Laboratory Bulletin

Date: November 22, 2012

To: All Zones - Physicians and Laboratory Directors and Managers

From: Calgary Laboratory Services (CLS)

Re: Globin Gene Deletion Screening for Alpha Thalassemia

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Key Messages:

Effective November 19, 2012, Calgary Laboratory Services (CLS) Molecular Hematology Laboratory initiated the transition towards performing province-wide, DNA-based screening for 7 common globin gene deletional variants associated with alpha thalassemia. It is expected that this transition will be completed for all labs within the province over the next few months.

Why this is important:

Alpha thalassemia is one of the major hemoglobin disorders prevalent in individuals of particular ethnic backgrounds from world regions where malaria is endemic. It is associated with large gene deletions at the alpha globin gene locus of which 7 common deletional variants account for more than 90% of all alpha thalassemia cases. Screening for common gene deletions can aid in diagnosis of alpha thalassemia, and the results can be used for carrier determination/genetic counselling in affected families.[1,2]

References:

1. Langois S, Ford JC, Chitayat D *et al*. (2008) Carrier Screening for Thalassemia and Hemoglobinopathies in Canada. JOGC: 218:950.
2. Ryan K, Bain B, Worthington D *et al.* (2010) Significant haemoglobinopathies: guidelines for screening and diagnosis. Brit J Haematol 149:35.

Action Required:

Individuals with suspected alpha thalassemia should be referred for biochemical testing (iron studies and hemoglobinopathy screening) to rule out other causes of microcytosis/anemia (iron deficiency and beta thalassemia). When indicated, individuals will be selected for follow-up molecular analysis by the consulting hematopathologist / biochemist.

This molecular analysis will only be performed on:

- Individuals for whom other causes of microcytosis and/or anemia have been ruled out by initial biochemical assays.
- Family members of known alpha thalassemia patients.

Fetal testing in the context of genetic counseling will continue to be performed at the Molecular Genetics Service Laboratory at Alberta Children's Hospital.

Target test turn-around-time is 10 business days from time of sample receipt at the Molecular Hematology Laboratory. Results will be available electronically through Alberta Netcare.

Inquiries and feedback may be directed to:

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