ALBERTA PRECISION LABORATORIES
Leaders in Laboratory Medicine

Laboratory Bulletin

DATE:	2021 June 28
TO:	All Zones
FROM:	Genetics and Genomics
RE:	**CFTR Sequencing for Intronic Variants**

PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Message

- The Molecular Genetics Laboratory (MGL) North has updated the analysis performed for *CFTR* sequencing to include the intronic variants c.1680-886A>G (legacy name 1811+1.6kbA->G) and c.1680-877G>T (legacy name 1811+1643G->T) based on scientific evidence regarding pathogenicity. These variants are in addition to the intronic variant c.3140-26A>G (historical nomenclature 3272-26A→G) described in the lab bulletin dated December 14, 2020 (*CFTR* sequencing for intronic variant c.3140-26A>G).

How this will impact you

This change applies to samples received in the laboratory after January 1, 2019.

- For cases received after January 1, 2019, this region was analyzed and these variants were reported if detected.
- For cases received prior to January 1, 2019, this region was not analyzed and these variants would not have been reported.

Action Required

- For patients with one pathogenic *CFTR* variant and an elevated sweat chloride value, where *CFTR* sequencing was performed prior to January 1, 2019, the clinician may wish to request testing for these intronic variants. **A new blood sample is required to complete this testing.** This testing can be initiated by ordering *CFTR* testing in Connect Care or using the Molecular Genetic Requisition (Molecular Genetics Requisition (Edmonton) (albertahealthservices.ca). Please note in the Connect Care order or on the requisition that the request is for “intronic variant analysis”.

Questions/Concerns

- G&G North (Edmonton) MGL Genetic Counsellors at 780-407-1015

Approved by

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

References

ClinVar entry for c.1680-886A>G (www.ncbi.nlm.nih.gov/clinvar/variation/53338/)
ClinVar entry for c.1680-877G>T (www.ncbi.nlm.nih.gov/clinvar/variation/53331/)