

Whole Genome Sequencing Reanalysis, Varies

Test ID: WGSR

Explanation: On the effective date, WGSR 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

New Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Current Specimen Required	
<p>Patient Preparation: 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>	
Specimen Type:	Whole blood
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. Do not aliquot.

New Specimen Required	
<p>For most patients, a new specimen submission will not be required. Testing can be performed using stored DNA from the original whole genome sequencing test. To order testing on the stored specimen, see Additional Testing Requirements.</p>	
<p>Patient Preparation: 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>Submit only 1 of the following specimens:</p>	
Specimen Type:	Whole blood
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. Do not aliquot.
Additional Information:	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.

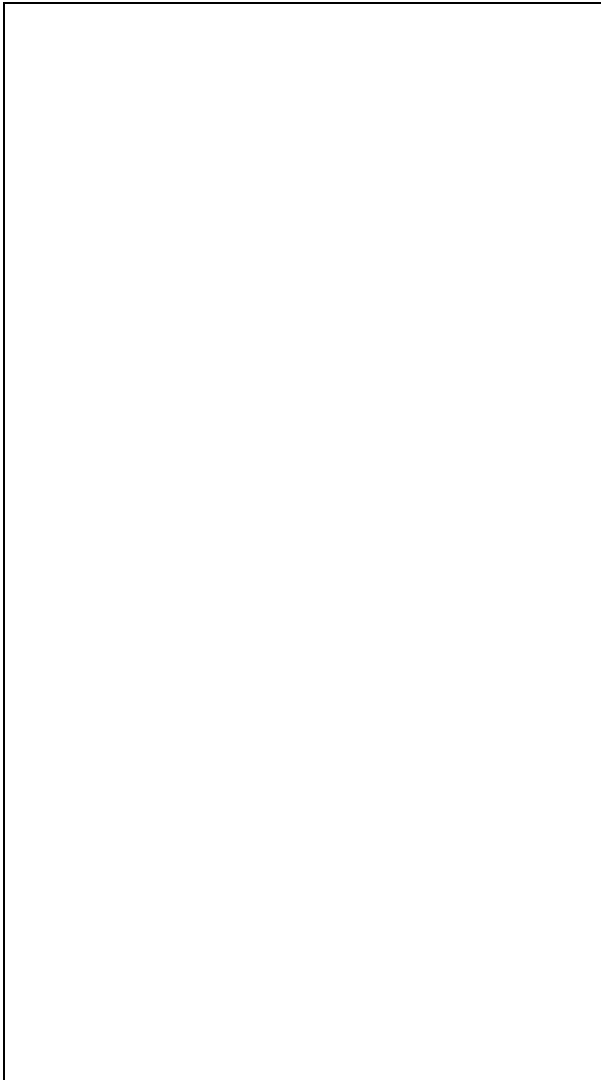
Specimen Stability Information: Ambient (preferred)/ Refrigerated (<24 hours)

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.