ALBERTA PRECISION LABORATORIES

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Laboratory Bulletin

Date: **December 14, 2020**
To: **All Zones**
From: **Genetics and Genomics**
Re: **CFTR Sequencing for Intronic Variant c.3140-26A>G**

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Key Message:

- The Molecular Genetics Laboratory (MGL) North has changed the analysis performed for *CFTR* sequencing to include the intronic variant c.3140-26A>G (historical nomenclature 3272-26A-->G) due to scientific evidence that this variant is pathogenic[1].

Why this is important:

- This change applies to samples received in the laboratory after March 1, 2019.
  - For cases reported since this change, this region was analyzed and this variant was reported if detected.
  - For cases reported prior to this change, this region was not analyzed, therefore this variant would not have been reported.

Action Required:

- For patients with one pathogenic variant and an elevated sweat chloride value, where CFTR sequencing was performed prior to March 1, 2019, the clinician may wish to request review of the sequencing data. This can be initiated by faxing a requisition that includes the patient demographics, ordering provider's information, and request for CF sequencing reanalysis to MGL at 780-407-1761. Providers using Connect Care should NOT make this request via Connect Care.

Inquiries and feedback may be directed to:

- Edmonton MGL genetic counsellors at 780-407-1015.

This bulletin has been reviewed and approved by:

- Dr. Dennis Bulman, Medical/Scientific Director, Genetics & Genomics, APL
- Dr. Carolyn O'Hara, Chief Medical Laboratory Officer (Interim), APL

References

1. https://www.ncbi.nlm.nih.gov/clinvar/variation/35864/