Retinoblastoma Genetic Testing

Applicability
This document applies to all healthcare providers who are ordering retinoblastoma genetic testing for an Alberta patient.

Purpose
This policy provides information regarding when retinoblastoma genetic testing should be performed at a facility outside Alberta.

Policy
Retinoblastoma genetic testing may be performed at a facility outside Alberta in the following situations:

1. Tumor sample testing - genetic testing of RB1 tumor tissue is not presently offered in Alberta
2. Proband testing (tumor and/or blood sample) when the patient is being treated at the Hospital for Sick Children in Ontario
3. Clinically indicated rush testing in which the GLS South Molecular Diagnostic Lab (MDL) is unable to complete testing within the required turn-around-time
   a. Expedited proband testing (blood sample) can be completed within 4 weeks at MDL
   b. Expedited targeted testing for a familial variant can be completed within 2 weeks at MDL providing a locus-specific assay has been developed in advance (MDL must be notified minimally 3 weeks in advance of commencing testing to order locus-specific reagents and ensure functionality).

Funding for out-of-province testing will be provided through the Genetic Resource Center.

For all other indications, retinoblastoma genetic testing will be performed at MDL in Alberta. Testing situations include:

1. Proband testing (blood sample) when the patient is Alberta Health-insured and not undergoing treatment in Ontario at the time of testing
2. Familial testing (including prenatal diagnosis) for a known pathogenic variant
   a. It is the responsibility of the ordering healthcare provider to coordinate collection of a positive control sample if DNA is not banked at MDL

Responsibility
Ordering healthcare providers and Genetic Lab Services personnel are responsible for implementing this policy

Reference
N/A