
Lab Dept: Chemistry

Test Name: 7-DEHYDROCHOLESTEROL

General Information

Lab Order Codes: 7DEH

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

CPT Codes: 82542 – Column includes mass spectrometry, if performed, none drug analyte(s) not elsewhere specified, qualitative or quantitative, each specimen

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

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 (MML Test: SLO)

Phone Numbers: MIN Lab: 612-813-6580

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 3 - 9 days, performed Tuesday and Friday

Special Instructions: Deliver to lab as soon as possible

Specimen

Specimen Type: Blood

Container: Green top (Lithium heparin) tube
Alternate: Green (Na Heparin), Lavender (EDTA) or Yellow ACD (A/B)

Draw Volume: 1.5 mL (Minimum: 0.6 mL) blood

Processed Volume:	0.5 mL (Minimum: 0.2 mL) plasma
Collection:	Routine blood collection. Mix specimen thoroughly by gentle inversion.
Special Processing:	Lab Staff: Centrifuge specimen, separate and freeze plasma. Store and ship frozen in a plastic vial. Forward promptly.
Patient Preparation:	N/A
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	7-Dehydrocholesterol: < or =2.0 mg/L 8-Dehydrocholesterol: < or =0.3 mg/L 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.
Critical Values:	N/A
Limitations:	Cholesterol screening tests are unreliable for diagnosis for Smith-Lemli-Opitz syndrome Aripiprazole and trazodone cause false elevations in 7-dehydrocholesterol. On rare occasions, 7-dehydrocholesterol is not elevated in patients with Smith-Lemli-Opitz syndrome.
Methodology:	Gas Chromatography/Mass Spectrometry (GC/MS)
References:	Mayo Medical Laboratories (May 2021)
Updates:	1/28/16: CPT update. 2/15/17: Collection tube update. 11/3/17: Draw volume update. 5/6/21: Updated reference ranges