



*Maritime Medical Genetics Service
Phone: (902) 470-8754 Fax: (902) 470-8709*

The Maritime Medical Genetics Service is no longer accepting referrals for genetic counselling for individuals with beta thalassemia minor (also known as a carrier for beta thalassemia or beta thalassemia trait). We will only accept a referral if BOTH reproductive partners are carriers (or their partner is a carrier of another beta globin disorder, such as sickle cell trait) AND are planning a pregnancy/currently pregnant.

We would like to direct your attention to the following resources about beta thalassemia minor and hemoglobinopathy carrier screening in Canada:

<https://thalassemia.org/What-is-Thalassemia#a3>

https://geneticseducation.ca/uploads/POC/POC_carrier_screening_Canada_hemoglobinopathies.pdf

Beta thalassemia minor typically does not cause symptoms. People with beta thalassemia minor may have anemia that does not respond to iron supplementation. As beta thalassemia minor does not typically cause a hematologic disorder, people with beta thalassemia minor do not need to be seen by a hematologist in the absence of other concerns.

We suggest consideration of testing in all interested reproductive age first-degree relatives your patient with a complete blood count (CBC), ferritin level, and hemoglobin electrophoresis. Hemoglobinopathy carrier screening is not indicated in healthy children.

A person with beta thalassemia minor is at risk of having children with beta thalassemia major (also known as Cooley's anemia) if their reproductive partner ALSO has beta thalassemia minor. They may also be at risk of having children with a serious hematologic condition if their partner is a carrier of a different beta globin disorder (such as sickle cell trait). Beta thalassemia is inherited in an autosomal recessive manner. If both partners have beta thalassemia minor, there is a 25% chance of having a child with beta thalassemia major with each pregnancy. Reproductive counselling should be provided to any individual with beta thalassemia minor by their primary care provider. Hemoglobinopathy screening should be offered to their reproductive partner and be completed prior to pregnancy, when possible.

If two adults have beta thalassemia minor or are carriers of another beta globin disorder (including sickle cell trait) *and* are planning to have children or are currently pregnant, they can be referred to the Maritime Medical Genetics Service for genetic counselling. **A referral with BOTH individuals' hemoglobinopathy screening reports should be included.**

Children with beta thalassemia minor or adult couples where both individuals have been screened and only one partner is a carrier do not need to be seen by Medical Genetics.

Provider questions can be directed to the Maritime Medical Genetics Service at 902-470-8754.