

#### Leaders in Laboratory Medicine

# **Laboratory Bulletin**

DATE:	2022 July 25
TO:	All Pathologists and Hematologists/Oncologists in the South Sector
FROM:	Molecular Pathology Program, Alberta Precision Laboratories
RE:	In-House Testing for ASXL1 Mutations

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## **Key Message**

 As of July 18, 2022, for all samples received for Alberta Precision Laboratories Myeloid Panel testing in Molecular Pathology South, ASXL1 mutations will be covered by in-house testing. Sendout ASXL1 testing performed in Colorado will cease. ASXL1 mutations, if detected, will be listed in the Myeloid Panel NGS report together with all other findings on the NGS panel (not as a separate report or addendum).

## **Background**

- ASXL1 mutations are prognostic biomarkers with clinical importance in acute myeloid leukemia, myelodysplastic syndrome, myeloproliferative neoplasms, and other myeloid malignancies. Due to a limitation in the ThermoFisher Oncomine Myeloid Assay run in Alberta Precision Laboratories South Sector, a significant proportion of clinically relevant ASXL1 mutations could not be detected in-house. Therefore, since clinical testing on the panel began in June 2022, samples have been sent to Colorado for supplemental testing to cover for ASXL1 mutations.
- A laboratory developed test has been designed and validated in the FMC Molecular Pathology laboratory to cover the specific ASXL1 mutations that cannot be reliably detected on the ThermoFisher Oncomine Myeloid Assay. The in-house test uses a PCR sizing methodology run on capillary electrophoresis to detect indels in the region of interest in ASXL1.

## How this will impact you

- All Myeloid NGS testing performed in Molecular Pathology South will have supplemental testing for ASXL1
  mutations. The combination of NGS and supplemental testing will ensure the vast majority of clinically relevant
  ASXL1 mutations are detected in-house. All NGS and ASXL1 assay findings will be integrated into a single report.
- Samples tested on Myeloid NGS in Molecular Pathology South will no longer be sent to Colorado for supplemental ASXL1 testing.

## **Action Required**

Pathologists, Oncologists and Hematologists: Be aware when reviewing Myeloid NGS results that ASXL1
testing is now incorporated in APL Molecular Pathology South in-house testing, and integrated into a single
report. You no longer need to look for ASXL1 addendum reports from sendout testing.

## Effective July 18, 2022

#### **Questions/Concerns**

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#### Approved by

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