minds for minds
Unlocking Autism Together

Newsletter December 2023

Kia ora from Minds for Minds and Te Ara Hāro !

As we near the end of the year, we hope you and your whānau have an enjoyable and relaxing summer break. In this newsletter, we summarise some research updates in Aotearoa and abroad. For those involved in the genetics project – if you have not been emailed regarding results or next steps, please email arnnz@auckland.ac.nz for an update. We are busy working toward transitioning our research into the clinical setting in Aotearoa to support early and accurate genetic diagnosis for those who would like it. Warm wishes to you and your whānau over the holiday season.

Research Report

Autistic co-led autism research priorities for Aotearoa New Zealand: A partnership with Autistic people, families and whānau, service and support providers, and researchers

Currently there are no priorities or strategy for autistic research in Aotearoa New Zealand. A group of researchers and advocates came together to establish a set of national priorities for autism research.

The report was made in partnership with autistic adults and other non-autistic members of the community, such as parents/carers, education and health practitioners, and researchers.

The research group consulted with the Autistic and autism community through focus groups and online surveys, and interviews with young autistic people. They asked questions about what future research should focus on and how it can help autistic people lead the lives they want to live.

The researchers identified 13 autism research priorities within four main areas. These research areas include establishing the needs of Autistic people, improving available supports for Autistic people, understanding of life from the point of view of Autistic people, and how we can improve our structures and environments to be inclusive of Autistic people.

Read the full report here.

New Projects

PhD student Suzanne Musgrave is working with Dr Jessie Jacobsen at the University of Auckland to understand the genetics of neurodevelopmental conditions.

Many neurodevelopmental conditions are caused by genetic variants, and identifying these variants can be incredibly useful as it can provide a more appropriate and descriptive diagnosis which can alter what kind of supports the individual receives.

However, identifying causative genetic variants comes with its own challenges. Limitations in standard sequencing technology make identifying large structural changes to the genome difficult, so they are often missed or not investigated in typical genetic analyses. Also, when translating genetic variants to clinicians and families, we need to be very confident that potential new genes or variants are functionally evaluated before they are returned.

Suzanne's PhD project will involve analysing the genomes of individuals with neurodevelopmental conditions, looking for large structural changes, and reanalysing genomes where a causative genetic variant was not found previously. The project will also involve assessing the biological consequences of new genes and variants in order to understand their role in neurodevelopment.

Masters student Sarah Jardine has been working with Hiran Thabrew at Te Ara Hāro to improve care for gender diverse and autistic young people.

Current clinical guidelines address one, but not both issues. A survey of 152 New Zealand clinicians found that clinicians are more likely to investigate gender diversity when young people present with autism than to investigate autism when they present with gender diversity.

These results were consistent both regionally and between public and private services. Bidirectional screening was more common among clinicians who worked in both autism and gender-related clinical settings.

Improvement of practice guidelines is urgently needed to improve care for this group.

New Zealand Publications

Community perspectives on the appropriateness and importance of support goals for young autistic children.

There has been limited research on the perspectives people in the autism community have about support goals for young autistic children.

Researchers at Victoria University of Wellington and their colleagues aimed to address this gap by surveying autistic adults, parents of autistic children, and clinical professionals living in Aotearoa New Zealand and Australia. They asked questions about the appropriateness and the importance of common support goals for young autistic children.

All participant groups rated goals related to the adult supporting the child, replacement of harmful behaviours, and improving child quality of life as the highest priorities, and ranked goals around play and academic skills, and autism characteristics as the lowest priority. While the groups agreed on the general order of priority, autistic adults were more likely to rate goals related to play skills and autism characteristics as inappropriate compared to parents or professionals.

More work needs to be done to understand the reasons why participants rated certain goals as lower or higher than others, and to ensure that the goals of clinical autism research aligns with these priorities.

Article Authors: Waddington, H., Minnell, H., Patrick, L., van Der Meer, L., Monk, R., Woods, L., & Whitehouse, A. J. Read the article here.

Identifying neurodevelopmental disabilities from nationalised preschool health check.

The Before School Check (B4SC) is a nationwide pre-school health assessment offered to all 4 year olds. It includes a general physical health screen and standardised questionnaires about behaviour and development.

Researchers at the University of Auckland and the University of Otago investigated whether combining screening measures in the B4SC could improve the early detection of neurodevelopmental conditions (NDCs).

Through looking at linked health data, the researchers identified an NDC in almost 11,000 4-5 year olds, 3.8% of their cohort. They found that combining scores from the different questionnaires in the B4SC was better at predicting which children had NDCs than any single measure. Including information about vision and biological sex alongside the questionnaires further improved the prediction. They suggest using these combined measures to improve early detection of children with NDCs in Aotearoa.

Article Authors: Mujoo, H., Bowden, N., Thabrew, H., Kokaua, J., Audas, R., & Taylor, B. Read the article here.

National SCIENCE Challenges

A BETTER START

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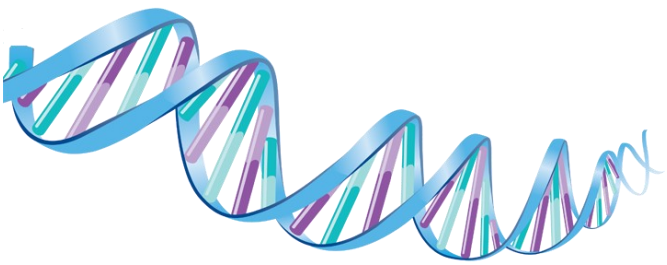
International Autism Research

Molecular diagnosis of 405 individuals with autism.

A group of researchers in Japan performed genetic analysis of 405 autistic individuals and their families, sequencing the coding regions of their genomes to find genetic variants that would lead to a molecular diagnosis. The researchers identified a causative variant in 66 participants (16.3%). They found that female participants were significantly more likely to have a causative genetic variant, reflecting a common trend in this research.

Interestingly they found the molecular diagnostic rate to be much lower in families with multiple autistic siblings, than in families with only one autistic child. In fact, of the three genetic variants they found in these families, only one was shared by the autistic siblings. This indicates that the genetics in families with multiple autistic individuals is more complicated than families with just one autistic individual.

The researchers also looked at the rate of genetic diagnosis over time. Based on the increasing knowledge of genes involved in autism, they estimated that the rate of genetic diagnosis increased 0.63% per year in their cohort, and that only 7.2% of their cohort could have been diagnosed before 2010. This highlights how important it is to reanalyse genetic sequences of autistic individuals who did not receive a genetic diagnosis.



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