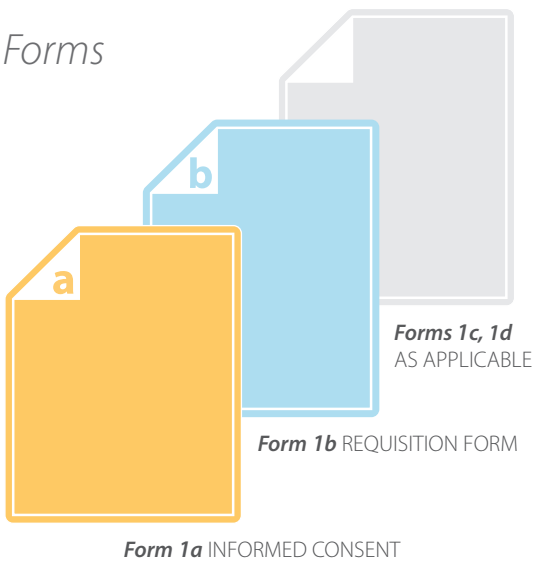


impact genetics

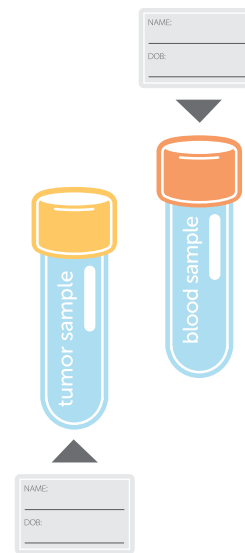
Retinoblastoma

Genetic Test Submission Guide

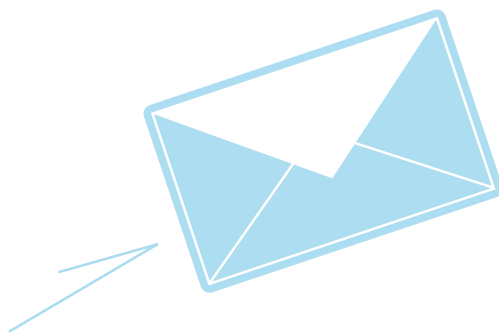
1 Forms



2 Samples



3 Ship



Impact Genetics
1100 Bennett Road - Unit 4
Bowmanville, ON LIC 3K5
1-877-624-9769

Results





impact genetics

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: _____.

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: _____

**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 FemalePregnant: 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 historyFamily previously tested: Yes NoMutation identified: Yes No*If mutation identified at lab other than 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: _____

Specialty: _____

Contact: _____

Telephone: _____ Fax: _____

Email: _____

Signature: _____

Institution: _____

Address: _____

City: _____ Prov/State: _____

Postal code: _____ Country: _____

Additional copies to: _____

Email: _____ Fax: _____

Pedigree**Billing** a) InstitutionProvide details: _____

_____ b) Patient Pay*Complete Form 1d: Credit Card Authorization for Non-Covered Services.* c) Third party insurance (USA only)*Complete Form 1c: U.S. Insurance Information and to expedite testing, complete Form 1d: Credit Card Authorization for Non-Covered Services.***1100 Bennett Road - Unit 4, Bowmanville, ON L1C 3K5****t: 647-478-4902 or 1-877-624-9769 f: 905-697-9786****e: info@impactgenetics.com** *Please ensure to use secure email*

Rev 15Mar2017



Form 1c: *U.S. Insurance Information*

Impact Genetics is committed to providing the highest quality genetic testing to all patients. In many situations, genetic testing improves outcomes and quality of life and decreases total costs to the patient and healthcare system.

Processing medical insurance claims is usually challenging and time consuming. Many insurance companies require pre-authorization prior to testing. Impact Genetics supports insurance billing, completing coverage checks and pre-authorization.

It is important for patients to understand that insurance rarely covers 100% of the cost of genetic testing and that they will be financially responsible for some or all of the cost of testing. The patient is responsible for any portion of the test fee not covered by insurance for any reason, including but not limited to, co-payments, unmet deductibles, co-insurance and non-covered services. Prior determinations do not guarantee payment and the amount paid by insurance when the claim is submitted may be different from the coverage indicated during the pre-verification or pre-authorization process.

Pre-authorizations can take time to obtain depending on each individual insurance plan's policy and documentation requirements. Turnaround time for test results begins after the pre-authorization has been processed and approved.

Insurance process

1. Send **Form 1c:** *U.S. Insurance Information* as soon as possible.
2. Send **Form 1d:** *Credit Card Authorization for Non-Covered Services* to initiate testing immediately.
3. Insurance coverage will be investigated and patient/specialist will be contacted to provide coverage estimate.
4. Insurance claim will be submitted upon completion of testing.
5. After insurance payment is received patient will be billed for non-covered services.

Note: Timely and complete submissions will enable faster insurance checks.



Send this form to Impact Genetics
BY FAX TO 905-697-9786

Form 1c: U.S. Insurance Information - Retinoblastoma

Disease/Genetic Test

Retinoblastoma Genetic Test

ICD-10 Code *provide code here* ▶ _____

CPT Codes: 81479, 81265 or S3841

Insurance Information

Primary insurance Secondary insurance

If Patient has secondary insurance, include the information on an additional copy of this form with the secondary insurance box checked.

Name of insured (if not Patient):

Insurance company: _____

Claims address: _____

City: _____

State: _____ Zip code: _____

Country: _____

Group #: _____

Subscriber/member #: _____

Physician Information

Physician's name: _____

NPI: _____

Practice name: _____

Practice Address: _____

Telephone: _____

Fax: _____

*Testing process will be initiated when
Form 1d: Credit Card Authorization for Non-Covered Services
is received or confirmation is received from insurance provider.*

Patient Information

Last name: _____

First name: _____

Date of birth: Y _____ M _____ D _____

Address: _____

City: _____

State: _____ Zip code: _____

Country: _____

Telephone: _____

Contact Information

*Details of insurance coverage will be communicated.
Please provide preferred telephone number(s):*

Patient Referring specialist

Telephone: _____

Alternate telephone: _____

Email: _____

In the event Patient cannot be reached a voice message related to retinoblastoma genetic testing may be left at the above phone number(s)

Please Attach All of the Following

- Copies of both the front and back of insurance membership card(s)
- Letter of Medical Necessity, signed by Referring Specialist (contact Impact Genetics for template if needed)
- Clinic notes demonstrating the Patient's need for testing and confirmation of diagnosis
- Insurance approval details *if* prior pre-approval completed

Performing Lab - Impact Genetics

1100 Bennett Road - Unit 4

Bowmanville, ON L1C 3K5 CANADA

t: 1-877-998-7837 f: 1-888-598-7568

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

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.



impact genetics

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
 HHT Genetic Test

- Uveal Melanoma Prognostic Genetic Test
 Uveal Melanoma 5 Gene Panel (SF3B1, EIF1AX, GNAQ, GNA11, BAP1)
 BAP1-TPDS (BAP1 Tumor Predisposition Syndrome) 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: _____

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: _____



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).



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
1100 Bennett Road - Unit 4
Bowmanville, ON L1C 3K5

tel: 1-877-624-9769

fax: 905-697-9786