ALBERTA PRECISION LABORATORIES

Leaders in Laboratory Medicine

Laboratory Bulletin

Date: November 16, 2020
To: All Zones
From: Genetics and Genomics (G&G)
Re: *PTEN* Testing Methodology Update

PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Message:

- Pathogenic variants in *PTEN* are indicative of *PTEN* Hamartoma Tumor syndrome and increased risk of breast cancer.
- Variants detected in the *PTEN* promoter region are classified as benign or variants of uncertain significance, and the associated cancer and/or health risks are unknown[1,2].
- Effective October 1, 2020, the Molecular Genetics laboratories are no longer analyzing the *PTEN* promoter region when *PTEN* is included in next-generation sequencing panel or single-gene testing.

Why this is important:

- Effective October 1, 2020, analysis of the *PTEN* promoter region is no longer being performed, and variants in this region will not be reported.

Action Required:

- None

Inquiries and feedback may be directed to:

- G&G North Genetic Counsellors (phone: 780-407-1015)
- G&G South Genetic Counsellors (phone: 403-955-3097)

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. Black MH, Li S et al. *PTEN* Promoter variants are not associated with common cancers: implications for multigene panel testing. JCO Prec Onc. October 2017
2. Landrum MJ, Lee JM, Benson M, et al. ClinVar: public archive of interpretations of clinically relevant variants. Nucleic Acids Res. 2016 Jan 04;44(D1):D862-8