



Retinoblastoma Solutions

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Retinoblastoma (RB1) Genetic Testing Requisition

Patient

Last Name _____
First Name _____
Date of Birth yyyy / mmm / dd
Gender M F

Patient History

Affected
 Bilateral Unifocal Multifocal
 Unilateral Unifocal Multifocal
 Phenotype Unknown
Diagnosis Date yyyy / mmm / dd
 Unaffected
 Fetus/newborn, Delivery Date yyyy / mmm / dd
 Sample to test Maternal Cell Contamination

Isolated Case Positive Family History
Family Previously Tested? Yes No
Mutation Identified? Yes No
Proband Name _____
Mutation _____

Relationship to Proband

Proband (first person in a family to be studied)
 Parent of Proband
 Brother or Sister of Proband
 Child of Proband: Son Daughter Prenatal
 Other _____

Specimen Info

Blood Sample ACD/EDTA(5-10 mls)
 DNA from Blood
 Fresh Tumor
 Frozen Tumor
 DNA from Tumor
 Tumor to follow **No Tumor to follow**

Pre-Natal:
 Cord Blood CVS Cultured Amniocytes

Date of Collection: yyyy / mmm / dd

Time of Collection: HH:MM (use 24h clock)

Referring Specialist

Name _____
Specialty _____
Contact _____
Tel _____ Fax _____
e-mail _____
Institution _____
Address _____

City _____ Prov (St) _____
Postal Code _____ Country _____
Additional Copies to: _____
e-mail _____ Fax _____

Billing Information

Payment enclosed
 Bill Institution Bill Patient
 Bill Third Party Insurer
 Information and Pre-approval enclosed

Billing Address

Name _____
Address 1 _____
Address 2 _____
Address 3 _____

Bill Credit Card
 VISA Mastercard

Cardholder _____
Card Num _____
Expiration yyyy / mmm / dd

Pedigree



INFORMED CONSENT to PERFORM GENETIC TESTING FOR RETINOBLASTOMA

I, _____, consent to participate, or as applicable, to have my child _____ participate, in a DNA-based test to identify an abnormality in the retinoblastoma gene (*RB1* gene). I understand this test requires a blood sample from the person to be tested and in some cases, a fresh tumor sample, and may require blood samples from other people related by blood. I understand that the blood and tumor samples will be used to determine if the subject and the subject's relatives carry a genetic abnormality believed to cause retinoblastoma tumors.

By signing below, I acknowledge that:

1. My participation and as applicable, my child's participation, in this DNA testing is voluntary.
2. The removal of up to 10 ml of blood (5 ml for infants) required for the test carries a low risk of discomfort and infection.
3. Although the lab makes every effort to ensure the accuracy of test results, there remains a small possibility of human error and a very small chance that there exists in the subject's DNA a second mutation not identified by the test. Consequently, a remote but real possibility remains that the DNA test results lead to an inaccurate diagnosis.
4. A person whose DNA contains a genetic abnormality associated with retinoblastoma does not necessarily develop the disease. A negative test result does not imply that the subject has no chance to develop retinoblastoma.
5. All test results are confidential; no information will be printed or released that discloses the patient's identity without my additional written permission. Only the referring physician I designate on the Retinoblastoma Requisition Form will receive a written report of test results.
6. *Retinoblastoma Solutions* is not a DNA banking facility and patient DNA samples may not be available for future testing.
7. An error in diagnosis may occur if the true biological relationships of the family members are not as stated in the pedigree submitted with the Retinoblastoma Requisition Form. It is possible that the test may disclose paternity and I consent that this finding be reported to the physician designated on the Retinoblastoma Requisition Form.
8. Until the results of this test are reported, the patient and members of the patient's family should still undergo retinal examinations as prescribed by my physician.
9. My referring physician reviewed this consent form with me, point by point, and explained the implications of the test results to me. Any questions that I asked have been answered to my satisfaction. I know that I (or members of my family) may ask any questions we have about the collection, use and disclosure of our personal genetic information.
10. If necessary to obtain reimbursement of test fees, *Retinoblastoma Solutions*, its agents and legal representatives may disclose information that identifies me or my children who are subject to *RB1* genetic testing.
11. I received a copy of this consent form and the referring physician whom I designate on the Retinoblastoma Requisition Form received a copy of this consent form.
12. *Retinoblastoma Solutions* has my consent to use the subject's DNA in an anonymous fashion for research to improve the sensitivity of *RB1* genetic tests.

 Signature of Subject or Consenting Parent

 Date

Statement of Referring Physician: I reviewed this form with my client, point by point. I offered to answer any questions regarding personal genetic information for the client or the client's children.

 Signature of Referring Physician

 Date



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. Will test blood only - if no fresh or frozen tumor is available.

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 at room temp.
- CVS:** Send CVS tissue in sterile tissue culture medium at room temperature.
- 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 and seal container securely with parafilm.
 5. Tumor may be refrigerated until it is ready to be shipped. **DO NOT FREEZE** tumor in tissue culture.
 6. Retain the remainder of the eye for pathology.
 7. 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 International Priority service (Next-Day) and use a FedEx "Clinical Specimen Bag."
If you cannot use FedEx, please contact us.
2. For samples from outside Canada,
 - a. Include 4 copies of a Commercial or Pro-Forma Invoice (available online, phone us if help is required)
 - b. Mark on the waybill and Commercial or Pro-Forma Invoice that the specimen is non-hazardous, non-toxic and non-infectious and *Bill Customs charges to Recipient*.
 - c. To avoid Customs clearing delays, declare value at US\$10 on waybill and invoice, and do not include "Toronto Western Hospital" in the mailing address.
3. Send us the parcel tracking number soon after courier pick-up: (416) 603-5597, info@retinoblastomasolutions.org.