- Delayed walking
- Fine and gross motor disorder
- Receptive language impairment and/or cognitive impairment
- Dysmorphism

##### What does this mean?

Genetic testing for children with motor speech disorder should be prioritised for children with co-occurring motor, language and/or learning deficits

[Click the image for a Plain Language Summary](#)



[Click the image for a Childhood Apraxia of Speech Fact Sheet](#)



#### Red flags for speech impairment: who should we refer to speech therapy?

[Access article HERE](#)

New paper from our team published in Archives of Disease in Childhood "Red flags for speech impairment: who should we refer to speech therapy?".

The research describes child speech development in a normative sample of 1,186 children aged 2-12 years. Data was collected as part of the development of a digital speech assessment tool.

##### Key findings:

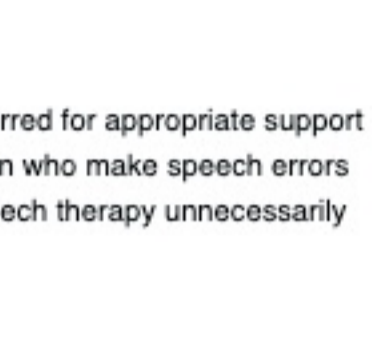
- Many children make speech errors until 6 years old
- Some parts of speech development was slower compared to 20 years ago, but not more disorder.
- We developed a tool to identify which children may be at risk of speech disorder.

##### What does this mean?

Children at risk of speech disorder may be identified and referred for appropriate support sooner, rather than a "watch and wait" approach and children who make speech errors similar to their peers may be less likely to be referred to speech therapy unnecessarily

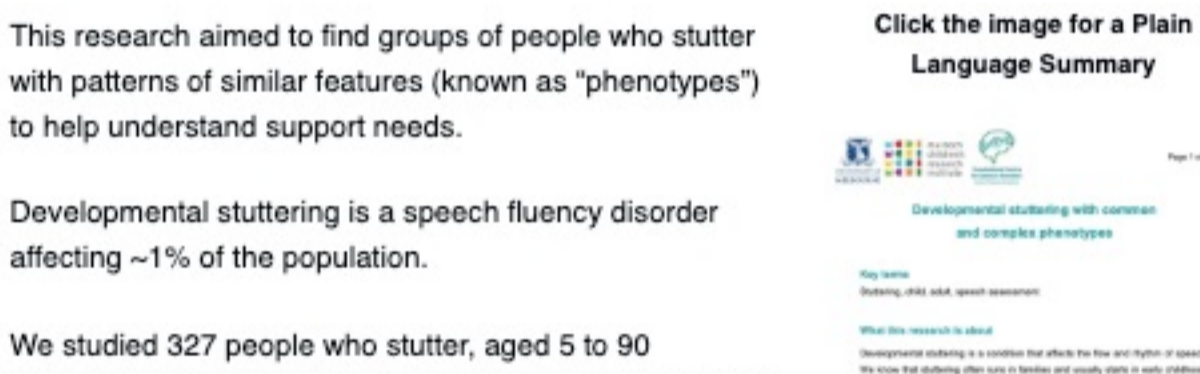
[Read the MCR news article about this research here](#)

[Click the image for a Plain Language Summary](#)



#### Tool for identifying children at risk of speech disorders

Article published 1/20 February 2023



This paper focuses on speech sound and phonological development. Analysis of oral motor and motor speech features is underway for this sample, so stay tuned for further publications from our team.

#### Developmental stuttering with common and complex phenotypes

[Access article HERE](#)

A new paper from our team led by Dr Sarah Horton is now published in Developmental Medicine and Child Neurology: "Developmental stuttering with common and complex phenotypes."

This research aimed to find groups of people who stutter with patterns of similar features (known as "phenotypes") to help understand support needs.

Developmental stuttering is a speech fluency disorder affecting ~1% of the population.

We studied 327 people who stutter, aged 5 to 90 years. We asked questions about their stuttering and how it affected them, other health conditions, and their development in early life. We also assessed their speech and thinking abilities. We used this to look for groups of people who stutter with patterns of similar features ("phenotypes").

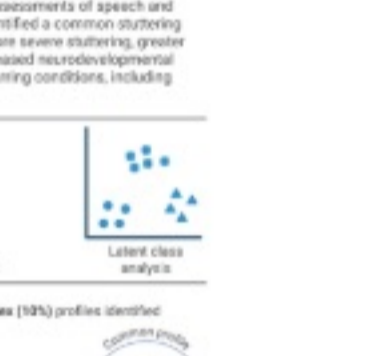
##### Key findings:

- Participants commonly reported co-occurring sleep, hearing, vision, immune system, and mental health conditions
- Stuttering was most severe in young adults
- 2 groups of participants: 90% with a "common phenotype" and 10% with a "complex phenotype."
- People in the complex group had more severe stuttering, greater negative impact of stuttering, higher anxiety, lower nonverbal intelligence, and developmental delays in early life.

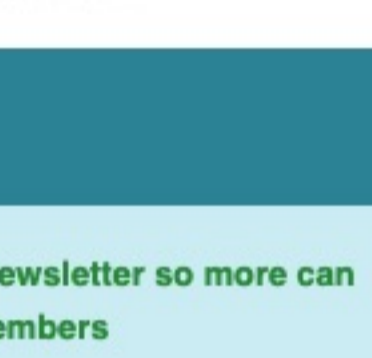
##### What does this mean?

This research shows that people who stutter often need multidisciplinary support beyond speech therapy alone. Identifying these two groups could help healthcare providers to recognise when extra support is needed.

[Click the image for a Plain Language Summary](#)



[Click the image for a Stuttering fact sheet](#)

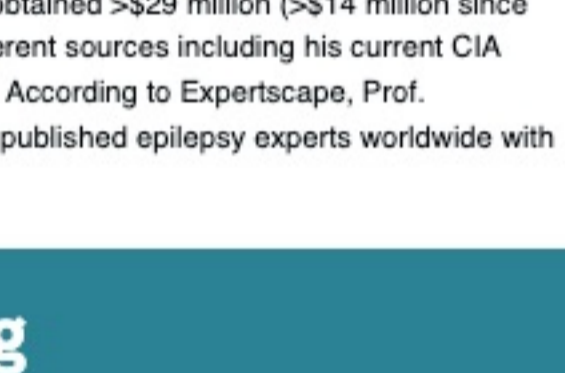


### Investigator profile feature

We are pleased to share a featured profile each newsletter so more can be learned about our CRE team members

**Professor Michael Hildebrand** is a senior neurogenetics researcher who heads the Translational Neurogenetics Laboratory and was promoted to Professor of Medicine at The University of Melbourne in 2023 at age 42 years.

Prof. Hildebrand's research has led to significant advancements in our understanding of the aetiology and genetic architecture of epilepsy, brain and vascular malformations, speech and language disorders, and hearing loss. He has identified many epilepsies, speech and language disorder, and hearing loss genes.



Conceptual advances include elucidation of the hidden genetics of epilepsies, particularly mosaic variants in focal epilepsies and vascular lesions ([Hum Mol Genet 2025](#), [JAMA Neurol 2023](#), [Neurology 2022](#), [Hum Mutat 2022](#), [Hum Mol Genet 2022](#), [Neuro Genet 2018](#), [Am J Hum Genet 2018](#), [Neuro Genet 2016](#)), the first high depth gene panel for focal epilepsy ([Neurology 2016](#)), and the innovative CSF liquid biopsy ([Brain Commun 2021](#), [Ann Neurol 2022](#)) and first SEEG mosaic gradient ([Neurology 2022](#)) to detect brain mosaicism.

Genetic contributions to speech and language disorders were poorly understood until Prof. Hildebrand led discovery of 33 new genes via genome-wide sequencing ([Mol Psych 2022](#), [Neuro 2020](#), [Mol Psych 2019](#)). These discoveries have had clinical impact with new genetic diagnoses informing prognosis, genetic counselling and targeted therapies including a MRFF RCRDUN funded clinical trial of mTOR/Ras inhibitors for epilepsy lesions. Conceptual advances include elucidation of the hidden genetics of epilepsies, particularly mosaic variants in focal epilepsies and vascular lesions ([Hum Mol Genet 2025](#), [JAMA Neurol 2023](#), [Neurology 2022](#), [Hum Mutat 2022](#), [Hum Mol Genet 2022](#), [2021](#), [Neuro Genet 2018](#), [Am J Hum Genet 2016](#)), the first high depth gene panel for focal epilepsy ([Neurology 2016](#)), and the innovative CSF liquid biopsy ([Brain Commun 2021](#), [Ann Neurol 2022](#)) and first SEEG mosaic gradient ([Neurology 2022](#)) to detect brain mosaicism.

Prof. Hildebrand has authored > 175 articles mostly in leading journals (e.g. Brain, Neurology, Am J Hum Genet), including >60 in the last 5 years. Michael's work has been cited >10,900 times (Google Scholar). He has obtained >\$29 million (>\$14 million since 2021) in competitive research funding from different sources including his current CIA MRFF Genomics Health Futures funding grant. According to Expertscape, Prof. Hildebrand ranks in the top 0.33% of >127,000 published epilepsy experts worldwide with h-index 58.

### Studies Recruiting

#### Speech, language, and feeding in individuals with KCNQ2-related condition

We are recruiting for a study looking at speech, language, and feeding in people with KCNQ2 Developmental and Epileptic Encephalopathy.

##### Eligibility:

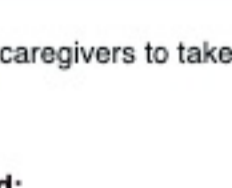
- Aged 6 months to adulthood
- Diagnosed with a KCNQ2 variant by genetic test
- Verbal or non-verbal/non-speaking

##### What's Involved:

- Online or in-person meeting
- Surveys about communication, feeding, epilepsy, 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:  
[genetics@speech@mcri.edu.au](mailto:genetics@speech@mcri.edu.au)



#### Understanding the genetics of stuttering in families

We are currently recruiting for a new project to understand more about the genetics of stuttering. This study will look at individual gene changes in a person who stutters and how these are the same or different to their family members.

##### Eligibility:

- Aged 5 years +
- *parents may also be invited to take part*

##### What's Involved:

- Provide a saliva or blood sample
- Complete a 1-hour speech and language assessment online or in person
- Complete online surveys about communication and health

[Register Interest HERE](#)

Register interest or ask any questions at:  
[genetics@speech@mcri.edu.au](mailto:genetics@speech@mcri.edu.au)



#### Experiences of support-seeking in adolescents who stutter and their caregivers

We are seeking adolescents and young adults who stutter and their caregivers to take part in research interviews.

##### Eligibility:

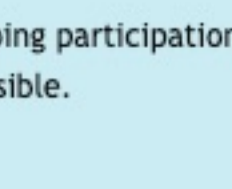
- Adolescents and young adults aged 10 to 25 years who stutter who are living in Australia.
- Caregivers of adolescents who stutter who are living in Australia.

##### What's Involved:

- Completing an online survey (approximately 15 minutes)
- Meeting a researcher for an online interview (approximately 1 hour)

[Register Interest HERE](#)

Register interest or ask any questions at:  
[sarah.horton@mcri.edu.au](mailto:sarah.horton@mcri.edu.au)



#### High-tech augmentative and alternative communication for childhood dementia

We are recruiting for a new study for individuals with childhood dementia to participate in a communication therapy trial.

##### Eligibility:

- Aged 3 to 12 years
- Diagnosed with a childhood dementia genetic condition
- Minimally speaking or non-verbal
- Not currently using a communication device proficiently
- Living in Victoria

##### What's Involved:

- 6 weeks of speech therapy sessions (2 x 1-hour sessions per week)
- 6 weeks of usual care (regular phone or video calls to check in)
- Online surveys and in person assessments to measure progress

[Register Interest HERE](#)

Register interest or ask any questions at:  
[aactrial@mcri.edu.au](mailto:aactrial@mcri.edu.au)



### Rare disease day

February 28th, was rare disease day!

Our research works to understand speech and language in rare conditions to improve diagnosis, prognosis and translational outcomes for children. We know too often those with rare conditions have their story go unheard. Learn more about Rare Disease Day [here](#)

Please check out our Instagram or Facebook pages for more insights!



### 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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