



# Laboratory Service Manual

**Lab Dept:** Chemistry

**Test Name:** 7-DEHYDROCHOLESTEROL

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## ***General Information***

**Lab Order Codes:** 7DEH

**Synonyms:** Smith-Lemli-Opitz Screen, RSH syndrome, 8-dehydrocholesterol

**CPT Codes:** 82541 – Column chromatography/mass spectrometry, analyte not elsewhere specified; single stationary and mobile phase

**Test Includes:** 7-dehydrocholesterol level reported as positive or negative. Quantitative results are provided when positive.

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## ***Logistics***

**Test Indications:** Useful in diagnosing Smith-Lemli-Opitz (SLO) Syndrome (3 beta-hydroxysterol-delta 7-reductase deficiency).

**Lab Testing Sections:** Chemistry – Sendouts

**Referred to:** Mayo Medical Laboratories (Test# 81595)

### **Phone Numbers:**

Minneapolis: 612-813-6580

Saint Paul: 651-220-6550

**Test Availability:** Daily, 24 hours

**Turnaround Time:** Varies, test performed 1 day per week (2 - 8 days)

**Special Instructions:** [See Patient Preparation](#)

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## ***Specimen***

**Specimen Type:** Blood

**Container:** Green top (Lithium heparin) tube

**Draw Volume:** 3.0 mL blood

**Processed Volume:** 1.0 mL (Minimum: 0.2 mL) plasma



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<b>Collection:</b>	Routine venipuncture. Mix specimen thoroughly by gentle inversion.
<b>Special Processing:</b>	Lab Staff: Centrifuge immediately after collection, separate and freeze plasma. Store and ship frozen in a plastic vial. Forward promptly.
<b>Patient Preparation:</b>	4 or more hour fasting specimen preferred (infants before next feeding).
<b>Sample Rejection:</b>	Warm specimens, anticoagulants other than heparin or EDTA, mislabeled or unlabeled specimens

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## *Interpretive*

<b>Reference Range:</b>	Negative (if positive, quantitative results are provided)  Interpretation: Elevated plasma concentrations of 7- and 8-DHC are highly suggestive of a biochemical diagnosis of SLO.  Mild elevations of these cholesterol precursors can be detected in patients with hypercholesterolemia and patients treated with haloperidol. However, the 7-DHC to cholesterol ratio is only elevated in SLO patients.
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	The enzymatic tests for measuring plasma cholesterol quantitate all three hydroxysterols and are unreliable for diagnosis for SLO.
<b>Methodology:</b>	Gas Chromatography/Mass Spectrometry (GC/MS)
<b>References:</b>	<a href="#">Mayo Medical Laboratories Web Page</a> (February 2010)