

SPECIMEN COLLECTION

Biochemical Genetics:

<i>Sample Type</i>	<i>Code</i>	<i>Collection Container(s)</i>	<i>Collection and Processing Instructions</i>	<i>Shipping Instructions and Preferred Sample Condition</i>
Red Blood Cells, Washed	RBC	Sodium Heparin (Green Top)	<p>Washed Red Blood Cell Preparation:</p> <p>Step 1: Centrifuge the whole blood at 3000rpm (1800rcf) for 5 minutes.</p> <p>Step 2: Remove plasma and buffy coat layer.</p> <p>Step 3: Resuspend the red cells in normal saline (0.9% NaCl) with approximately 2 times the volume of the red cells, and invert the tube to mix.</p> <p>Step 4: Centrifuge for 5 minutes at 2000 rpm and discard the supernatant.</p> <p>Step 5: Repeat Steps 3 & 4 twice for a total of 3 washes or until the supernatant is clear.</p> <p>Step 6: Completely remove the supernatant and freeze the red cells at -20°C, preferably at -80°C.</p> <p>Ship frozen.</p>	Ship frozen sample on dry ice with overnight delivery.
Serum	SRM	Clean container without additives	<p>Spin down, transfer, and ship frozen.</p> <p>1-2 ml (each): AG, CZ, TT</p> <p>3-5 ml (each): BM</p>	Ship frozen sample on dry ice with overnight delivery.
Whole Blood (Sodium heparin)	WBG	Sodium Heparin (Green Top)	3-5 ml (each): GP	<p>Ship sample at room temperature for receipt at NTD within 24 hours of collection. Do not refrigerate or freeze.</p> <p>NOT ACCEPTED ON SATURDAYS</p>

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Whole Blood (No additive)	WBR	No Additive (Red Top) Or SST (Serum Separator Tube - no additives)	1-2 ml (each): AG, CZ, TT, 3-5 ml (each): BM	Ship sample at room temperature for receipt at NTD within 24 hours of collection. Do not refrigerate or freeze. NOT ACCEPTED ON SATURDAYS

Molecular:

<i>Sample Type</i>	<i>Code</i>	<i>Collection Container(s)</i>	<i>Collection and Processing Instructions</i>	<i>Shipping Instructions and Preferred Sample Condition</i>
DNA, Isolated *Only accept DNA that has been isolated or extracted in a CLIA certified laboratory or a laboratory meeting equivalent requirements.	DNA	Microtainer	<p>*Next Generation Sequencing (NGS): 30µg</p> <p>* Sanger Sequencing: 5-25µg (Varies by size of gene. Contact the laboratory for specific amounts.)</p> <p>* Non-Sequencing Tests: 20 µg</p> <p>* Array-Based Tests: 10 µg</p> <p>Isolation using the Qiagen™ Puregene or Perkin Elmer™ Chemagen kit for DNA extraction is recommended</p>	Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery
<p>*Research Laboratories: DNA extracted in research laboratories is not acceptable. Only under exceptional circumstances (e.g. proband not available) will DNA extracted in a research laboratory be accepted for clinical testing. Additional testing (e.g. of other family members) may be required to confirm results.</p> <p>* Non-US Laboratories: Non-US laboratories, which are not subject to CLIA regulations, will be reviewed on a case-by-case basis. Please call to speak with a laboratory genetic counselor prior to submitting a DNA sample from any non-CLIA-certified laboratory.</p>				
Liquid Buccal Swab	LBC	ORAcollection-DX (OCD-100) Assisted saliva collection kit	<p>DNA Genotek ORAcollection-DX kit used according to manufacturer instructions.</p> <p>Contact NTD for a saliva collection kit for patients that cannot provide a blood sample.</p>	Do not refrigerate or freeze saliva sample. Store and ship at room temperature.

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			Note: Whole blood is the preferred sample.	
Nasal/ NasoPharyngeal	NSP	Sterile container containing viral transport media (VTM) or provided universal transport media (UTM)	Contact NTD for Collection kits.	Do not refrigerate or freeze saliva sample. Store and ship at room temperature. *Ensure caps are tightened securely prior to shipment
Saliva	SLV	Oragene™ Saliva Collection kit	Oragene™ Saliva Collection kit used according to manufacturer instructions. Contact NTD for a saliva collection kit for patients that cannot provide a blood sample. Note: Whole blood is the preferred sample.	Do not refrigerate or freeze saliva sample. Store and ship at room temperature.
Whole Blood (EDTA) Preferred sample type	WBP	EDTA (Purple Top)	Infants (<2 years): 2-3 ml Children (>2 years to 10 years old): 3-5 ml Older Children & Adults: 5-10 ml Special Notes and Exceptions: <ul style="list-style-type: none"> • Clotted or hemolyzed samples are not accepted • Southern Blot Analysis: minimum 3 ml blood 	* Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery. * Samples older than 7 days may be acceptable if sufficient and suitable DNA is obtained