

Step 2: Sample Preparation Accepted Somatic/Tumour Samples Reference Sheet—Retinoblastoma

All samples **must** be labelled with **at least 2** patient identifiers (e.g. full name and DOB).

We do not accept formalin-fixed (FFPE) tissue.

Sample Preparation Instructions (instructions for enucleating surgeon or pathology lab)

Fresh tumour in medium:

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 tumour from inside the eye, leaving tumour-optic nerve and tumour-choroidal relationship undisturbed for pathological evaluation.
- Place fresh retinoblastoma tumour in a sterile tissue culture media, such as RPMI or Alpha MEM with antibiotic added (100 U/mL penicillin and 100 ug/mL streptomycin) and seal container securely with parafilm.
- Tumour may be kept at room temperature until it is ready to be shipped. **Do not freeze** tumour in tissue culture.
- Retain the remainder of the eye for pathology.
- Send tumour sample at room temperature.

Frozen tumour sample preparation:

- Flash freeze tumour sample and ship on enough dry ice to keep frozen for maximum transit time.
- Detailed tumour extraction information available on our website:
<http://impactgenetics.com/testing-services/retinoblastoma/info-for-rb-clinicians>

DNA extracted from fresh or frozen RB tumour:

- Extracted DNA **must** be performed by a CLIA-accredited laboratory.
- Extraction method **must** be provided
- DNA concentration: 100 ng/ μ L
- DNA Quantity: 150 μ L
- Ship at room temperature

Step 2: Sample Preparation

Accepted Germline Samples Reference Sheet

All samples **must** be labelled with **at least 2** patient identifiers (e.g. full name and DOB).

Sample Type for Germline Analysis	Sample Requirements	Shipment Instructions
Blood for DNA analysis	<ul style="list-style-type: none"> • 5 mL in lavender top (EDTA) tube, or 10 mL in yellow top (ACD) tube • 2-5 mL in pediatric/small tubes for infants 	<ul style="list-style-type: none"> • Ship at room temperature • To be received within 5 days of blood draw
Blood for RNA analysis	<ul style="list-style-type: none"> • 5 mL in lavender top (EDTA) tube • 2-5 mL in pediatric/small tubes for infants 	<ul style="list-style-type: none"> • Ship on 4°C cool pack • Must be received within 48 hours of blood draw
DNA from blood Provide DNA extraction method. Must be performed at a CLIA-accredited lab.	<ul style="list-style-type: none"> • DNA concentration: 100 ng/μL • DNA Quantity: 150 μL 	<ul style="list-style-type: none"> • Ship at room temperature
Buccal swab	<ul style="list-style-type: none"> • Select cases only • Contact us for collection kit • See <i>Buccal Swab Collection Instructions</i> sheet for details 	<ul style="list-style-type: none"> • Ship at room temperature
Direct CVS	<ul style="list-style-type: none"> • Send CVS tissue in a sterile tissue culture medium 	<ul style="list-style-type: none"> • Ship at room temperature
DNA extracted from CVS Provide DNA extraction method. Must be performed at a CLIA-accredited lab.	<ul style="list-style-type: none"> • DNA concentration: 100 ng/μL • DNA Quantity: minimum 20 μL 	<ul style="list-style-type: none"> • Ship at room temperature
Direct amniotic fluid	<ul style="list-style-type: none"> • As much volume as possible in conical tube 	<ul style="list-style-type: none"> • Ship at room temperature • Must be received within 48 hours of collection
Cultured amniocytes/ CVS	<ul style="list-style-type: none"> • Two T25 flasks of cultured amniotic cells 	<ul style="list-style-type: none"> • Ship at room temperature
DNA extracted from amniocytes Provide DNA extraction method. Must be performed at a CLIA-accredited lab.	<ul style="list-style-type: none"> • DNA concentration: 100 ng/μL • DNA Quantity: minimum 20 μL 	<ul style="list-style-type: none"> • Ship at room temperature
Maternal blood for MCC	<ul style="list-style-type: none"> • 5 mL in lavender top (EDTA) tube, or 10 mL in yellow top (ACD) tube 	<ul style="list-style-type: none"> • Ship at room temperature
Maternal DNA extracted from blood for MCC Provide DNA extraction method. Must be performed at a CLIA-accredited lab.	<ul style="list-style-type: none"> • DNA concentration: 100 ng/μL • DNA Quantity: 150 μL 	<ul style="list-style-type: none"> • Ship at room temperature
Other	<ul style="list-style-type: none"> • For alternative collection methods please contact us for confirmation 	<ul style="list-style-type: none"> • Ship according to method instructions