

GENERAL GENETICS - REFERRAL FORM

- Our centre accepts referrals for patients for many indications, including the genetic evaluation of intellectual disability/autism, multiple malformations, cardiac, neurological, or metabolic genetic disorders. Please see page 2 for indications that are no longer accepted.
- Please use the Familial Oncology Program referral form for referrals regarding cancer.
- We may request for you to arrange genetic testing prior to accepting a referral / scheduling an appointment.
- We also accept e-consults through OTNHub (<https://otnhub.ca/>). Please consider sending an e-consult if your referral is regarding management of a diagnosed condition or question of appropriateness of referral. If we have overlooked information, please contact us by phone (613-549-6666 x2800) or fax (613-548-1348).

REFERRING PROVIDER INFORMATION	PATIENT DEMOGRAPHICS
Name _____	Name _____
Phone _____	DOB _____
Fax _____	Phone _____
Signature _____	MOH _____
Date _____	Address _____

Please select the applicable criteria below and include consult notes, imaging, and labs with referral

Suspected genetic syndrome	
Rule out a specific syndrome	
Result counselling	Reason for testing: _____ <input type="checkbox"/> Genetic test result attached (must be included with referral) Has patient been informed of result? Yes _____ No _____
Familial Variant Testing	Relative's name: _____ Relationship to patient: _____ <input type="checkbox"/> Genetic test result/ family letter attached (must be included with referral)
Re-analysis	<input type="checkbox"/> VUS reinterpretation (copy of previous test result must be included with referral) <input type="checkbox"/> Updated testing (copy of previous test result must be included with referral)
Other Reason (Please specify)	

Is this referral urgent? No _____ Yes _____ If yes, why? _____

Please be advised of changes to our triage process

We are no longer accepting referrals for

- Hemochromatosis
- Consanguinity
- Hypermobile Ehlers Danlos Syndrome
- Coagulopathy - please refer to Hematology
- Alpha-Thalassemia outside of pregnancy with only one partner affected
- Ancestry screening if only one partner is of that ancestry (French Canadian, Ashkenazi Jewish)

Familial Hypercholesterolemia (FH)

Our clinic is accepting referrals for patients who have Possible, Probable or Definite FH based on Simon Broome Criteria or Dutch Lipid Network Criteria. We will only accept referrals that include:

- Clear documentation of FH clinical diagnosis by criteria above
- Documentation of cholesterol profile off meds
- All subspecialty clinic notes (eg. endocrinology or cardiology) and/or relevant results in affected family members

Hemoglobinopathies

Our clinic is only accepting referrals for patients who are at significant risk of having a child with a hemoglobinopathy. Their partner must also be referred.

We will only accept referrals in individuals where:

- MCV is <80 in both patient and partner OR
- ONE partner has MCV<75 + normal ferritin OR
- Abnormal Hb electrophoresis in both partners
- CBC, ferritin, hemoglobin electrophoresis is included in referral for both patient and partner

Marfan syndrome

Our clinic accepts referrals for patients who have a clinical diagnosis of Marfan syndrome or who are suspected to have the condition. Please send any ophthalmology records, cardiology records or any family history records.

We will only accept referrals that include:

- Recent (<1yr) echocardiography with aortic dimensions

Congenital Adrenal Hyperplasia (CAH)

We will only accept referrals that include:

- 17-OH-P and/or other relevant endocrine lab results
- Endocrinology clinic notes
- CYP21A2 genetic test results (requisition found here) if 21-OH deficiency is suspected

Retinal Dystrophy or Retinitis Pigmentosa (RP)

We will only accept referrals that include:

- Electroretinogram (ERG) and/or
- Ophthalmology records/clinic notes