Condition Description

Smith-Lemli-Opitz syndrome (SLOS) is an inherited disorder of cholesterol biosynthesis. Cholesterol is an essential structural component of membranes and the precursor of all other sterols, steroids and bile acids in the body. A little more than half the cholesterol of the body arises by synthesis (about 700 mg/d).

The biosynthesis of cholesterol is a complex pathway catalyzed by enzymes. Reduced activity of 7-dehydrocholesterol reductase leads to the accumulation of 7- and 8-dehydrocholesterol and reduced total cholesterol in plasma. Biochemical diagnosis of the disorder is by an elevated 7-dehydrocholesterol/cholesterol ratio.

The severity of the SLOS clinical phenotype can be highly variable ranging from individuals who have minor features and normal development to those with severe mental retardation and congenital anomalies. These anomalies may include prenatal and postnatal growth retardation, microcephaly, characteristic facial features, cleft palate, cardiac anomalies, postaxial polydactyly, 2-3 syndactyly of the toes and males with underdeveloped genitalia.

Please click here for a GeneReviews clinical summary.


Indications

This test is indicated for:

• Patients whose clinical evaluations are suggestive of Smith-Lemli-Opitz syndrome

**This test is not useful for carrier screening.**

Methodology

Qualitative and quantitative determination performed by gas chromatography/mass spectrometry.

Detection

Mild elevations of 7-dehydrocholesterol can occur secondary to certain medications. These elevations can overlap with those seen in mildly affected individuals. Further testing may be necessary to clarify mild abnormalities. A detailed medication history can aid in the interpretation of results.

Reference Range

The reference range is 7DHC ≤ 2.0 µg/mL.

Specimen Requirements

Type: Plasma

Specimen Requirements:

In sodium heparin (green top) tube: 1-5 ml

Sample should be collected while fasting or 2-4 hours postprandial.

Centrifuge to separate plasma and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Special Instructions

Patient name, date of birth, and clinical information required.