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Laboratory Bulletin

DATE:	29 September 2025
TO:	All Zones
FROM:	Molecular Genetics Laboratory South, Genetics and Genomics, APL and Clinical Biochemistry, APL
RE:	**Alpha Thalassemia Reflex Testing for Pregnant Patients**

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Key Message

- Effective October 1, 2025, the mean corpuscular volume (MCV) threshold for triggering reflex alpha thalassemia molecular testing following a Hemoglobinopathy Investigation in pregnant patients will be reduced from <85 fL to <80 fL.
- Based on a retrospective review of Alberta patients, this change maintains 100% sensitivity for alpha thalassemia trait carrier detection.

Background

- The Hemoglobinopathy Investigation Panel is a first-line test used to screen patients for hemoglobinopathies and thalassemia. During pregnancy, one purpose of this test is to identify individuals with alpha thalassemia trait, which is defined as having two non-functional copies of the *HBA* gene (out of four total).
- If the Hemoglobinopathy Investigation suggests alpha thalassemia trait, reflex molecular genetic testing is performed to confirm carrier status. Partner testing may be recommended to identify couples at risk of having offspring with clinically significant alpha thalassemia, allowing them to make informed decisions regarding reproductive options and prenatal diagnosis.
- Previously, individuals with an MCV <85 fL, no previous MCV ≥85 fL, and no previous alpha thalassemia molecular testing were reflexed for *HBA* molecular genetic testing. The updated MCV threshold for reflex testing will be reduced to <80 fL. This aligns with current Canadian and international guidelines, as well as evidence-based literature and data from Alberta patients.
- Based on historical data, the implementation of this change is expected to eliminate the unnecessary testing of approximately 350 pregnant individuals annually.

How this will impact you

- The updated MCV threshold will not affect the sensitivity of detecting alpha thalassemia trait carriers, which, based on a retrospective review of Alberta patients, will be maintained at 100%.
- A comprehensive assessment for alpha thalassemia silent carrier status (defined as one non-functional copy of the *HBA* gene) is not included with the current hemoglobinopathy investigation testing algorithm, as this state is typically clinically and hematologically silent. However, genetic testing may be warranted if a current or prospective partner carries two alpha thalassemia deletions/mutations in cis (i.e., alpha thalassemia trait in cis).
- Note that the change to the MCV reflex threshold in pregnant patients does not alter reflex testing or access to molecular genetic *HBA*-related disorder testing for other indications for pregnant or non-pregnant populations, including:

- Clarification of other abnormal hemoglobinopathy investigation results (e.g. Hb variants)
- Confirmation of diagnosis (including MCV ≥80 fL in the presence of other suspicious indices)
- Silent carrier testing in the presence of a carrier partner (alpha thalassemia trait in cis)
- Family studies

Action Required

- Providers can continue to order the Hemoglobinopathy Investigation Panel following the current process, and reflex molecular testing will be coordinated by the laboratory when indicated.
- If genetic testing is indicated to identify silent carrier status (i.e., in the context of a partner with alpha thalassemia trait in cis), this can be coordinated directly through the Molecular Genetics Laboratory South by ordering “HBA-related disorder”.

Effective **October 1, 2025**

Questions/Concerns

- G&G South MGL Genetic Counsellors, 403-955-3097
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Approved by

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