

## Whole Genome Sequencing for Hereditary Disorders, Varies

**Test ID:** WGSDX

**Explanation:** On the effective date, WGSDX will include Skin biopsy, Cultured fibroblast, Saliva, and Muscle tissue biopsy as acceptable specimens. Additionally, CULFB will be added to accommodate possible reflexes for the new specimen types.

Current Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed
G227	Number of Comparators for WGSDX	No (Bill Only)	No
MATCC	Maternal Cell Contamination, B	Yes	No

New Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed
G227	Number of Comparators for WGSDX	No (Bill Only)	No
MATCC	Maternal Cell Contamination, B	Yes	No
<b>CULFB</b>	<b>Fibroblast Culture for Genetic Test</b>	<b>Yes</b>	<b>No</b>

Current Specimen Required	
<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p>	
<b>Specimen Type:</b>	<b>Whole blood</b>
<b>Preferred:</b>	Lavender top (EDTA) or yellow top (ACD)
<b>Acceptable:</b>	Any anticoagulant
<b>Specimen Volume:</b>	3 mL
<b>Collection Instructions:</b>	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. <b>Do not aliquot.</b>

New Specimen Required	
<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p>	
<p><b>Submit only 1 of the following specimens:</b></p>	
<b>Specimen Type:</b>	<b>Whole blood</b>
<b>Preferred:</b>	Lavender top (EDTA) or yellow top (ACD)
<b>Acceptable:</b>	Any anticoagulant
<b>Specimen Volume:</b>	3 mL
<b>Collection Instructions:</b>	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. <b>Do not aliquot.</b>

**Additional Information:** If a cord blood specimen is received, MATCC / Maternal Cell Contamination, Molecular Analysis, Varies will be performed at an additional charge; maternal blood sample is required.

**Specimen Stability Information:** Ambient (preferred)/Refrigerated

**Additional Information:** 1. If a cord blood specimen is received, MATCC / Maternal Cell Contamination, Molecular Analysis, Varies will be performed at an additional charge; maternal blood sample is required.  
2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

**Specimen Type:** Skin biopsy

**Supplies:** Fibroblast Biopsy Transport Media (T115)

**Container/Tube:** Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

**Specimen Volume:** 4-mm punch

**Specimen Stability Information:** Refrigerated (preferred)/Ambient

**Additional Information:** A separate culture charge will be assessed under CULFB/Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

**Specimen Type:** Cultured fibroblast

**Container/Tube:** T-25 flask

**Specimen Volume:** 2 Flasks

**Collection Instructions:** Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

**Specimen Stability Information:** Ambient (preferred)/ Refrigerated (<24 hours)

**Additional Information:** A separate culture charge will be assessed under CULFB/Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to

culture fibroblasts before genetic testing can occur.

**Specimen Type:** Saliva

**Patient Preparation:** Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

**Supplies:** Saliva Swab Collection Kit (T786)

**Specimen Volume:** 1 Swab

**Collection Instructions:** Collect and send specimen per kit instructions.

**Specimen Stability Information:** Ambient 30 days

**Additional Information:** Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

**Specimen Type:** Muscle tissue biopsy

**Supplies:** Muscle Biopsy Kit (T541)

**Collection Instructions:** Prepare and transport specimen per instructions in Muscle Biopsy Preparation Instructions.

**Specimen Volume:** 10 to 80 mg

**Specimen Stability Information:** Frozen (preferred)/Ambient/Refrigerated

## Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.