



Quarterly Newsletter Of Autism Research – December 2017

Welcome

It's our final newsletter of the year and the Christmas season seems to have come around faster than usual!

End of years are a time to take stock of achievements and prepare for the coming year. Despite some challenges in 2017, the biggest being funding to carry out the sequencing for those registered on our database, we've had some amazing successes from the research team, thanks to the collaborative efforts of scientists, researchers and clinicians (see below).

Thank you to all volunteers and supporters; we do appreciate all your interest in the research and your support of the Trust. As Christmas looms, on behalf of the Minds for Minds Trust we wish you a very happy Christmas and safe New Year and hope that you all have the opportunity to spend time with your family and friends over the festive season. And if you're looking for a gift replacement, go to our website and donate to the research at www.mindsforminds.org.nz/donate

Best Wishes everyone,
Josephine Wallis



Josephine Wallis
Minds for Minds Chairperson

Minds for Minds Research

Below are summaries from two papers published by the Minds for Minds genetics team in the last month – don't let the scientific jargon in the titles put you off!

Compound Heterozygous Inheritance Of Mutations In Coenzyme Q8A results In Autosomal Recessive Cerebellar Ataxia And Coenzyme Q10 Deficiency In A Female Sib-pair

JIMD Reports:

Thanks to the collaborative effort with some of our stellar New Zealand clinicians, the genetics team recently published results of a family, which have two girls with ataxia (loss of coordination of gait).

After identifying the genetic cause in the two girls (a DNA variation in a gene called coenzyme Q8A), it quite brilliantly led to the family trialing an approved treatment (oral Coenzyme Q10). After a year of treatment, the girls' movements have improved along with their energy levels and classroom performance.

The genetics group is very grateful to the researchers and clinicians who helped them reach such a great outcome for this family.

The article can be found here:
https://link.springer.com/chapter/10.1007%2F8904_2017_73

Compound Heterozygous SLC19A3 Mutations Further Refine The Critical Promoter Region For Biotin-Thiamine responsive Basal Ganglia Disease

Molecular Case Studies: Results from another family were recently published by the genetics team in partnership with New Zealand clinicians. In this family, two siblings presented with neurological deterioration, seizures and gait ataxia. The genetic cause was determined to be two genetic variants in a gene called solute carrier family 19, member 3 (SLC19A3), including one deletion (the focus of PhD student, Whitney Whitford's research). Unfortunately, the elder sibling passed away before this variant was found, however the surviving child has responded well to treatment with biotin and thiamine (B-vitamins important for brain function). Following 10 months of treatment, the girl is running independently, eating a full diet and communication and cognition is improving.

The article can be found here:
<http://molecularcasestudies.cshlp.org/content/3/6/a001909.full>

Without the involvement of families we would not have these successes, so thank you to all those enrolled in the research. We are continuing to sequence as many participants as funding allows, apologies if we have not been in touch to collect a saliva sample yet. We hope to collect and sequence many more families in 2018.

Ingenio – Auckland University Alumni Magazine

In 2010 a young neuroscientist was named The University of Auckland's Young Alumna of the Year. That young doctor, Jessie Jacobsen, is one of the lead researchers in our Minds for Minds team. She recently spoke about the team's genetics work with the University of Auckland's Ingenio magazine.



Photo credit: Charlotte Johnson

The article can be found here:
<http://www.ingenio-magazine.com/cracking-the-autism-code/>

Minds for Minds Masters Student's Update



Chris and Lydia, our two bright young Masters students we introduced last newsletter, have been working with our genetics team. They are due to hand in their theses in February 2018; we wish them all the best for their write up. Here is a summary of what they have found out.

Lydia has looked at five families, each where the child has a rare, clinically undiagnosed, neurodevelopmental disorder. She found causative variants in three children, all of which have been validated in our lab. Two have also been independently validated by a clinician and are in the process of being returned to the family. The causative variants were found in genes crucial to the rapid neuronal expansion process that occurs during development.

Chris found causative variations in four of the five cases he looked at. These variations occur in a variety of genes including those involved in brain development and in cell communication. Two of these variations have been clinically validated and are being returned to the families whilst the others require further analysis to fully characterise their role and function.



Spring Week On Auckland University Campus

It's been a busy year for our Dr Jessie Jacobsen who recently had the privilege of speaking at the University of Auckland's Spring Week on Campus – a public programme run by the University. The week opens up a window for enthusiastic adult students with a love of learning, into some of the world-class research being carried out at the University. Sessions covered child health, stroke recovery, contemporary New Zealand artists in Berlin and of course our very own Minds for Minds genetic research. The Trust takes every opportunity to share its research with the wider community. If you are keen to attend public lectures like this, keep an eye out on the university website early next year to find out what might appeal to you. The next "week" will be in winter 2018 (Winter Week on campus).

Minds for Minds - Health Research Council Grant

A project grant from the Health Research Council was recently awarded to Associate Professor Johanna Montgomery (Physiology). Johanna is a member of the wider Minds for Minds research



team. This project will investigate a dietary zinc link in Autism Spectrum Disorders. It is known that zinc can regulate brain cell communication and low zinc levels have been identified in people with ASD. In this project, Prof Montgomery will examine whether maternal zinc levels can influence the development of ASD-related behaviours in animal models of ASD. In collaboration with international experts, she will also determine whether dietary zinc can improve ASD-related behaviours in multiple animal models that express ASD-related genetic changes. In addition, together with Associate Professor Mike Taylor, they will investigate how dietary zinc alters the gut microbiome and whether this can contribute to positive effects of dietary zinc. Once we have this knowledge we aim to then advance to human studies of dietary zinc in ASD. We look forward to reporting the results.

Minds for Minds T-Shirts For Sale



Josephine Wallis & son Michael with t-shirts

Ken Griffen is a young New Zealand artist, based in New York and has had sold out exhibitions locally. He has gifted an exclusive limited-edition design for 250 Minds for Minds t-shirts to raise funds for the autism research project. Ken was originally Creative Director for Huffer brand. As one commentator said of his art "I have not been to an exhibition or come across a new artist that has made me this excited in a very long time." to see his works visit www.kengriffen.com. There were only 250 t-shirts produced, but there are still some available in a limited size range with his signature on the sleeve. Plenty of children's sizes in white only. These will make great Christmas presents but will need to be ordered by 17 December. After that, orders received will be dispatched from 10 January. All t-shirts are \$50. Colours are black on white and white on black. There are a number of sizes in men's, women's and youth/kids. All proceeds go directly to the Minds for Minds autism research project.

To order a t-shirt click here:

www.mindsforminds.org.nz/shop

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Help us continue our research

www.mindsforminds.org.nz/donate

Speaking Circuit

Remember if you would like any one of our scientific team to speak to your group or conference about Minds for Minds and its research, please email us at info@mindsforminds.org.nz