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MAYO CLINIC LABORATORIES

Test Definition: SLO

Smith-Lemli-Opitz Screen, Plasma

Overview

Useful For

Diagnosing Smith-Lemli-Opitz syndrome (7-dehydrocholesterol reductase deficiency) and fetal fentanyl syndrome

Genetics Test Information

Smith-Lemli-Opitz syndrome (SLO) is a multiple congenital anomaly disorder caused by defective cholesterol biosynthesis due to deficiency of the enzyme 7-dehydrocholesterol (7-DHC) reductase.

Clinical variability even within families has been noted and severity of SLO ranges from severe to mild.

Elevated plasma concentrations of 7-DHC and 8-dehydrocholesterol are highly suggestive of a biochemical diagnosis of SLO.

Special Instructions

- Biochemical Genetics Patient Information

Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type

Plasma

Necessary Information

Biochemical Genetics Patient Information (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Required

Supplies: Sarstedt Aliquot Tube, 5 mL (T914)
Collection Container/Tube:
Preferred: Green top (sodium or lithium heparin)
Acceptable: Lavender top (EDTA), pearl white top (EDTA plasma gel), yellow top (ACD solution A or B)
Submission Container/Tube: Plastic vial
Specimen Volume: 0.5 mL plasma
Collection Instructions:
1. Centrifuge and aliquot plasma into plastic vial.

2. Send plasma frozen.

Forms

1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

Plasma: 0.1 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen (preferred)	92 days	
	Ambient	14 days	
	Refrigerated	28 days	

Clinical & Interpretive

Clinical Information

Cholesterol plays an essential role in many cellular and developmental processes. In addition to its role as a membrane lipid, it is the precursor to numerous molecules that play important roles in cell growth and differentiation, protein glycosylation, and signaling pathways. The biosynthesis of cholesterol and its subsequent conversion to other essential compounds is complex, involving a number of intermediates and enzymes. Disorders that result from a deficiency of these enzymes lead to an accumulation of specific intermediates and inhibit the formation of important biomolecules. Clinical findings common to cholesterol biosynthesis disorders include congenital skeletal malformations, dysmorphic facial features, psychomotor retardation, and failure to thrive.

Smith-Lemli-Opitz syndrome (SLO) is an autosomal recessive disorder caused by variants in the *DHCR7* gene leading to a deficiency of the 7-dehydrocholesterol reductase enzyme. It is characterized biochemically by markedly increased plasma concentrations of 7-dehydrocholesterol and 8-dehydrocholesterol levels. Clinical features can include microcephaly, growth retardation, developmental delay, dysmorphic facial features, cleft palate, limb abnormalities (especially 2-3 syndactyly of the toes and postaxial polydactyly), and heart and kidney malformations. However, the clinical spectrum ranges from mild to severe with some mildly affected individuals presenting with only 2-3 toe syndactyly and mild cognitive impairment. The reported incidence is between 1 in 10,000 and 1 in 60,000, but it may be more prevalent due to underdiagnoses of mildly affected individuals.

Fetal fentanyl syndrome is a newly described syndrome in infants who were exposed to high levels of fentanyl prenatally and presented with clinical features similar to SLO. These features include growth deficiency, microcephaly, congenital anomalies, and distinctive facial features. In addition to clinical findings being suggestive of SLO, biochemical studies demonstrated elevated 7-dehydrocholesterol and/or 8-dehydrocholesterol levels after birth that normalized over time.

Other disorders of cholesterol biosynthesis, including desmosterolosis (desmosterol reductase deficiency) and sitosterolemia, may present with similar manifestations. These disorders can be detected biochemically by performing a quantitative profile of plasma sterols (STER / Sterols, Plasma).

Reference Values

7-dehydrocholesterol
< or =2.0 mg/L

8-dehydrocholesterol
< or =0.3 mg/L

Interpretation

Elevated plasma concentrations of 7-dehydrocholesterol (7-DHC) and 8-dehydrocholesterol are highly suggestive of a biochemical diagnosis of Smith-Lemli-Opitz (SLO) syndrome.

Transient elevations of 7-dehydrocholesterol (7-DHC) or 8-dehydrocholesterol in newborns in the setting of known prenatal fentanyl exposure can be associated with fetal fentanyl syndrome.

Mild elevations of these cholesterol precursors can be detected in patients with hypercholesterolemia and patients treated with some antipsychotic or antidepressant medications, including haloperidol, aripiprazole, and trazodone. However, the 7-DHC to cholesterol ratio is typically elevated only in patients with SLO syndrome.

Cautions

On very rare occasions, 7-dehydrocholesterol (7-DHC) is not elevated in patients with Smith-Lemli-Opitz (SLO) syndrome.

Cholesterol screening tests are unreliable for diagnosis for SLO syndrome.

Some antipsychotic or antidepressant medications, such as aripiprazole and trazodone, cause false elevations in 7-DHC.

Clinical Reference

1. Donoghue SE, Pitt JJ, Boneh A, White SM. Smith-Lemli-Opitz syndrome: clinical and biochemical correlates. J Pediatr Endocrinol Metab. 2018;31(4):451-459
2. Nowaczyk MJM, Wassif CA. Smith-Lemli-Opitz syndrome. In: Adam MP, Mirzaa GM, Pagon RA, et al., eds. GeneReviews [Internet]. University of Washington, Seattle; 1998. Updated January 30, 2020.Accessed November 02, 2023. Available at www.ncbi.nlm.nih.gov/books/NBK1143/
3. Wadman E, Fernandes E, Muss C, et al. A novel syndrome associated with prenatal fentanyl exposures. Genet Med Open. 2023;1(1):1-7
4. Khoja I, Pino G, Peck D, et al. P051: Impact of prenatal fentanyl exposure on sterol analysis for Smith-Lemli-Opitz syndrome in newborns. Genet Med Open. 2025;3(S2):28
5. Hall P, Michels V, Gavrilov D, et al. Aripiprazole and trazodone cause elevations of 7-dehydrocholesterol in the absence of Smith-Lemli-Opitz syndrome. Mol Genet Metab. 2013;110(1-2):176-178

6. Genaro-Mattos TC, Tallman KA, Allen LB, et al. Dichlorophenyl piperazines, including a recently-approved atypical antipsychotic, are potent inhibitors of DHCR7, the last enzyme in cholesterol biosynthesis. Toxicol Appl Pharmacol. 2018;349:21-28. doi:10.1016/j.taap.2018.04.029

Performance

Method Description

The plasma specimen is hydrolyzed and then extracted followed by evaporation to dryness under nitrogen. The sterols are derivatized and then analyzed using selected ion-monitoring electron impact gas chromatography mass spectrometry to quantitate 7-dehydrocholesterol and 8-dehydrocholesterol.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Tuesday, Friday

Report Available

3 to 7 days

Specimen Retention Time

1 month

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value

SLO	Smith-Lemli-Opitz Scrn, P	73852-6

Result ID	Test Result Name	Result LOINC® Value
29974	Reviewed By	18771-6
29972	Interpretation	59462-2
610625	7-Dehydrocholesterol	33275-9
610626	8-Dehydrocholesterol	34671-8