

DATE:	20 March 2023
TO:	All Zones
FROM:	Genetics and Genomics, Alberta Precision Laboratories
RE:	New Tyrosinemia and Alkaptonuria Monitoring Test

PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Message

- APL Genetics and Genomics now offers dried blood spot (DBS) monitoring for known Tyrosinemia and Alkaptonuria patients.

Background

- Patients with inborn errors of tyrosine catabolism (i.e., Tyrosinemia Type 1-3 and alkaptonuria) require regular monitoring of specific amino acids and succinylacetone. The new DBS Tyrosinemia monitoring will fill this need and utilize existing Newborn Screening workflows for efficiency. This testing option may be used to replace both in-province plasma amino acids quantification (AAQ) and out of province DBS testing for this patient population. Reducing in province AAQ shifts workload away from limited capacity instruments while providing a more convenient collection type for patients. Additionally, repatriation of Tyrosinemia monitoring enhances turnaround times and achieves cost savings. Testing includes quantification of tyrosine, phenylalanine, methionine and succinylacetone with age specific reference ranges.

How this will impact you

- Ordering DBS Tyrosinemia monitoring for regular monitoring of Tyrosinemia and Alkaptonuria patients. Note that this DBS test does not include Nitisinone quantification, which must be ordered as a Miscellaneous send-out test.

Action Required

- Information for and ordering of Tyrosinemia Monitoring, Dried Blood Spot, Test code 14342 is available in ConnectCare. Alternatively, the test can be ordered on the recently updated Genetics and Genomics Biochemical Genetics Requisition.

Effective **Immediately**

Questions/Concerns

- Dr. Ross Ridsdale, Biochemical genetics North, Ross.Ridsdale@albertaprecisionlabs.ca, 780-407-4570
- Dr. Iveta Sosova, Biochemical genetics North, Iveta.Sosova@albertaprecisionlabs.ca, 780-407-7379
- Dr. Lauren MacNeil, Biochemical genetics North, Lauren.MacNeil@albertaprecisionlabs.ca, 780-407-1275

Approved by

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