
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: Varies, test performed 1 day per week (3 - 9 days)

Special Instructions: Deliver to lab as soon as possible, must be processed within 45 minutes of collection. [See Patient Preparation](#)

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

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| Processed Volume: | 0.5 mL (Minimum: 0.2 mL) plasma |
| Collection: | Routine venipuncture. Mix specimen thoroughly by gentle inversion. |
| Special Processing: | Lab Staff: Centrifuge within 45 minutes of collection, separate and freeze plasma. Store and ship frozen in a plastic vial. Forward promptly. |
| Patient Preparation: | 12 or more hour fasting specimen preferred (infants before next feeding). |
| Sample Rejection: | Mislabeled or unlabeled specimens |

Interpretive

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| Reference Range: | 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. |
| 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. |
| Methodology: | Gas Chromatography/Mass Spectrometry (GC/MS) |
| References: | Mayo Medical Laboratories Web Page (February 2017) |
| Updates: | 1/28/16: CPT update. 2/15/17: Collection tube update. 11/3/17: Draw volume update. |