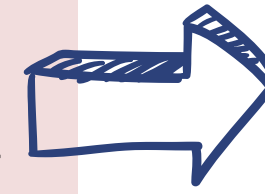




For clinicians in Saskatchewan first-tier genetic testing **cannot** be ordered by primary care clinicians. Referral to Genetics, Developmental Pediatrician or other appropriate specialist for assessment is required.



[Genetics referral information here](#)

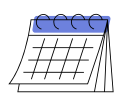


**1** Patient meets eligibility criteria.

**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](mailto:grc@saskhealthauthority.ca)

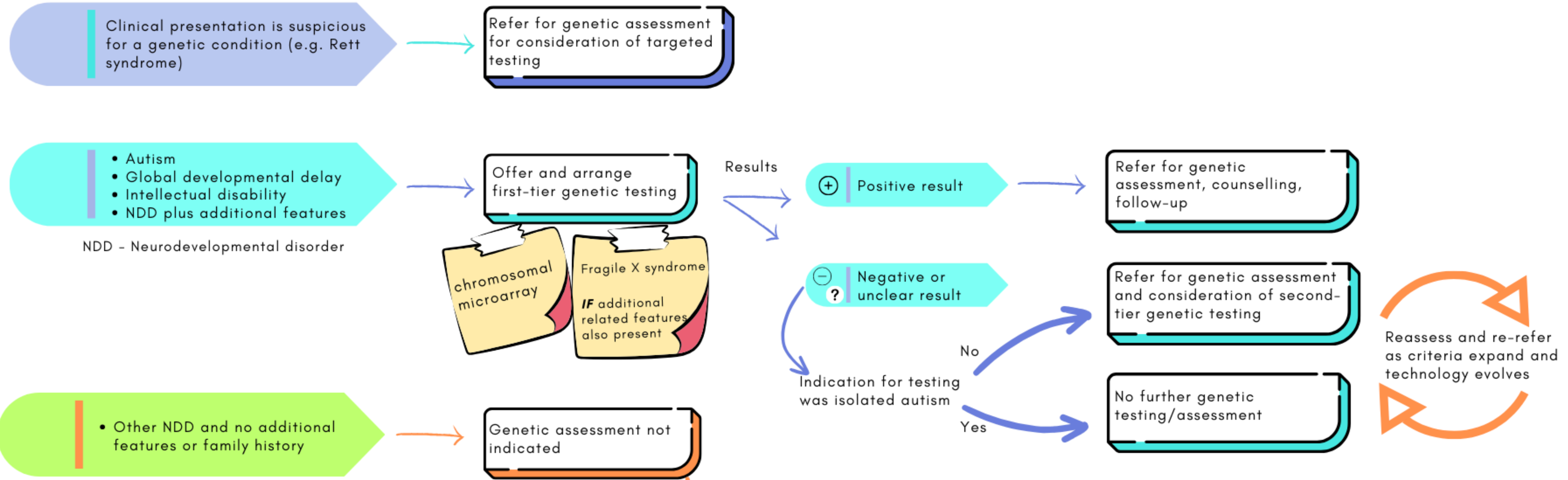
FXS (and other) - [Requisition form](#)



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

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](mailto:grc@saskhealthauthority.ca)



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)

(Links accessed July 2024)