

Genetic diagnosis of Autism Spectrum Disorder in New Zealanders

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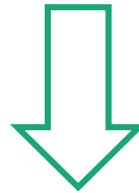


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Research question

How can we improve genetic diagnosis of Autism Spectrum Disorder in New Zealand to provide these children with the best start in life?



Objective

Develop materials and protocols for early and accurate gene-targeted genetic diagnosis of ASD in NZ

Why does it matter?

- Prevalence estimated to exceed 1% of our population (1 in 68 and 1 in 42 boys)
- Extrapolation of lifetime healthcare costs = NZ\$2.2bn p.a.
- ASD can be a significant challenge for families, particularly accessing appropriate social and educational support
- It is evident there is a strong genetic component - 83%¹
- ‘High confidence’ genes responsible for ~10-30% of cases²
– not routinely screened in the clinic in NZ

¹ Sandin, S. et al. (2017) The heritability of autism spectrum disorder. *Jama* **318**, 1182-1184

² Vorstman, J. A., et al. (2017) Autism genetics: opportunities and challenges for clinical translation. *Nat Rev Genet.* **18**, 362-376

Our plan and current status

- Enrol 300 participants with ASD in NZ
 - *Have >1600 participants interested*
- Collect samples (saliva or blood)
 - *Extracted DNA from first cohort (100)*
- Develop/optimize DNA sequencing + bioinformatics protocols
 - *Refined gene list to 95 'high confidence' genes*
 - *Currently contracting to develop customised ASD panel*
- Assess 300 participants for causative variations in 'high confidence' ASD genes
 - *Next step*
- Return findings to participants (where appropriate)
- Clinical certification of materials/protocols

Ultimate hopes

- An accurate genetic diagnosis will improve long term health outcomes for New Zealanders living with ASD
- The identification of genetic variants particular to New Zealand's unique population mix will improve clinical interpretation
- The discovery of genetic variants will lead to improved understanding of relevant biological pathways associated with ASD and overlapping conditions

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Community groups

Autism NZ

Children's Autism Foundation

Autism Intervention Trust

NZORD



minds for minds

Unlocking Autism Together

www.mindsforminds.org.nz