GENETIC TESTING FOR NEURODEVELOPMENTAL DISORDERS (NDDs)

How to arrange genetic testing for NDDs in Saskatchewan

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

For clinicians in Saskatchewan first-tier genetic testing cannot be ordered by primary care clinicians.
Referral to Genetics, Developmental Pediatrician or other appropriate specialist for assessment is required.

Genetics referral information here

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)

Genetic Resource Centre – Roy Romanow Provincial Laboratory

Requests can be made directly to the Genetic Resource Centre directly for requisitions.
Tel: 306-655-6450
Email: grc@saskhealthauthority.ca
FXS (and other) - Requisition form

Ordering of CMA requires review by the Genetic Resource Centre as it is an out-of-province test. Contact the GRC here grc@saskhealthauthority.ca

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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