

The Canadian Guidelines for Prenatal Diagnosis recommend preconception counselling and screening/testing for individuals belonging to population groups known to have an increased risk for carrying certain genetic disorders (Tables 1 and 2) in order to arrange for prenatal testing if appropriate.<sup>1</sup>

**Table 1. Geographic distribution of ethnic populations at risk for hemoglobinopathies [reproduced from [2]].**

It is recommended that all pregnant women from an ethnic background at increased risk of hemoglobinopathy and/or thalassemia (Table 1) be screened by both CBC, to assess the MCV and MCH, and hemoglobin electrophoresis or high performance liquid chromatography (HPLC).<sup>2</sup>

Regions of Origin	Thalassemia	Sickle Cell Disease (SCD)
<b>Africa</b>	✓	✓ <i>SCD carrier frequency among African Americans is ~ 8-10% and in many regions of Africa it is as high as 25-35%</i>
<b>Mediterranean region</b> e.g. Sardinia, Corsica, Sicily, Italy, Spain, Portugal, Greece, Cyprus, Turkey, Egypt, Algeria, Libya, Tunisia, Morocco, Malta	✓	✓
<b>Middle East</b> e.g. Iran, Iraq, Syria, Jordan, Saudi Arabia and other Arabian peninsula countries, Qatar, Lebanon, Palestine, Israel (both Arabs and Sephardic Jews affected), Kuwait	✓	✓
<b>South East Asia</b> e.g. India, Afghanistan, Pakistan, Indonesia, Bangladesh, Thailand, Myanmar	✓	✓ <i>in parts of India</i>
<b>Western Pacific region</b> e.g. Southern China, Vietnam, Philippines, Malaysia, Cambodia, Laos	✓	
<b>Caribbean countries</b>	✓	✓
<b>South American countries</b>	✓	✓

Note: Japanese, Koreans, Caucasians of Northern European ancestry, Native Americans (First Nations in Canada), and Inuit are not at increased risk of hemoglobinopathies.<sup>2</sup>

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**Table 2. Genetic disorders where a higher carrier frequency is found in individuals of a particular ancestry.**

Disease	Carrier frequency in at-risk population	Recommendation for offering carrier testing
<b>Tay Sachs disease<sup>1</sup></b>	Ashkenazi Jewish (AJ) 1/30  French Canadians (FC) in Eastern Quebec 1/14	DNA-based mutation analysis is recommended for: <ul style="list-style-type: none"> <li>✓ all at-risk couples</li> </ul> <i>(where one individual of the couple is non-AJ or non-FC from Eastern Quebec, biochemical testing can be considered)</i>
<b>Canavan disease<sup>3</sup></b> <b>Familial dysautonomia<sup>3</sup></b>	Ashkenazi Jewish 1/37-57 Ashkenazi Jewish 1/32	DNA-based mutation analysis is recommended for: <ul style="list-style-type: none"> <li>✓ all at-risk couples</li> </ul>
<b>Cystic fibrosis<sup>1</sup></b>	Northern European 1/25-30 Ashkenazi Jewish 1/24	DNA-based mutation analysis is recommended for: <ul style="list-style-type: none"> <li>✓ Individuals with a family history of CF (and their partners)</li> <li>✓ Both parents of a fetus with an ultrasound diagnosis of echogenic bowel</li> </ul> Canadian College of Medical Geneticists does <b>not</b> recommend carrier testing for the general population at this time.

*NOTE: Genetic testing for conditions more often seen in Ashkenazi Jewish ancestry is frequently offered as a panel. Contact your [contact your local genetics centre for more information](#).*

Contact [your local genetics centre](#) to learn more about the community you serve and if there is an at-risk population you should be considering for referral for genetic consultation (e.g. individuals from Saguenay-Lac-St-Jean/Charlevoix regions of Quebec).

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### References

- [1] Chodirker BN, Cadrin C, Davies GAL et al. Canadian guidelines for prenatal diagnosis. Genetic indications for prenatal diagnosis. SOGC Clinical Practice Guideline No. 105, June 2001. *J Obstet Gynaecol Can* 2001;23:525–31
- [2] Langlois S, Ford JC, Chitayat D et al. Carrier screening for thalassemia and hemoglobinopathies in Canada. SOGC Clinical Practice Guideline No. 218, October 2008. *J Obstet Gynaecol Can* 2008; 30(10): 950-971
- [3] Langlois S, Wilson RD, Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada. Prenatal Diagnosis Committee of the Canadian College of Medical Geneticists Carrier screening for genetic disorders in individuals of Ashkenazi Jewish descent. *J Obstet Gynaecol Can* 2006; 28(4):324–43