Date: December 6, 2021

DISCIPLINE: GENOMICS

Testing for KIT D816V Mutation

Date effective: December 6, 2021

Background Information:

KIT gene sequencing is now available for patients diagnosed with systemic mastocytosis and acute myelogenous leukemia (AML). Prior to September 22, 2021, D816V point mutation analysis and c-Kit gene sequencing on blood and bone marrow was sent to the Mayo Clinic Laboratories, and D816V point mutation analysis on FFPE tissue was sent to Stanford Clinical Laboratories for testing. The new in-house sequencing covers the D816 codon and replaces both send-out tests. Eligible specimens include blood, bone marrow and formalin-fixed paraffin embedded (FFPE) tissue.

This test can be ordered by Shared Health Hematopathologists and CancerCare Manitoba Hematologists & Oncologists.

Refer to the current versions of requisitions and laboratory policies for specimen requirements.

Change in Test Procedure:

- This testing is now offered in-house through the Molecular Diagnostics Laboratory, Genomics, Diagnostic Services.
- Samples will be tested using next-generation sequencing on the Q31 Hotspot Tumour panel, which covers exons 2, 8-11, 13-15 and 17, including the codon D816.

References/Resources:

- R250-10-29 Molecular Hematopathology Test Requisition

Patient Impact:

- None anticipated.

System Improvements:

- By performing the test in-house, a shorter turn-around-time for results is expected.

For More Information Contact:

- Dr. Elizabeth Spriggs, Medical Director, Genomics, Diagnostic Services bspriggs@sharedhealthmb.ca
- Dr. Paul Park, Associate Medical Director, Genomics, Diagnostic Services, Shared Health ppark@sharedhealthmb.ca