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Centre of Research Excellence in Speech and Language

Newsletter

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.

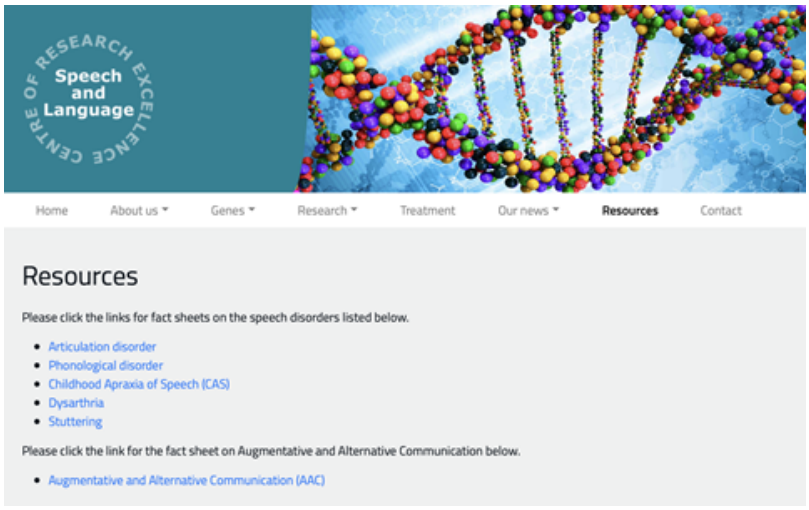
Our vision is to transform speech pathology practice by identifying, understanding and targeting the underlying causes of developmental speech and language disorders.

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 this research would not be possible.

Latest News



Visit our website!

New resourced added to Genetics of Speech Disorders website

We have recently added new fact sheet resources for articulation disorder, phonological disorder, childhood apraxia of speech, dysarthria, stuttering, and augmentative and alternative communication (AAC).

Our website also has other information about genes associated with speech and language disorders for clinicians and families. There is information on speech and language growth after diagnosis, evidence based interventions and ongoing research.

Childhood Apraxia of Speech: Pharmaceutical Trial

Our group is trialling a new treatment for children with childhood apraxia of speech (CAS). This treatment is the drug methylphenidate (MPH), commonly known as Ritalin. We want to find out whether MPH can help improve the speech and language outcomes for children aged 6-12 years with CAS.

MPH is approved as a treatment for ADHD in children. We want to find out if it is effective for children with CAS.

Learn more about the trial explained by Prof Angela Morgan and Clinical Trial Coordinator Elana Forbes in the video below.

We have already had some children finish the trial! Register your interest below or email speechtrial@mcri.edu.au.

Register here!

Social media

If you would like to keep up to date with what the CRE Speech and Language team is doing, please follow us on Twitter, at MCRI_SpeechLang. Alternatively, you can still find us on Facebook 'Genetics of Speech and Language Disorders'.

Awards, Conferences and Events

KAT6A and *KAT6B* Symposium

On March 23rd 2022, CRE PhD student, Miya St John presented at the 2nd KAT6A and KAT6B Virtual Symposium. The Symposium was an opportunity for researchers across the globe to collaborate and share their research regarding KAT6A and KAT6B syndromes. We thank the KAT6A Foundation founders, Emile and Natacha Najm, as well as research coordinator, Tanya Tripathi, for putting on such a wonderful and engaging event.

Rare Disease Day - SWAN Australia

On February 28th 2022, we celebrated Rare Disease Day, marking an important time to reflect on so many of the communities we work with. CRE PhD candidate, Miya St John, was invited to speak at one of this year's virtual events, hosted by SWAN Australia, Community Junction and Carer Gateway-Wellways Australia. We extend a special thanks to Heather Renton, Meg Salisbury and Jane Ellul for their invitation to be a part this fabulous event.

Holland Bloorview Pursuit Award

Dr Ruth Braden was awarded the international Hollen Bloorview Pursuit Award in June 2022. The award recognised Dr Braden for her for exemplary contributions and achievements to the field of childhood disability research.

Apraxia Kids Research Symposium

The Apraxia Kids Research symposium was held in Las Vegas over July 5-7th. Prof Angela Morgan was an invited keynote who opened the conference and spoke on the

history and current findings of the genetics of childhood apraxia of speech.

Selected recent findings and publications

DYRK1A Publication

Our team recently published a study on the speech and language features in 38 individuals with *DYRK1A* gene changes. The results showed that individuals with *DYRK1A* disorder have a range of speech and language features. Few individuals developed sufficient oral language skills to rely solely on talking to communicate. This meant that many individuals used Augmentative and Alternative Communication (AAC) to communicate. Social motivation was a relative strength of the cohort. These findings help to inform our understanding of speech and language in individuals with *DYRK1A* syndrome and will help us improve therapy approaches.

ARTICLE

Check for updates

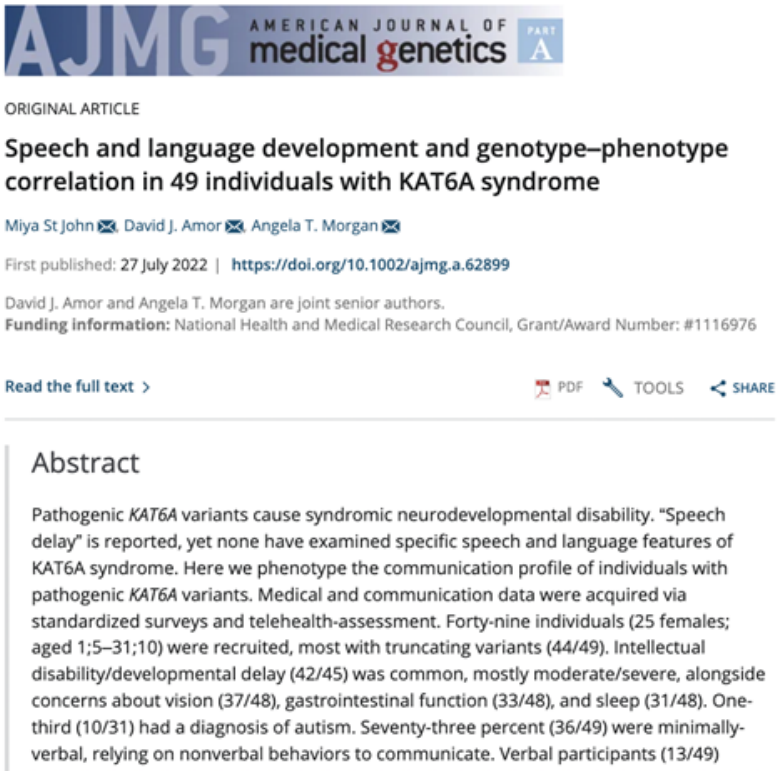
Social motivation a relative strength in DYRK1A syndrome on a background of significant speech and language impairments

Lottie D. Morison[1,7], Ruth O. Braden[1,7], David J. Amor[1,2,3], Amanda Brignell[1,4,5], Bregje W. M. van Bon[6,8] and Angela T. Morgan[1,2,3,8]✉

Speech and language impairments are commonly reported in *DYRK1A* syndrome. Yet, speech and language abilities have not been systematically examined in a prospective cohort study. Speech, language, social behaviour, feeding, and non-verbal communication skills were assessed using standardised tools. The broader health and medical phenotype was documented using caregiver questionnaires, interviews and confirmation with medical records. 38 individuals with *DYRK1A* syndrome (23 male, median age 8 years 3 months, range 1 year 7 months to 25 years) were recruited. Moderate to severe intellectual disability (ID), autism spectrum disorder (ASD), vision, motor and feeding impairments were common, alongside epilepsy in a third of cases. Speech and language was disordered in all participants. Many acquired some degree of verbal communication, yet few (8/38) developed sufficient oral language skills to rely solely on verbal communication. Speech was characterised by severe apraxia and dysarthria in verbal participants, resulting in markedly poor intelligibility. Those with limited verbal language (30/38) used a combination of sign and graphic augmentative and alternative communication (AAC) systems. Language skills were low across expressive, receptive, and written domains. Most had impaired social behaviours (25/29). Restricted and repetitive interests were most impaired, whilst social motivation was a relative strength. Few individuals with *DYRK1A* syndrome use verbal speech as their sole means of communication, and hence, all individuals need early access to tailored, graphic AAC systems to support their communication. For those who develop verbal speech, targeted therapy for apraxia and dysarthria should be considered to improve intelligibility and, consequently, communication autonomy.

European Journal of Human Genetics; https://doi.org/10.1038/s41431-022-01079-w

Read the article



AJMG AMERICAN JOURNAL OF medical genetics PART A

ORIGINAL ARTICLE

Speech and language development and genotype–phenotype correlation in 49 individuals with KAT6A syndrome

Miya St John ✉, David J. Amor ✉, Angela T. Morgan ✉

First published: 27 July 2022 | https://doi.org/10.1002/ajmg.a.62899

David J. Amor and Angela T. Morgan are joint senior authors.
Funding information: National Health and Medical Research Council, Grant/Award Number: #1116976

Read the full text › PDF TOOLS SHARE

Abstract

Pathogenic *KAT6A* variants cause syndromic neurodevelopmental disability. "Speech delay" is reported, yet none have examined specific speech and language features of KAT6A syndrome. Here we phenotype the communication profile of individuals with pathogenic *KAT6A* variants. Medical and communication data were acquired via standardized surveys and telehealth-assessment. Forty-nine individuals (25 females; aged 1;5–31;10) were recruited, most with truncating variants (44/49). Intellectual disability/developmental delay (42/45) was common, mostly moderate/severe, alongside concerns about vision (37/48), gastrointestinal function (33/48), and sleep (31/48). One-third (10/31) had a diagnosis of autism. Seventy-three percent (36/49) were minimally-verbal, relying on nonverbal behaviors to communicate. Verbal participants (13/49)

Read the article

KAT6A Publication

Research led by PhD student Miya St John has been published this month. The study of 49 individuals with *KAT6A* syndrome found that most individuals are minimally-verbal, with contributions from underlying

motor deficits and cognitive-linguistic impairment. Alternative/augmentative communication (AAC) approaches are required for many individuals into adult life. These findings highlight the importance of early uptake of tailored AAC options to support communication outcomes.

Current Studies

Speech Tracker

We launched a new online study called “Speech Tracker” at the end of last year. This study builds on our previous research showing that speech and language is a core developmental challenge for children with certain genetic conditions.The aim of Speech Tracker is to examine speech & 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 better understand prognoses for children and families, and will help us develop more targeted speech therapies.

To date we have have been recruiting children and adults with:
Koolen-de Vries Syndrome
***SETBP1* loss of function or missense variants**

So far

· over 80 families have registered interest across the two groups

· almost half of these families have completed the first stage of the study

· some families are now completing their 6 month follow-up

We encourage any families who have started their tasks to complete these as soon as possible, or contact the research team with any queries.

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

We aim to expand the use of Speech Tracker to further genetic conditions of interest in coming months, so we encourage you to watch this space for updates.

Email: speechtracker@mcri.edu.au

Register via the link below or scan the QR code.

Register here!

Stuttering Sub-phenotyping Study

PhD student, Sarah Horton, has recruited almost 100 participants for her Stuttering Sub-Group Study so far. This study aims to investigate sub-groups of people who stutter based on features such as family history of stuttering, speech characteristics, and other health and medical conditions. Participants complete online surveys and take part in a videoconference meeting to provide speech samples. We hope that identifying sub-groups of people who stutter with similar features will help us to better understand differences in stuttering such as why some people recover naturally and some don't, or why some people respond to treatments and others don't.

Sarah will soon launch a follow-up to this study where participants will record some further speech samples at different times. We hope that this will help us to better understand stuttering variability.

This project is being undertaken in conjunction with the international Genetics of Stuttering Study (https://www.geneticsofstutteringstudy.org.au/), which is currently recruiting participants who stutter or who have stuttered at some point in their lifetime.

Learn more!

Thank you!

Our team wishes to thank families and clinicians for their ongoing participation and support.

In the past year, we have expanded our interstate participation to reach more families across Australia.

A sincere thanks to all the families who have taken part, without your help this research would not be possible.

The Centre of Research Excellence in Speech and Language

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