

Date: December 1, 2015
To: All Zones
From: Genetic Laboratory Services – South, Molecular Diagnostic Laboratory
Re: Changes to molecular testing for FGFR3-related skeletal dysplasias

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Key Messages:

- Effective immediately, the Calgary Molecular Diagnostic Laboratory (MDL) will offer an FGFR3-related skeletal dysplasia panel consisting of mutations reported to be associated with hypochondroplasia (HCH), achondroplasia (ACH) or thanatophoric dysplasia (TD).
- 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 familial mutations will continue to be available for at-risk individuals or pregnancies.
- The turnaround time will remain 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 each disease individually; however, there is significant phenotypic overlap between HCH and ACH, and ACH and TD. Many patients had multiple tests performed prior to identifying the molecular cause.
- Any patient sample referred for testing for HCH, ACH or TD will be tested for FGFR3 exons encompassing known mutations responsible for all three diseases simultaneously.

Action Required:

- If you suspect your patient has an FGFR3-related skeletal dysplasia, complete a Calgary MDL requisition (available at <http://www.albertahealthservices.ca/lab/page8667.aspx>) requesting FGFR3-related skeletal dysplasia panel and give it to your patient. They can take it to their local phlebotomy lab for bloodwork.
- Physicians with FGFR3-related skeletal dysplasias ordering privileges include clinical geneticists, paediatricians and maternal-fetal medicine specialists.

Inquiries and feedback may be directed to:

- Genetic Laboratory Services – South, Genetic Counsellors (403-955-3097)

This bulletin has been reviewed and approved by:

Dr. Martin Somerville, Medical/Scientific Director, Genetic Laboratory Services
Dr. James Wesenberg, Provincial Medical/Scientific Director, Laboratory Services