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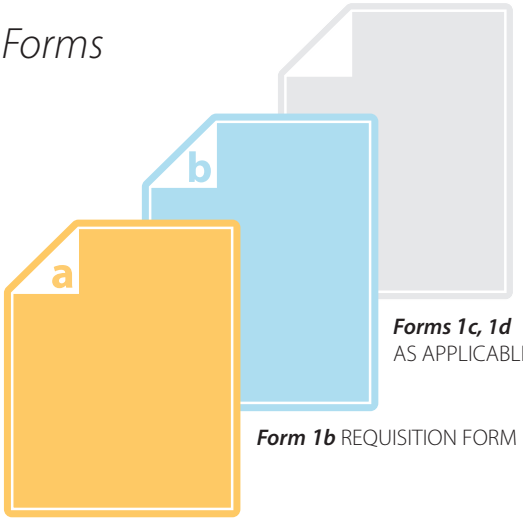
Retinoblastoma

Genetic Test Submission Guide

1 Forms

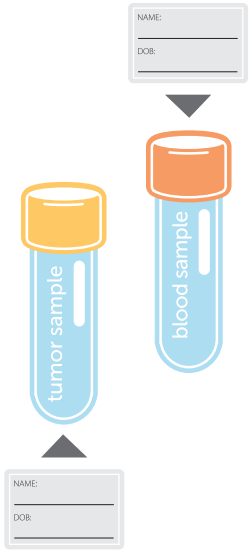


Forms 1c, 1d AS APPLICABLE

Form 1b REQUISITION FORM

Form 1a INFORMED CONSENT

2 Samples



3 Ship

Impact Genetics
115 Midair Court
Brampton, ON L6T 5M3
1-877-624-9769

Results

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Form 1a: *Informed Consent to Perform Genetic Testing*

The purpose of my DNA test/or my child's DNA test is to look for mutation(s) or genetic alterations known to be associated with the following genetic condition or disease: Retinoblastoma.

By signing below, I acknowledge that:

1. My participation or, as applicable, my child's participation in this DNA testing is voluntary. The decision to consent to, or to refuse the above testing is entirely mine.
2. This testing is done on small biological samples.
3. It is possible that the quantity or quality of sample submitted may be inadequate for testing or that a mutation cannot be identified.
4. When DNA testing shows a mutation or alteration, then the person is a carrier or is affected with that condition or disease, or, in the case of cancer genetic testing, the person is a carrier of a mutation or alteration that may be associated with an increased risk for certain cancer(s) compared to the general population. Consulting a doctor or genetic counsellor is recommended to learn the full meaning of the results and to learn if additional testing might be necessary.
5. When the DNA testing does not show a known mutation or alteration, the chance that the person is a carrier or is affected is reduced, or, in the case of cancer genetic testing, the person's risk for certain cancer(s) compared to the general population will depend on additional personal factors. There is still a chance to be a carrier or to be affected because the current testing cannot find all the possible changes within a gene.
6. Impact Genetics will disclose the test results ONLY to the specialist designated on the Requisition Form (***Form 1b***), or to his/her agent, unless otherwise authorized by the patient or required by law, except as described in point 11 below, no information will be printed or released that discloses the patient's identity, or other confidential information.
7. Impact Genetics is not a DNA banking facility and patient DNA samples may not be available for future testing.
8. 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 Requisition Form (***Form 1b***). It is possible that the test may disclose non-paternity (someone who is not the biological father), or some other previously unknown information about family relationships, such as adoption, and I consent that this finding be reported to the referring specialist designated below.
9. There is a chance that the test may reveal unexpected abnormalities that may be of medical value in the patient's care. Impact Genetics will inform the referring specialist designated below.
10. Until the results of this test are reported, the patient and members of the patient's family should still undergo examinations as prescribed by the referring specialist.
11. If necessary to obtain reimbursement of test fees, Impact Genetics, its agents and legal representatives may disclose information that identifies the patient or other confidential information (including test results).
12. I have read or have had read to me, the above information and I understand it. I have also read or had explained to me the specific disease or condition tested for, and the specific test(s) I am having, including the test descriptions, principles and limitations. I have had the opportunity to discuss the purposes and possible risks of this testing with my doctor or someone my doctor has designated.

Consent for Future Research:

After all analysis required to reach a genetic diagnosis is complete, Impact Genetics has my consent to use any surplus DNA or RNA in an **anonymous** fashion for research. No tests(s) will be performed and reported on my sample other than the one(s) authorized by my doctor.

☐ YES ☐ NO

Signature of Patient: _____ Date: _____

Signature of Witness: _____ Date: _____

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LAB USE ONLY DO NOT FILL OUT

Date received: Y M D

Specimen type:

Condition:

MRN: Tech:

Form 1b: Retinoblastoma Genetic Test Requisition

Patient

Last name:

First name:

Date of birth: Y M D

Gender: ☐ Male ☐ Female

Pregnant: ☐ Yes ☐ No

Delivery date: Y M D

Patient History

☐ Affected

☐ Bilateral ☐ Unilateral ☐ Phenotype unknown

Diagnosis date: Y M D

☐ Unaffected

Family History

☐ Isolated case ☐ Positive family history

Family previously tested: ☐ Yes ☐ No

Mutation identified: ☐ Yes ☐ No

If mutation identified at lab other then Impact Genetics please provide report.

Proband name (first person in a family to be studied):

Mutation:

Relationship To Proband

☐ Proband ☐ Parent of proband

☐ Brother or sister of proband ☐ Child of proband

☐ Other:

Specimen Information

Sample:

☐ Blood sample for DNA

☐ Blood sample for RNA (at Impact Genetics' request)

☐ DNA from blood

☐ DNA from tumor

☐ Fresh tumor

☐ Frozen tumor

☐ Buccal swab (only for preapproved familial mutation confirmation, contact lab directly before submitting)

☐ Other:

Pre-natal:

☐ Cord blood ☐ CVS ☐ Cultured amniocytes

☐ Direct amniotic fluid ☐ DNA extracted from CVS

☐ DNA extracted from amniocytes

Date of collection: Y M D

Time of collection: HH:MM (24hr)

☐ Sample to test maternal cell contamination

☐ Tumor to follow

☒ No tumor to follow

Referring Specialist

Name: Scott A. Larson, MD

Specialty: Ophthalmology

Contact:

Telephone: 319-256-2859 Fax: 319-353-7996

Email: scott-a-larson@uiowa.edu

Signature: Scott A. Larson

Institution: University of Iowa

Address: 200 Hawkins Drive

City: Iowa City Prov/State: Iowa

Postal code: 52242 Country: USA

Additional copies to:

Email: Fax:

Pedigree

Billing

☒ a) Institution

Provide details: University of Iowa

☐ b) Patient Pay

*Complete **Form 1d:** Credit Card Authorization for Non-Covered Services.*

115 Midair Court, Brampton, ON L6T 5M3

t: 647-478-4902 or 1-877-624-9769 f: 905-697-9786

e: info@impactgenetics.com *Please ensure to use secure email*

Rev 21Feb2020 (CAN)

PLEASE DO NOT SEND FORM WITH SAMPLE;
Send this form to Impact Genetics
BY FAX TO 905-697-9786
For patient pay, testing will be held pending receipt of this completed form.

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Form 1d: *Credit Card Authorization for Non-Covered Services*

To be completed by and returned to Impact Genetics ***directly by the cardholder****.*

Billing Information

Laboratory Test:

☐ Retinoblastoma Genetic Test
☐ Uveal Melanoma Prognostic Genetic Test
☐ NeuroSURE℠: Epilepsy Gene Panel Test
☐ *BAP1*-TPDS (*BAP1 Tumor Predisposition Syndrome*) Genetic Test
☐ HHT Genetic Test
☐ *MLH1/MSH2/MSH6/PMS2/EPCAM* Somatic Tumor MMR Genetic Test

Patient name: _____ Date of birth: Y ____ M ____ D ____

☐ VISA ☐ MASTERCARD

Name on card: _____

Billing address: _____

City: _____

Province/State: _____ Postal/Zip code: _____

Country: _____

Card #: _____ Expiration date: _____

CVC # *(3-digit Card Verification Code at back of card)*: _____

Contact Information

Impact Genetics will contact the cardholder prior to placing the credit card charge, to confirm the date and amount of the charge. Please ***provide at least 2 contact methods*** *and check preferred:*

☐ Phone: _____

☐ Email: _____

☐ Fax: _____

Statement of Financial Responsibility U.S. PATIENTS ONLY

Box below must be checked for testing to proceed.

☐ I understand that my insurance plan is not expected to pay for these test(s) at 100% and I agree to be personally and fully responsible for payment.

Cardholder's signature: _____ Date: _____

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Step 2: *Retinoblastoma Genetic Test Sample Requirements*

U.S. insurance patients: a completed ***Form 1c****: U.S. Insurance Information* must be provided.
All submitted samples must be labeled with at least two patient identifiers (ie. name and date of birth).

Sample Requirements

Bilateral proband or positive family history:

- Blood or DNA from blood.
- Fresh tumor sample is useful if available.

Unilateral proband with no family history:

- Both blood sample and fresh or flash-frozen tumor sample are useful for complete analysis.

Pre/peri-natal sample options:

- Cultured amniocytes
- Direct amniotic fluid
- CVS tissue
- Cord blood
- DNA extracted from amniocytes
- DNA extracted from CVS

Maternal blood or DNA from blood (required for all pre/perinatal samples, to be submitted at same time or prior to procedure).

Genetically related family member for known mutation:

- Blood or DNA from blood;
- Or buccal swab, (select cases only, contact lab to confirm).

Provide family member's genetic test report if genetic testing has been performed at a lab other than Impact Genetics

Sample Preparation Instructions

Blood samples for DNA:

- 10 mls in yellow-topped ACD tubes **or** lavender-topped EDTA tubes (for infants 2-5 mls in pediatric or small tubes) at room temperature, to be received within 5 days after draw.

DNA from blood:

- DNA concentration—100 nanograms/microliter.
- DNA quantity—150 microliters.

Blood samples for RNA:

- 10 mls in lavender-topped EDTA tubes (for infants 2-5 mls in pediatric or small tubes) on 4° C cool packs, to be received within 48 hours after draw.

Buccal swab:

- Kit available (select cases only contact lab directly to request); follow included instructions.

Amniocytes:

- Two T25 flasks of cultured amniotic cells **or** DNA extracted from amniotic cells at room temperature (100 nanograms/microliter: minimum 20 microliters).

Direct amnio:

- As much volume as possible in conical tube at room temperature, to be received within 48 hours after draw.

CVS:

- Send CVS tissue in sterile tissue-culture medium at room temperature **or** DNA extracted from CVS tissue at room temperature (100 nanograms/microliter: minimum 20 microliters).

Fresh tumor in medium

(instructions for enucleating surgeon or pathology lab):
After removal of an eye containing retinoblastoma,

- Cut off optic nerve and retain as separate specimen for pathology lab.
- Open globe by pupillary-optic nerve section as in routine eye pathology.
- Excise or scoop the bulk of the tumor from inside the eye, leaving tumor-optic nerve and tumor-choroidal relationship undisturbed for pathological evaluation.
- Place fresh retinoblastoma tumor in a sterile tissue culture media, such as RPMI or Alpha MEM with antibiotic added (100U/ml penicillin and 100ug/ml streptomycin) and seal container securely with parafilm.
- Tumor may be kept at room temperature until it is ready to be shipped. **DO NOT FREEZE** tumor in tissue culture.
- Retain the remainder of the eye for pathology.
- 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.
- Detailed tumor extraction information available on our website:

  http://impactgenetics.com/testing-services/retinoblastoma/info-for-rb-clinicians/

Sample Identification

- Label each sample **with at least two** patient identifiers (e.g. name and date of birth).

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Step3: *Shipping Requirements*

Shipping Instructions

For all samples:

- Ship samples in rigid, leak-proof packaging to Impact Genetics at address shown on this page.
- Include Informed Consent and Requisition Forms (***1a*** and ***1b)*** with the samples. Patients in the U.S. must also include U.S. Insurance Information (***Form 1c***) if required and not provided previously.
- Within Canada, use **Purolator Express** (next-day) or **FedEx Priority** service (next-day). Outside of Canada use **FedEx Priority** service (next-day) and **use a FedEx "Clinical Pak"**. If you cannot use Purolator or FedEx, please contact us.
- Provide us with the parcel tracking number soon after courier pick-up: **647-478-4902**, **info@impactgenetics.com**.
- For emailed PDF FedEx waybills and customs forms, please contact us directly.

For samples from outside Canada:

- Complete all fields and sign 4 copies of the Pro-Forma Invoice (*available on our website; phone us if help is required*). Place the 4 Pro-Forma Invoice copies in the document pouch.
- Mark on the Air Waybill and Pro-Forma Invoice:
  EXEMPT HUMAN DIAGNOSTIC SPECIMEN – non-hazardous, non-toxic and non-infectious.
- To avoid Customs clearing delays, declare value at US $10 on the Air Waybill and Pro-Forma Invoice.
- Select **Bill Customs Charges to Recipient**.

Send to Impact Genetics

mail: Impact Genetics
115 Midair Court
Brampton, ON L6T 5M3

tel: 1-877-624-9769

fax: 905-697-9786