

Smith Lemli Optiz, *DHCR7* Gene, Full Gene Analysis, Varies

Test ID: DHCRZ

Explanation:

On the effective date, the changes below will occur, including additional acceptable specimen types and possible reflex tests.

Reflex Tests added:

Test ID	Reporting Name	Available Separately	Always Performed
FIBR	Fibroblast Culture	Yes	No
CRYOB	Cryopreserve for Biochem Studies	Yes	No

Method Name updated:

Current Method Name	New Method Name
Custom Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing	Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

Additional Specimens accepted:

Current Specimens Accepted	New Specimens Accepted
Whole Blood	Whole Blood Skin Biopsy Cultured Fibroblasts Blood Spot Saliva

New Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type:	Whole Blood
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant

Specimen Volume:	3 mL
Collection Instructions:	<ol style="list-style-type: none"> 1. Invert several times to mix blood. 2. Send specimen in original tube. Do not aliquot.
Specimen Stability Information:	Ambient (preferred) 4 days/Refrigerated 14 days
Minimum Volume:	1 mL
Specimen Type:	Skin Biopsy
Supplies:	Fibroblast Biopsy Transport Media (T115)
Container/Tube:	Sterile container with any standard cell culture media (e.g., 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
Specimen Type:	Cultured fibroblast
Container/Tube:	T-25 flask
Specimen Volume:	2 Flasks
Collection Instructions:	<p>Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory.</p> <p>Cultured cells from a prenatal specimen will not be accepted.</p>
Specimen Stability Information:	Ambient (preferred)/Refrigerated (<24 hours)
Additional information	A separate culture charge will be assessed under FIBR / Fibroblast Culture, Tissue. An additional 4 weeks is required to culture fibroblasts before genetic testing can occur.
Specimen Type:	Blood spot
Supplies:	Card-Blood Spot Collection (Filter Paper) (T493)
Preferred:	Collection card (Whatman Protein Saver 903 Paper)
Acceptable	PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card
Specimen Volume:	5 Blood spots
Collection Instructions:	<ol style="list-style-type: none"> 1. An alternative blood collection option for a patient older than 1 year of age is fingerstick. 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours. 3. Do not expose specimen to heat or direct sunlight. 4. Do not stack wet specimens. 5. Keep specimen dry.
Specimen Stability Information:	Ambient (preferred)/Refrigerated
Additional information	Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.
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 concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Testing Duration Changed:

Current Report Available
3 to 4 weeks

New Report Available
4 to 6 weeks

Additional CPT Codes Added:

Current CPT Codes
81405

New CPT Codes
81405 88233-Tissue culture, skin, solid tissue biopsy (if appropriate) 88240-Cryopreservation (if appropriate)

Questions

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