

## Sandhoff Disease, *HEXB* Gene, Full Gene Analysis, Varies

Test ID: HEXBZ

### 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

<b>Specimen Volume:</b>	3 mL
<b>Collection Instructions:</b>	<ol style="list-style-type: none"> <li>1. Invert several times to mix blood.</li> <li>2. Send specimen in original tube. Do not aliquot.</li> </ol>
<b>Specimen Stability Information:</b>	Ambient (preferred) 4 days/Refrigerated 14 days
<b>Minimum Volume:</b>	1 mL
<b>Specimen Type:</b>	<b>Skin Biopsy</b>
<b>Supplies:</b>	Fibroblast Biopsy Transport Media (T115)
<b>Container/Tube:</b>	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.
<b>Specimen Volume:</b>	4-mm punch
<b>Specimen Stability Information:</b>	Refrigerated (preferred)/Ambient
<b>Specimen Type:</b>	<b>Cultured fibroblast</b>
<b>Container/Tube:</b>	T-25 flask
<b>Specimen Volume:</b>	2 Flasks
<b>Collection Instructions:</b>	<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>
<b>Specimen Stability Information:</b>	Ambient (preferred)/Refrigerated (<24 hours)
<b>Additional information</b>	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.
<b>Specimen Type:</b>	<b>Blood spot</b>
<b>Supplies:</b>	Card-Blood Spot Collection (Filter Paper) (T493)
<b>Preferred:</b>	Collection card (Whatman Protein Saver 903 Paper)
<b>Acceptable</b>	PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card
<b>Specimen Volume:</b>	5 Blood spots
<b>Collection Instructions:</b>	<ol style="list-style-type: none"> <li>1. An alternative blood collection option for a patient older than 1 year of age is fingerstick.</li> <li>2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.</li> <li>3. Do not expose specimen to heat or direct sunlight.</li> <li>4. Do not stack wet specimens.</li> <li>5. Keep specimen dry.</li> </ol>
<b>Specimen Stability Information:</b>	Ambient (preferred)/Refrigerated
<b>Additional information</b>	Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.
<b>Specimen Type:</b>	<b>Saliva</b>
<b>Patient Preparation:</b>	Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

<b>Supplies:</b>	Saliva Swab Collection Kit (T786)
<b>Specimen Volume</b>	1 Swab
<b>Collection Instructions:</b>	Collect and send specimen per kit instructions.
<b>Specimen Stability Information:</b>	Ambient 30 days
<b>Additional information</b>	Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

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### ***Testing Duration Changed:***

<b>Current Report Available</b>
3 to 4 weeks

<b>New Report Available</b>
4 to 6 weeks

### ***Additional CPT Codes Added:***

<b>Current CPT Codes</b>
81479

<b>New CPT Codes</b>
81479 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.