Alberta Health Services

Laboratory Bulletin

Date: April 21, 2016
To: All Zones
From: Genetic Laboratory Services (GLS)
Re: Changes to molecular testing for TWIST-related and FGFR1, 2, and 3-related craniosynostosis

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Key Messages:

- Effective immediately, the GLS South Molecular Diagnostic Laboratory is offering a craniosynostosis panel consisting of mutations reported to be associated with craniosynostosis in the FGFR1, FGFR2, FGFR3, and TWIST genes.
- Sequencing of the entire FGFR3 gene will no longer be available as it does not increase the detection rate of mutations for the above diseases.
- Testing for known mutations will continue to be available for at-risk individuals or pregnancies.
- The turn around time is expected to be approximately 6-8 weeks for non-urgent samples and approximately 1-2 weeks for urgent samples.

Why this is important:

- Previously, testing was performed for craniosynostoses individually based on phenotype, however, there is a significant phenotypic overlap. As a result, many patients had multiple tests performed sequentially before identifying the molecular cause.
- All patient samples referred for craniosynostosis testing will be tested with the new panel which includes mutations in FGFR1, FGFR2, FGFR3, and TWIST genes.

Action Required:

- The GLS South Molecular Diagnostic Laboratory requisition is available on the GLS webpage located at http://www.albertahealthservices.ca/lab/page8667.aspx). Please ensure that the requisition is completed in full.
- Craniosynostosis ordering privileges are limited to clinical geneticists, paediatricians, and maternal-fetal medicine specialists.

Inquiries and feedback may be directed to:

GLS South Genetic Counsellors (403-955-3097)

This bulletin has been reviewed and approved by:

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