Summary of Carrier Screening Guidelines for Hemoglobinopathies

Objective: These guidelines ((Wilson et al., 2016); (Aul et al., 2025)) were written to update and provide recommendations for Canadian reproductive care clinicians on pre- and post-conceptional reproductive carrier screening for autosomal recessive (AR) and X-linked (XL) conditions.

Thalassemia/hemoglobinopathies

1. **Screening should be done in the pre-conception period or as early into the pregnancy as possible.** Carrier screening for hemoglobinopathies should be offered to women/families from ethnic backgrounds with a reported increased carrier frequency, when red blood cell indices reveal a mean cellular volume < 80 fl, or electrophoresis reveals an abnormal hemoglobin type. However, the use of ethnicity alone in the carrier risk identification process may create screening inconsistency and missed opportunity for carrier identification, with both obstetrical and fetal implications. High clinical suspicion is required as well.

2. **Carrier screening for thalassemia/hemoglobinopathies should be offered by the most responsible health care provider or reproductive genetic provider and include the following**:

- Complete blood count
- Hemoglobin (Hb) electrophoresis (HE) or Hb high performance liquid chromatography (HHPLC)
- Serum ferritin

***Note:*
*- A qualitative solubility test (e.g., SICKLEDEX) should **not** be used as carrier screening for hemoglobinopathies. This test can miss relevant hemoglobin traits (false negative) or misidentify traits.*
*- **If the patient had previous hemoglobinopathy screening done outside of Canada**, we recommend **repeating** hemoglobinopathy carrier screening due to the chance of false negatives or incorrect results with the initial test.*

3. **If the female thalassemia/hemoglobinopathy screening results are abnormal, a hemoglobinopathy screening protocol should be undertaken for the male partner.**

4. If **both reproductive partners are found to be carriers of thalassemia, hemoglobin variants, or a combination of thalassemia and hemoglobin variant, they should be referred for formal genetic counselling** (reproductive risks, optional prenatal testing, and diagnostic management). *Patients in the Maritimes can be referred to Maritime Medical Genetics Service (MMGS; fax: 1-902-470-8709), based at IWK Health, to receive this genetic counselling.*

Contact the Maritime Newborn Screening genetic counsellor at 1-902-470-2783 for more information.

References:

Aul, R. B., Canales, K. E., De Bie, I., Laberge, A.-M., Langlois, S., Nelson, T. N., Walji, S., Yu, A. C., & Lazier, J. (2025). Reproductive carrier screening for genetic disorders: position statement of the Canadian College of Medical Geneticists. 0, 1–9. https://doi.org/10.1136/jmg-2025-110871

Wilson, R. D., De Bie, I., Brown, R. N., & Armour, C. M. (2016). Joint SOGC-CCMG Opinion for Reproductive Genetic Carrier Screening: An Update for All Canadian Providers of Maternity and Reproductive Healthcare in the Era of Direct-to-Consumer Testing. 38(8), 742–762. https://doi.org/10.1016/j.jogc.2016.06.008 PMID: 27638987 [RETIRED]