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Newsletter

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

Latest Updates

SPEECH PATHOLOGY WEEK

Happy Speech Pathology Week (25-31 August)!

This year's theme is 'Communicate your way'.

Many of the people we work with use different ways to communicate. Some use speech, some use sign language, some use alternative devices or eye gaze – to name a few.

We would like to thank all the wonderful kids and their families who have participated in our research and who have visited our Speech Apraxia and Genetics Clinic at the Royal Children's Hospital. Since opening a few years ago, our clinic has already supported 250 children and their families. We are honoured to be part of their journey, while we work and learn together.

Please look out for our Instagram and Facebook this week to find out how our research is involved in some of these unique ways of communicating.

STUTTERING SUB-GROUP STUDY

Researchers from the Speech and Language team are investigating subgroups of people aged 5 years and older, who stutter (past or present), based on family history, speech characteristics, personal experience of stuttering, and other health and medical conditions.

Identifying subgroups of people who stutter with similar features will help us to better understand differences in stuttering such as why some people resolve naturally and some don't, or why some people respond to speech pathology treatments and others don't.

Participation in the study is free and easy. Participants are expected to complete online surveys (around 45 minutes) and attend an online Zoom session with a speech pathologist from our team (around 45 minutes). No formal diagnosis of stuttering is needed.

Please note recruitment for this study will cease in October.

Learn more about this study

GLUT-1 DEFICIENCY SYNDROME: speech and language STUDY

Researchers at the University of Melbourne in collaboration with Redenlab and our Speech and Language team at MCRI, are running the largest global study of communication in GLUT-1 Deficiency Syndrome.

The study is investigating speech and language outcomes in individuals aged 6 months to adulthood with GLUT-1 Deficiency Syndrome. By improving our understanding of speech and language in this group, we hope to develop more targeted strategies and therefore improve prognoses. The findings will also contribute to the development of objective measures of communication for use in clinical trials.

Participation in the study is free and involves a combination of online surveys, online speech tasks and home recordings. Interested families are welcome to register their interest here.

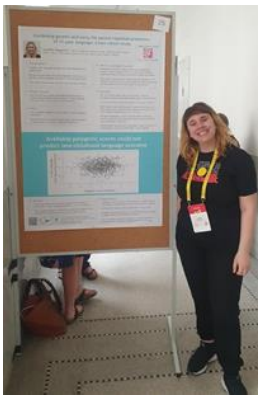
We would like to thank the Glut1 Deficiency Foundation in the USA for their generous funding and support of this project.

Awards, Conferences & Events

XVIth INTERNATIONAL CONGRESS FOR THE STUDY OF CHILD LANGUAGE (IASCL)

Speech & Language team member Lottie Gasparini presented a poster on genetic and early life parent-reported language predictors at the IASCL in Prague, Czech Republic (15-19 July).

Her poster titled ‘Combining genetic and early life parent-reported predictors of 11-year language: A two-cohort study’ and supporting information can be accessed here. So far, available genetic measures (polygenic scores) could not predict late-childhood language outcome. However, the analysis is ongoing and updates can be found here.



Lottie Gasparini's poster presentation

APRAXIA KIDS NATIONAL CONFERENCE

CRE Director Prof Angela Morgan was invited to co-present with Dr Ruth Stoeckel (Mayo Clinic emeritus SLP) to discuss 'Genetics of Childhood Apraxia of Speech' at the Apraxia Kids National Conference in Pittsburgh, USA (11-13 July).

In the past three years, genetic analysis has revealed over 30 causative genes implicated from 122 children across 3 cohorts. These results show that one in three children have a genetic variant that explains their Childhood Apraxia of Speech (CAS). Many of the new genes for CAS are associated with previously described conditions that include intellectual disability, autism and epilepsy. Clinical genetic testing should be implemented for those with CAS, in order to parallel many other neurodevelopmental disorders where this testing is already standard of care.

We would like to thank Apraxia Kids for their invitation to present at this annual conference which attracts an international audience.

Audience members at the Apraxia Conference.

INTERNATIONAL RESEARCH CONFERENCE ON FOXP1 SYNDROME

CRE team member Dr Miya St John presented at the International Research Conference on FOXP1 Syndrome on 8-9 July at Duke University (North Carolina, USA). The International FOXP1 Foundation partnered with the Duke University, School of Medicine to host the event which provided an opportunity for families and individuals with FOXP1 syndrome from all over the world to come together with researchers to accelerate the development of therapies to help FOXP1 families.

Miya spoke about speech and language in FOXP1 syndrome. She presented the work of the team (including CRE investigators Dr Ruth Braden and Olivia van Reyk) of our FOXP1 Speech Tracker study and how this will be crucial work for future drug therapy trials.

We would like to thank the organising committee and the FOXP1 Foundation board for inviting us to present, and for hosting such a successful conference.

Dr Miya St John (bottom row – second from the right) was one of the presenters.

NCCR SYMPOSIUM

The National Centre of Competence in Research (NCCR) Evolving Language organised a Symposium 'From genes to communication' at the University of Geneva, Switzerland (10-11 June). The event brought together leading researchers in the field of genetics to discuss topics related to genetics of language, including genetics of language-related disorders, genetics of animal communication, (epi-)genetics of linguistically-relevant computation and memory.

Prof Angela Morgan was an invited speaker who presented on 'Genetic bases of child speech and language disorders'.

We would particularly like to thank NCCR directors Daphné Bavelier and Nina Kazanina (Geneva), Balthasar Bickel (Zurich) and Klaus Zuberbühler (Neuchatel) for the invitation to present.

Prof Morgan speaks about the genetic bases of child speech & language disorders.

CURE CLCN4 SCIENTIFIC CONFERENCE

The Cure CLCN4 annual (hybrid) conference was held in London, UK (7-8 June). Researchers and clinicians working on *CLCN4*-related condition and related areas were able to network, and engage in discussions about the latest research and advancements in the field.

CRE team member and student Alecka Garrett presented preliminary results from our international research study looking into speech and language outcomes in individuals (6 months to adulthood) with *CLCN4*-related neurodevelopmental disorder.

Recruitment for this research project has now been finalised. We would like to sincerely thank all the families who participated in our research. Our team is currently preparing a publication and we look forward to sharing the final results of this study.

We would like to thank Cure CLCN4 for their invitation to speak at this wonderful event.

Alecka Garrett presenting at the conference.

SYMPOSIUM 'WHAT'S NEW SINCE *FOXP2*: NEW DEVELOPMENTS IN SPEECH & LANGUAGE NEUROBIOLOGY'

Our NHMRC Centre of Research Excellence - Translational Centre for Speech Disorders was delighted to co-host their Symposium ‘What’s new since *FOXP2*: new developments in speech and language neurobiology' together with University College London (UCL) Great Ormond Street Institute of Child Health. This one-day in-person event held on 7 June 2024 (London), celebrated over 20 years of research since the ground-breaking identification of *FOXP2* as a gene critically involved in speech and language development. International experts, including Prof Vargha-Khadem (UCL) and Prof Fisher (Max Planck Institute for Psycholinguistics), presented on the most recent genetic and neuroimaging discoveries in the field of speech and language disorders. The event ended with a panel discussion on implications for clinical practice and future research directions.

A big thank you to all the wonderful speakers and

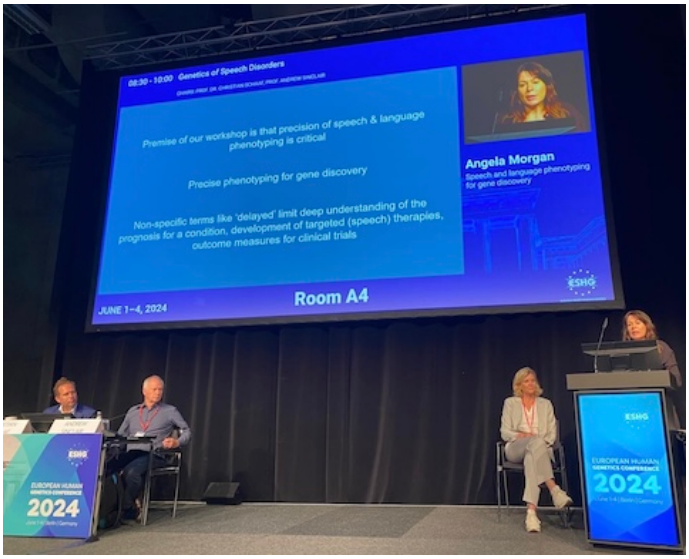
delegates who attended our CRE Symposium. For the full program and list of speakers, please click here.

Speakers & some delegates at our Symposium.

EUROPEAN HUMAN GENETICS CONFERENCE

CRE Director Prof Angela Morgan was an invited education session speaker at the European Human Genetics Conference in Berlin, Germany (1-4 June). She co-presented with our close collaborator Prof Tjitske Kleefstra of the Erasmus Medical Centre and Sophia Children's Hospital in the Netherlands. Angela and Tjitske discussed 'Genetics bases of speech and language in the neurodevelopmental disorders landscape.'

The conference was attended by more than 5,500 delegates onsite and 1,000 online participants from over 90 countries.



Prof Morgan (far right) co-presenting with Prof Kleefstra (seated).

SANFILIPPO CHILDREN'S FOUNDATION ANNUAL SCIENTIFIC SYMPOSIUM

The Sanfilippo Children's Foundation hosted their annual Solving Sanfilippo Symposium in collaboration with the

Laboratory for Human Neurobiology, South Australian Health and Medical Research Institute in Adelaide on 31 May-1 June. Over 85 researchers, clinicians and parents attended to discuss research on the childhood dementia, Sanfilippo syndrome (mucopolysaccharidosis type III).

CRE PhD candidate Lottie Morison presented on 'Planning for the future for children with genetic conditions that cause a loss of communication skills'.

We would like to sincerely thank the Sanfilippo Children's Foundation for their invitation to present and be part of this important Symposium.

Lottie Morison was one of the presenters.

Selected recent publications

Speech and language classification in the human phenotype ontology

CRE Director Prof Angela Morgan and collaborators recently published an article in the European Journal of Medical Genetics: *Speech and language classification in the human phenotype ontology*.

The Human Phenotype Ontology (HPO), introduced in 2008, provides a standardised vocabulary of phenotypic abnormalities of human disease, with each term describing a specific feature, such as 'language impairment'. The HPO contains over 16,000 terms and over 156,000 annotations to hereditary diseases. Its detailed descriptions of clinical abnormalities and computable disease definitions is used by researchers, clinicians, informaticians and electronic health record systems around the world for deep phenotyping in the field of rare disease.

In this paper, the authors discuss the need to refine classification of speech and language phenotype terms in the HPO in rare diseases. It describes the current speech and language phenotyping in the HPO and proposes a

revision and simplification of the speech and language hierarchy and sub-phenotypes.

Collaborators on the HPO project.

Read more about this article

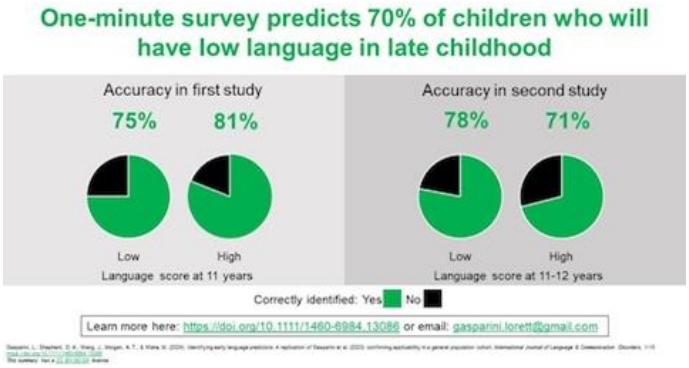
Identifying early language predictors: A replication of Gasparini et al. (2023) confirming applicability in a general population cohort

Speech & Language team member Lottie Gasparini together with collaborators including CRE Prof Angela Morgan, recently published in the International Journal of Language & Communication Disorders: *Identifying early language predictors: A replication of Gasparini et al. (2023) confirming applicability in a general population cohort*.

There are no robust variables able to predict which children will experience lasting language disorder with its far reaching negative impacts.

In this study, the authors aimed to examine the ability of variables collected at 2 years for predicting which children have language disorder at 11 years of age. An exciting approach was taken, using new machine learning methods, applied to one of Australia's largest birth cohorts of children, the Longitudinal Study of Australian Children (LSAC). The results confirmed the authors earlier findings from a previous large scale population-based cohort study, that 6 specific variables (taking only one-minute to collect) can predict 70% of children who will have low language in late childhood. This is an exciting step forward in the field for developing a low-resource and time-efficient recruitment tool for early language intervention studies. This could lead to improved clinical service provision for

young children likely to have persisting language difficulties.



Read more about this article

A plain language summary is 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.

Acknowledgement

We would like to take this opportunity to thank the Ainsworth 4 Foundation for their generous funding to support research into the genetics of 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.

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