



Sample Submission Instructions for RB1 Molecular Analysis

Samples Required, by Type of Analysis

Bilateral proband or positive family history: Blood sample required, fresh tumor sample useful if available.

Unilateral proband with no family history: Both blood sample and fresh or flash-frozen tumor sample necessary for complete analysis. Partial diagnosis is possible on blood only without flash-frozen or fresh tumor specimen.

Fetus: Cultured amniocytes or CVS tissue with maternal blood sample (at the same time).

Genetically related family member for known mutation: Blood sample only.

Sample Preparation Instructions

- Blood Samples for DNA:** 10 mls (for infants 2-5 ml in pediatric or small tubes) venous blood in yellow-topped ACD tubes or lavender-topped EDTA tubes at room temperature, to be received within 5 days after draw.
- Blood Samples for RNA:** 10 mls (for infants 2-5 ml in pediatric or small tubes) venous blood in yellow-topped ACD tubes or lavender-topped EDTA tubes on 4°C cool packs, to be received within **48 hours** after draw.
- Amniocytes:** Two T25 flasks of cultured amniotic cells or DNA extracted from amniotic cells.
- CVS:** Send CVS tissue.
- Fresh Tumor in Medium** (Instructions for enucleating surgeon or pathology lab)
After removal of an eye containing retinoblastoma,
 1. Cut off optic nerve and retain as separate specimen for pathology lab.
 2. Open globe by pupillary-optic nerve section as in routine eye pathology.
 3. Excise or scoop the bulk of the tumor from inside the eye, leaving tumor-optic nerve and tumor-choroidal relationship undisturbed for pathological evaluation.
 4. Place fresh retinoblastoma tumor in a sterile tissue culture media, such as RPMI with 100 milligrams/microliter of penicillin and streptomycin with 10-15% serum.
 5. Seal the container securely with parafilm.
 6. Tumor may be refrigerated until it is ready to be shipped. **DO NOT FREEZE** tumor in tissue culture.
 7. Retain the remainder of the eye for pathology.
 8. Send tumor sample at room temperature.
- Frozen Tumor Sample Preparation:** Flash freeze tumor sample and ship on enough dry ice to keep frozen for maximum transit time.

Sample Identification: Label each sample with at least two patient identifiers (name, date of birth, MRN, e.g.), plus the date and time sample was obtained.

Shipping: *Ship all samples in rigid, leak-proof packaging to Retinoblastoma Solutions at address shown above.*

1. Please use FedEx Next Day delivery and use a FedEx "Clinical Specimen Bag." On the waybill mark that FedEx is the Customs Clearing Agent for the package. If you cannot use FedEx, please contact us.
2. For samples from outside Canada,
 - a. Mark on the waybill that the specimen is non-hazardous, non-toxic and non-infectious.
 - b. To avoid Customs clearing delays, Declare value at US\$10, mark that the recipient will pay duties, and do not include "Toronto Western Hospital" in the mailing address.
 - c. Include a Commercial Invoice or Pro-Forma Invoice (available online, phone us if help is required).
3. Send us the parcel tracking number soon after courier pick-up: (416) 603-5597, info@retinoblastomasolutions.org

Turnaround Times: 4-14 wks (5.5 mean) unknown mut; 2-4 wks (2.5 mean) known mut; 7 working days rush case for known mut (prenatal or infants at risk aged 3 yrs or less).