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

Latest News

Stuttering Study Launch
Recruitment for the Genetics of Stuttering study is well underway. Thank you to the 800+ individuals who have already participated! This has been a fantastic response, and participants have been true partners in helping us further refine our survey.

We are still recruiting to reach our goal of 3,000. Last week, we welcomed 12 year old Sian (picture below) and her Mum, Azelene to Melbourne, to spread the word about our study. Study investigators Professor Melanie Bahlo, Professor Angela Morgan and Dr Vicki Jackson were interviewed alongside Sian and her Mum on the radio and TV. Check out our website or Twitter if you missed out on the action, or would like to learn more about how to participate!

Contact us at:
www.geneticsofstutteringstudy.org.au
twitter.com/GenStuttering

2019 Past and Upcoming Events

April 12th: Round Table meeting - the future of speech genetics research with VIP guest Professor Simon Fisher

June 27th: Koolen de Vries Patient Advocacy Summit- Utah USA

July 11th: Apraxia Kids Conference- Pittsburgh USA

August 16th: Stuttering study launch
Welcome to our UK Branch
We have just expanded our program to commence recruitment in the UK, with our collaborators Dr Frédérique Liégeois and Dr Daisy Thompson-Lake. They have a busy summer and autumn planned visiting families in the UK!

Parents Advocating for Change
A group of parents in Tasmania have been advocating for changes to the Tasmanian Department of Education’s policy and procedures as they relate to external NDIS service providers access to State Schools to deliver therapy. The parents each have a child with a diagnosis of CAS supported by the NDIS. They argue that the State Department’s policy and procedures allow for school principals to make decisions refusing or restricting NDIS provider access, that result in the discrimination of children who require early intervention and/or have permanent disabilities. They have been informed the Department is considering the matter and that legal advice has been sought.

Website
We are currently in the process of developing a website that will provide families with information about genes that are associated with speech and language disorders. It will also provide information on trajectories of speech and language growth after diagnosis, evidence based interventions and ongoing research conducted within these areas. Stay tuned!

Current Studies
Phelan-McDermid Syndrome
Dr Amanda Brignell, postdoctoral research fellow in the CRE has been investigating the speech and language characteristics of children with Phelan-McDermid Syndrome. This study recruited children from four states across Australia. Recruitment for the study is now complete and the findings are being prepared for publication in a scientific journal. Two University of Melbourne Masters of Speech Pathology students have also been working on this project as part of their independent studies projects. Amanda will present the findings at the Phelan-McDermid Syndrome Foundation of Australia family conference in Sydney in January 2020.

Genetics of Speech Disorders: We are recruiting!
We have been recruiting children and families with childhood apraxia of speech or severe or persistent speech disorder. You do not need a family history of speech disorder for a condition to be genetically-based. Some gene changes occur for the first time, or ‘de novo’ in the current generation of children with apraxia or other speech disorders. We are currently still recruiting for the Genetics of Speech and Genetics of Stuttering studies. If you are interested, or know someone that may be interested please do not hesitate to contact us at geneticsofspeech@mcri.edu.au OR (03) 9936 6334.

We are also investigating the relationship between speech and music as part of a study conducted at the University of Melbourne. We are recruiting children with Childhood Apraxia of Speech, who have already taken part in the Genetics of Speech study. The additional music testing will be a 30-40 min enjoyable session that can be conducted at the child’s house or at MCRI. If you are interested in your child’s musical abilities, or know someone that may be interested please do not hesitate to contact Masters student Amie Willis akwillis@student.unimelb.edu.au OR 0437 352 987.
Awards, Conferences and Events

European Academy of Childhood Disability Conference
Ruth Braden (PhD student, see right) presented her recent paper entitled ‘Speech and language in bilateral perisylvian polymicrogyria (PMG): a systematic review’ at the European Academy of Childhood Disability Conference in Paris in May. Children with PMG have many small folds on the surface of their brain, which can be seen on MRI. PMG may have significant impacts on speech and language in some individuals, although the specific strengths and challenges of communication have not been well characterised. Ruth is helping to support families with PMG by providing the most comprehensive study of speech and language in individuals with PMG to date, hoping to improve diagnostic and prognostic information for families and clinicians.

Koolen de Vries Symposium
Professor Angela Morgan (below left) and Miya St John (PhD student, below centre) presented at the Koolen de Vries Symposium, Salt Lake City Utah in June 2019. They discussed their work on reverse phenotyping of Koolen de Vries and the current status and progression of a clinical trial using Karaoke to try and improve speech clarity called the Karaoke in the Kool Kids trial.

Apraxia Conference
Associate Professors Michael Hildebrand and Frédérique Liégeois, and Professor Angela Morgan presented at the Apraxia Kids Conference, Pittsburgh in July 2019. Apraxia Kids is a US based, international support group for children with Childhood Apraxia of Speech (CAS). Over 400 families attended the event. Our CRE team had the opportunity to provide families and health professionals with the latest evidence on diagnosis, genetic cause and brain imaging findings in CAS.

Centre of Research Excellence Day with Simon Fisher
The team was lucky enough to host our CRE collaborator CI Professor Simon Fisher from the Max Planck Institute for Psycholinguistics in Nijmegen, Netherlands. Simon is the co-founder of the FOXP2 gene, the first gene associated with speech apraxia in the literature. Simon is a core co-investigator on our CRE team. Simon and co-investigators attended a one day symposium to discuss current findings in, and future research direction for, the genetics of speech and language. Figure right shows CRE team members inc. Simon Fisher (mid front row)
Selected recent findings and publications

A novel neurodevelopmental condition was identified by our CRE team in collaboration with US colleagues. Speech impairment is a core feature of the condition, including CAS in at least one known case. If you would like to find out more information regarding this paper, please see the reference and link below;
Khan et al. (2019). Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine. https://www.nature.com/articles/s41436-019-0523-0?fbclid=IwAR16REwcuqLmGiX38RHV-5vqz8jL40dYU2GlR_Lwazy_aZ8nwgXyLOm3Q

Our work phenotyping communication in children with Koolen de Vries Syndrome has been translated into a Gene Reviews guideline document. Gene Reviews documents are clinical guidelines to support care and management of children. If you would like to find out more information regarding this paper, please see the reference and link below;

A paper has been published in collaboration with our team looking at cases studies of children with changes in the DDX3X gene, which has been linked to CAS. If you would like to find out more information regarding this paper, please see the reference and link below;