

Step 2: Sample Preparation

Accepted Samples Reference Sheet – HHT and Retinoblastoma Genetic Tests

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)	<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)	<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)	<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)	<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