GENETIC TESTING FOR NEURODEVELOPMENTAL DISORDERS (NDDs)

How to arrange genetic testing for NDDs in Newfoundland and Labrador

GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

For clinicians in Newfoundland and Labrador first-tier genetic testing cannot be ordered by primary care clinicians.
Test ordering is restricted to Geneticists and Pediatricians.

Genetics referral information here

More on testing in NFLD here

Eastern Health

1 Patient meets eligibility criteria.

2 Patient accepts testing after value-based discussion weighing benefits and considerations.

- 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.

Who is offered genetic testing?

- All autistic individuals
- All individuals with unexplained GDD and/or ID
- Individuals with another NDD plus other health condition

GDD- Global Developmental Delay
ID - Intellectual Disability

3a 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.

b 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)

- Provincial Medical Genetics
- Eastern Health Laboratory Medicine
  - Ordering restrictions have been lifted to allow *pediatricians only* to request Fragile X A.FMR1 and Fragile X E. FMR2 when investigating children with global developmental delay and intellectual disability.
  - The proper requisitions must be *fully* completed and submitted to the Newfoundland and Labrador Health Services Genetics Laboratory. Test requests and specimens will be accepted only from pediatricians. Specimen collection instructions on the requisitions must be strictly adhered to.

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

Clinical presentation is suspicious for a genetic condition (e.g. Rett syndrome)

→ Refer for genetic assessment for consideration of targeted testing

- Autism
- Global developmental delay
- Intellectual disability
- NDD plus additional features

NDD - Neurodevelopmental disorder

→ Offer and arrange first-tier genetic testing

- chromosomal microarray
- Fragile X syndrome — ***IF*** additional related features also present

Results

⊕ Positive result → Refer for genetic assessment, counselling, follow-up

⊖ ? Negative or unclear result

Indication for testing was isolated autism

No → Refer for genetic assessment and consideration of second-tier genetic testing

Yes → No further genetic testing/assessment

Reassess and re-refer as criteria expand and technology evolves

- Other NDD and no additional features or family history

→ Genetic assessment not indicated

First-tier genetic testing can be ordered by any physician in:

- British Columbia
- Alberta
- Ontario
- Québec
- Maritimes

Referral for genetic assessment may require that first-tier genetic testing be completed first. Check your local Clinic's criteria.

First-tier genetic testing can only be ordered by a Geneticist or Specialist (e.g. developmental pediatrician) in:

- Saskatchewan
- Manitoba
- Newfoundland and Labrador (geneticist only)

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