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Translational Centre for Speech Disorders
Centre of Research Excellence

# Centre of Research Excellence

# Translational Centre for Speech Disorders

# Newsletter

## New Translational Centre for Speech Disorders

It has been a while since we have been in touch, but we have exciting news to share.

Funding for the Centre of Research Excellence (CRE) in Speech and Language ceased earlier this year, however the Speech and Language Team at the Murdoch Children's Research Institute was able to secure funding from the National Health and Medical Research Council (NHMRC) for a Centre of Research Excellence for a 5-year period (2023-2028) to launch a **Translational Centre for Speech Disorders.**

Please visit our updated Website which has also been enhanced with a search bar.

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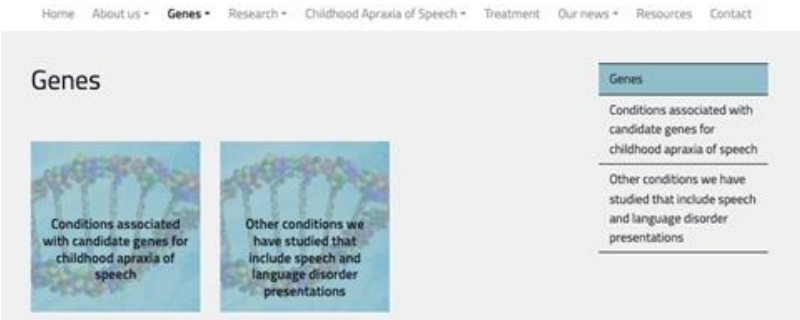
National and international experts will collaborate to study the neurobiology of speech disorders to better

understand the cause of speech disorders such as apraxia of speech. Findings from this research will help transform diagnosis and prognosis and enable precision therapies to improve long term outcomes for children with speech disorders.

**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.


# Latest News




See our Resources!

## New genes added to Genetics of Speech Disorders website

We have recently added more genes findings to our website such as *ARHGEF9*, *BRPF1* and *PHF21A*. We have also updated the website with our latest *NRXN1* findings!

We have also recently updated our fact sheet resources for articulation disorder, phonological disorder, childhood apraxia of speech, dysarthria, stuttering, and augmentative and alternative communication (AAC). In addition, a fact sheet for Koolen-de Vries syndrome is

available. 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.

## The ADAAPT Study: developing a digital speech test

The '**ADAAPT**: **A**ssessment of **D**ysarthria, **A**rticulation, **A**praxia and **P**honology with **T**reatment planning Study' aims to develop a new digital assessment tool for speech disorders in children and adolescents.

Our researchers Olivia van Reyk and Charlotte Boulton, have visited 9 schools and 14 childcare centres and kindergartens from a range of metro and regional locations, and have assessed more than 1100 children for the ADAAPT Study so far. To all the school, childcare centres, kindergartens, families and children who have participated in our research, we want to say a **big thank you**!

The next steps in the study include analysing the data from these assessments to assist further in understanding speech development and refining the tool.

**We are also continuing to recruit children and adolescents with speech difficulties to take part in this study.** The study involves a free speech assessment and summary report.

We are looking for participants:

- Aged 3-16
- Who use speech for phrases of 2 or more words
- Who may have speech difficulties as a result of:
  - Cerebral palsy
  - Brain tumour
  - Stroke or brain injury
  - Epilepsy
  - Genetic conditions
  - Autism spectrum disorder
  - Other complex neurodevelopmental conditions

*This project is funded by a National Health and Medical Research development grant.*


Learn more!


Learn more!

## Expansion of Speech Tracker study - *FOXP2*

The online study called '**Speech Tracker**', was launched end 2021 and builds on our previous research showing that speech and language is a core developmental change for children with certain genetic conditions. In this study, we are examining speech and language over time. Participants are invited to complete short (15 minutes) speech tasks from home two times per year for three years. This data will help us to better understand prognoses for children and families, and to develop more targeted speech therapies.

So far, we have been recruiting children and adults with ***FOXP1*-related disorder, Koolen-de Vries Syndrome** and ***SETBP1* haploinsufficiency disorder**. We are now also inviting children (aged 6 months to adulthood) with ***FOXP2*** to take part in **Speech Tracker**.

We would like to thank the Koolen-de Vries Foundation and SETBP1 Society for their input during the development of the study and their ongoing support of the work.

# 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.


## HGSA 45th Annual Scientific Meeting

On 24-27 November 2022, CRE team members Dr Emma Baker and Ms Miya St John attended the Human Genetics Society of Australasia (HGSA) 45th Annual Scientific Meeting which was held in Perth. The meeting was an opportunity for researchers to collaborate and share their research with a focus on recent developments in genetics and genomics. Miya presented on 'Expanding the speech and language phenotype in Koolen-de Vries syndrome: late onset and period stuttering a novel feature'. Emma presented a poster on the adaptive profiles of children with monogenic neurodevelopmental disorders.

## Kleefstra Syndrome conferences

In June of this year, PhD Student, Lottie Morison, presented at two European conferences on the speech and language in Kleefstra Syndrome: a presentation at the **Kleefstra Syndrome Scientific Conference** in Slovenia and a poster at the **annual European Society of Human Genetics Conference** in Scotland. These conferences were an opportunity for international researchers and clinicians to collaborate and share research about Kleefstra Syndrome and rare genetic conditions more broadly.

Our sincere thanks to Dr Tanja Draksler and Prof Tjitske Kleefstra for organising the Kleefstra Syndrome conference.



## 14th International Congress of Human Genetics

The 14th International Congress of Human Genetics was held on 25 February this year in Cape Town, South Africa. Prof Angela Morgan was an invited keynote speaker who spoke about the genetic bases of severe childhood speech disorders.

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

We are delighted to host our first **Symposium** on **Monday 9 October 2023** (9am- 16.45pm followed by light refreshments) at the Ella Latham Auditorium (The Royal Children's Hospital, Melbourne).

Our **Symposium** is a full-day (in-person only) event with talks by leading national and international experts in the fields of speech pathology, paediatric neurology, neuroscience and genetics.

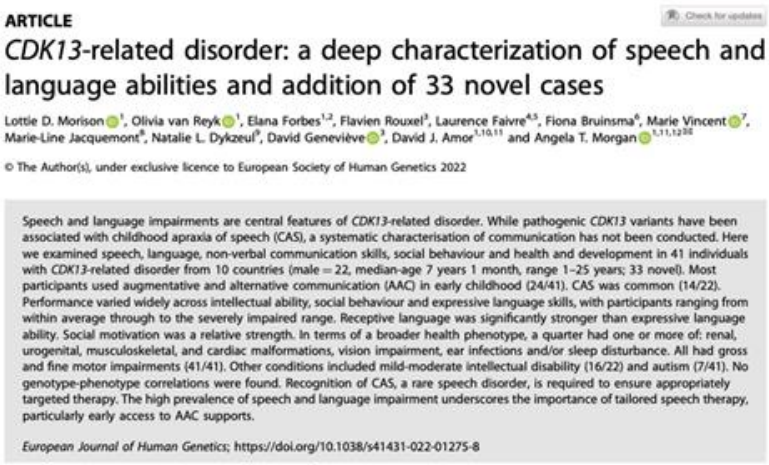
The event is free to attend and suited to speech pathologists, clinicians, allied health professionals and researchers across many disciplines including neurology, neuroscience, genetic counselling and genetics.

For our Preliminary Program please click HERE. Please note registration is required (closing date 28 September) and now open.

# Selected recent findings and publications

## *CDK13*-related disorder

Our team recently published a paper characterising 41 individuals with *CDK13*-related disorders (33 of these children have not been previously published in the scientific literature). This study identified that many individuals use augmentative and alternative communication (ACC) systems in early childhood. Receptive language skills were significantly stronger than expressive language skills across the group, and many children had childhood apraxia of speech (CAS). This study informs our understanding of speech, language, and social communication skills in *CDK13*-related disorder. This information can, in turn, help families and clinicians best support the communication development of individuals with *CDK13*-related disorder.



ARTICLE

*CDK13*-related disorder: a deep characterization of speech and language abilities and addition of 33 novel cases

Lottie D. Morison[1], Olivia van Reyk[1], Elana Forbes[1,2], Flavien Rouxel[3], Laurence Faivre[4,5], Fiona Bruinsma[6], Marie Vincent[7], Marie-Line Jacquemont[8], Natalie L. Dykzeul[9], David Geneviève[3], David J. Amor[1,10,11] and Angela T. Morgan[1,11,12]✉



Speech and language impairments are central features of *CDK13*-related disorder. While pathogenic *CDK13* variants have been associated with childhood apraxia of speech (CAS), a systematic characterisation of communication has not been conducted. Here we examined speech, language, non-verbal communication skills, social behaviour and health and development in 41 individuals with *CDK13*-related disorder from 10 countries (male = 22, median-age 7 years 1 month, range 1–25 years; 33 novel). Most participants used augmentative and alternative communication (AAC) in early childhood (24/41). CAS was common (14/22). Performance varied widely across intellectual ability, social behaviour and expressive language skills, with participants ranging from within average through to the severely impaired range. Receptive language was significantly stronger than expressive language ability. Social motivation was a relative strength. In terms of a broader health phenotype, a quarter had one or more of: renal, urogenital, musculoskeletal, and cardiac malformations, vision impairment, ear infections and/or sleep disturbance. All had gross and fine motor impairments (41/41). Other conditions included mild-moderate intellectual disability (16/22) and autism (7/41). No genotype-phenotype correlations were found. Recognition of CAS, a rare speech disorder, is required to ensure appropriately targeted therapy. The high prevalence of speech and language impairment underscores the importance of tailored speech therapy, particularly early access to AAC supports.

*European Journal of Human Genetics*; https://doi.org/10.1038/s41431-022-01275-8

Read the article here

## Systematic review of speech and language across chromatin disorders

Our team recently published a systematic review of speech and language across chromatin disorders in *Neuroscience & Biobehavioural Reviews*. In the human body, DNA is wrapped around proteins, which form a

structure called chromatin. Chromatin is important for all body systems and regulates how our genes work. Our bodies also contain many genes that affect the structure and function of chromatin itself. Changes to these genes cause chromatin disorders, which can have many impacts on the body. Our review looked at 70 chromatin disorders and what the research says about their impact on speech and language. 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. Understanding these relationships helps understand more about the impact chromatin has on the brain and speech and language. This research is also important because chromatin disorders may be targeted in future gene therapies.

Read the article here

# 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.

## Acknowledgements

We would like to take this opportunity to thank the **Ainsworth 4 Foundation** and the **Channel Foundation** for their generous funding to support research into speech disorders. Their incredible support is playing an important role in advancing our research and helping us progress our work in the diagnosis, prognosis and management of childhood speech disorders and specifically stuttering.

Centre of Research Excellence - Translational Centre
for Speech Disorders

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