

Who can order first-tier genetic testing?

Any British Columbia physician can order first-tier testing.

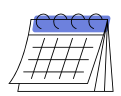
When to order first-tier genetic testing?

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.



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

Who is offered genetic testing?

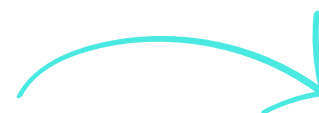
- All autistic individuals
- All individuals with unexplained GDD and/or ID
- Individuals with another NDD plus other health condition

GDD- Global Developmental Delay
ID - Intellectual Disability

 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.