

Who can order first-tier genetic testing?

Any British Columbia physician can order first-tier testing.

When to order first-tier genetic testing?

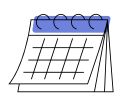
Some Genetics Clinics require that first-tier genetic testing be completed **prior to** referral. This is also the recommendation of the Canadian College of Medical Geneticists. Others permit referral while genetic testing is in progress, or may accept a referral without genetic testing. Check out the referral criteria on the Clinic website or call your local genetics specialist.

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)

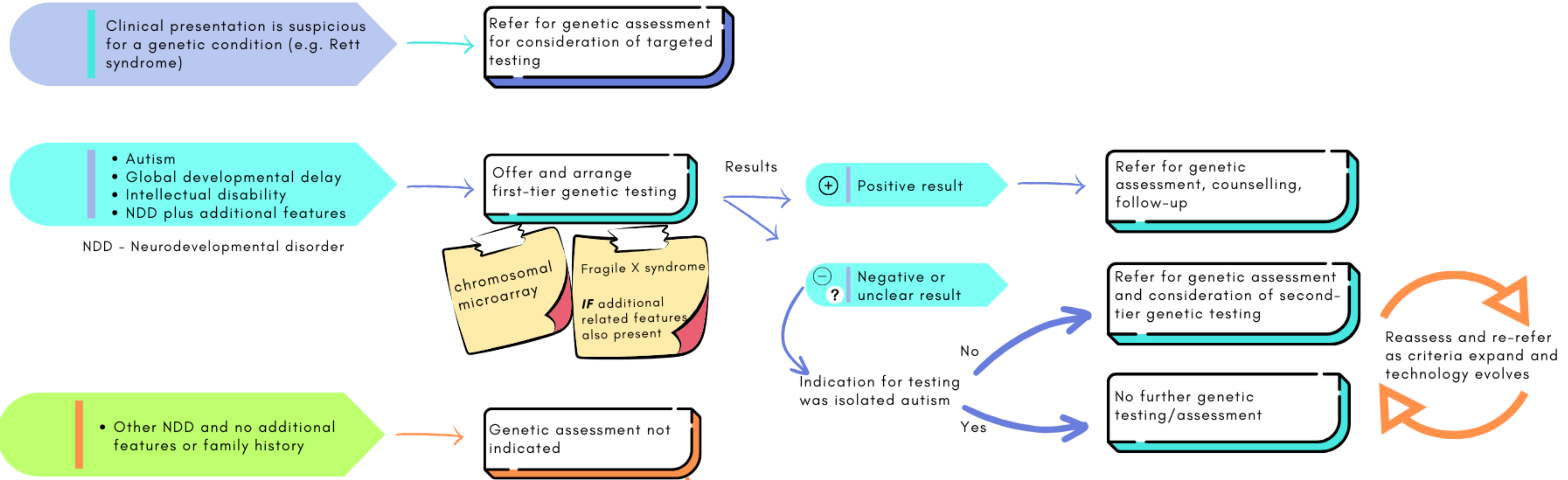
- Division of Genome Diagnostics BC Women's and Children's Hospitals
 - Website
 - CMA - Information | Requisition form
 - Tel: 604-875-2304
 - FXS - Information | Requisition form
 - Tel: 604-875-2852 | Email: moleculargenetics@cw.bc.ca



Refer to Genetics with the results, **unless** the results are normal and the only indication for testing was autism.



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



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)