



## **Retinoblastoma Genetic Testing**

### **APPLICABILITY**

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

### **PURPOSE**

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

### **GUIDELINE**

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

1. Tumor specimen testing - genetic testing of RB1 tumor tissue is not presently offered in Alberta
2. Proband testing (tumor and/or blood specimen) when the patient is being treated at the Hospital for Sick Children in Ontario
3. Clinically indicated rush testing in which the Genetics & Genomics South Molecular Genetics Lab (MGL) is unable to complete testing within the required turn-around-time
  - a. Expedited proband testing (blood specimen) can be completed within 4 weeks at MGL
  - b. Expedited targeted testing for a familial variant can be completed within 2 weeks at MGL providing a locus-specific assay has been developed in advance (MGL 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 Centre.

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

1. Proband testing (blood specimen) 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 specimen if DNA is not banked at MGL

### **RESPONSIBILITY**

Ordering healthcare providers and Genetics & Genomics personnel are responsible for implementing this guideline.