Alberta Health Services

Laboratory Bulletin

Date: December 03, 2015
To: All Zones
From: Genetic Laboratory Services (GLS) – South, Molecular Diagnostic Laboratory
Re: Fragile XE syndrome testing discontinued

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Key Messages:

- Effective January 1, 2016, Genetic Laboratory Services will no longer perform testing for Fragile XE syndrome (FRAXE) due to mutations in the FMR2 gene.
- Testing for Fragile XA syndrome (FRAXA) due to mutations in the FMR1 gene will continue to be available.

What is Fragile XE syndrome?

- FRAXE syndrome is a very rare, X-linked form of non-syndromic intellectual disability that primarily affects males.
- No Alberta patients have been diagnosed with FRAXE syndrome in the history of offering this test.

Action Required:

- Individuals with a non-syndromic intellectual disability should continue to be offered genetic testing for FRAXA syndrome and chromosomal microarray. A referral for genetic assessment for these individuals may also be appropriate.
- Testing for FRAXE syndrome for appropriate patients (for example, known family history) is still available through the Genetic Resource Centre.

Inquiries and feedback may be directed to:

GLS South Genetic Counsellors: Allison Sluyters and Kim Gall (403-955-3097)

Molecular Diagnostic Lab Requisitions and Genetic Resource Centre information and forms are located on the GLS website at http://www.albertahealthservices.ca/lab/Page8667.aspx

This bulletin has been reviewed and approved by:
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