

GENETIC TESTING FOR NEURODEVELOPMENTAL DISORDERS (NDDs)

How to arrange genetic testing for NDDs in Manitoba



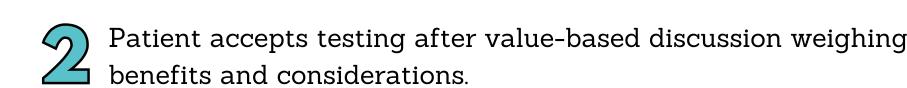
For clinicians in Manitoba first-tier genetic testing cannot be ordered by primary care clinicians.

Referral to Genetics or Developmental Pediatrician for assessment is required.





Patient meets eligibility criteria.





• Our patient/family information handout could be used to facilitate discussion.



• Genetic test results can take 4-8 weeks, depending on the laboratory. If results could affect an ongoing pregnancy you can request that they are expedited.



• Individuals with another

unexplained GDD and/or

Who is offered genetic testing?

All autistic individuals

All individuals with

NDD plus other health condition

> GDD- Global Developmental Delay ID - Intellectual Disability

Download and complete the Requisition for your regional/ provincial laboratory to order Chromosomal Microarray (CMA).

- Complete with as much information as possible, checking all applicable boxes, and include available family history (even if non-contributory).
- The laboratory scientists will use all available clinical information to interpret results.

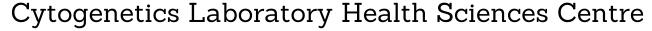
Determine if Fragile X Syndrome (FXS) testing should also be ordered and coordinate the blood draws (FMR1 gene). This test would be important in an individual with a diagnosis of autism, GDD and/or ID AND one or more of the following features:

- Macro-orchidism
- Macrocephaly, maybe mild or appear disproportionately large relative to body stature
- Large or prominent ears, long or narrow face, tall forehead, high arched palate, prominent jaw
- Soft velvety hands, redundant skin on dorsum of hands, hyperextensible joints, pes planus, mitral valve prolapse
- Maternal relatives with a diagnosis of autism, GDD and/or ID
- Maternal relatives assigned female at birth with premature menopause or ovarian insufficiency
- Maternal relatives with adult-onset tremor, ataxia, or parkinsonism
- Maternal relatives with a known diagnosis of FXS or FXS related condition



Requisitions and Laboratory contact information

CMA is a cytogenetic test and FXS is a molecular genetic test. (Links accessed July 2024)



- Website | Tel: 204-787-2489
- CMA Requisition form

Molecular Diagnostic Laboratory Health Sciences Centre

- <u>Website</u> | Tel: 204-787-1024
- FXS Requisition form





If you have questions, don't hesitate to call or email the Laboratory. There is often a genetic counsellor available to support your ordering.

