

Genetic Analysis of Autism Spectrum and Associated Neurodevelopmental Disorders



What is the purpose of the research?

Our genomics study will identify and characterize genes that confer risk to autism spectrum and related neurodevelopmental disorders. With this knowledge there will be an enhanced biological understanding of ASD, which we believe will facilitate earlier diagnosis and suggest opportunities for therapeutic interventions.

What does the research study involve?

- You will be requested to provide clinical information on your child and family
- A small blood sample will be requested from the participant(s) in order to extract DNA (genetic material)

What will the DNA sample be used for?

Microarray screening:

- To look at the chromosomes for missing or extra genetic material called a copy number variant (CNV) with the focus on the possible identification of genetic variants contributing to ASD in your family.

Genome Sequencing:

- To look for smaller changes in genetic material with the focus on the possible identification of the genetic factors contributing to ASD in your family.

What are the potential benefits to our family?

- The high-resolution microarrays used in this research study capture much more information than those currently offered at a diagnostic laboratory.
- Genome sequencing is not currently offered as a hospital-based clinical service and as far as we know can only be freely assessed through research studies.

Who can participate?

- Individuals having a formal diagnosis of Autism Spectrum Disorder, including Aspergers Syndrome, Autism and PDD-NOS.

For more information please contact:

Susan Day Fragiadakis at Holland Bloorview: 416-425-6220 ext. 6518
Email: sfragiadakis@hollandbloorview.ca