

Overview

Useful For

Biomarker for peroxisomal biogenesis disorders, such as Zellweger syndrome and single enzyme defects of bile acid synthesis, including D-bifunctional protein deficiency and alpha methyl CoA racemases

Monitoring patients receiving bile acid therapy, such as cholic acid, for liver disease due to peroxisomal biogenesis disorders or single enzyme defects in bile acid synthesis

Testing Algorithm

For more information see:

[-Bile Acid-Associated Tests Ordering Guide](#)

[-Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy](#)

Special Instructions

- [Bile Acid-Associated Tests Ordering Guide](#)
- [Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy](#)

Highlights

This is a serum test for the measurement of C27 bile acids, a diagnostic marker for peroxisomal biogenesis disorders and single enzyme defects of bile acid synthesis, including D-bifunctional protein deficiency and alpha methyl CoA racemase deficiency.

This test can also be used for monitoring of treatment efficacy.

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Serum

Ordering Guidance

For assessment of general liver dysfunction in adults or diagnosis or monitoring of intrahepatic cholestasis of pregnancy, order BAFS / Bile Acids, Fractionated and Total, Serum.

Specimen Required

Patient Preparation: Patient must be fasting for 12 to 14 hours.

Collection Container/Tube:

Preferred: Red top

Acceptable: Serum gel

Submission Container/Tube: Plastic vial

Specimen Volume: 0.5 mL

Collection Instructions: Centrifuge and aliquot serum into a plastic vial.

Forms

[If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request \(T798\)](#) with the specimen.

Specimen Minimum Volume

0.3 mL

Reject Due To

| | |
|-----------------|----|
| Gross hemolysis | OK |
| Gross lipemia | OK |

Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---------------|--------------------------|---------|-------------------|
| Serum | Refrigerated (preferred) | 90 days | |
| | Ambient | 90 days | |
| | Frozen | 90 days | |

Clinical & Interpretive

Clinical Information

Bile acids are formed in the liver from cholesterol, conjugated primarily to glycine and taurine, stored and concentrated in the gallbladder, and secreted into the intestine after the ingestion of a meal. In the intestinal lumen, the bile acids serve to emulsify ingested fats and thereby promote digestion. During the absorptive phase of digestion, approximately 90% of the bile acids are reabsorbed.

The efficiency of the hepatic clearance of bile acids from portal blood maintains serum concentrations at low levels in normal persons. An elevated fasting level, due to impaired hepatic clearance, is a sensitive indicator of liver disease. Following meals, serum bile acid levels have been shown to increase only slightly in normal persons, but markedly in patients with various liver diseases, including cirrhosis, hepatitis, cholestasis, portal-vein thrombosis, Budd-Chiari syndrome, cholangitis, Wilson disease, and hemochromatosis. No increase in bile acids will be noted in patients with intestinal malabsorption. Metabolic hepatic disorders involving organic anions (eg, Gilbert disease, Crigler-Najjar syndrome, and Dubin-Johnson syndrome) do not cause abnormal serum bile acid concentrations.

This bile acid test for peroxisomal disorders measures concentrations of C27 bile acids, which are diagnostic markers for peroxisomal biogenesis disorders such as Zellweger syndrome and single enzyme defects of bile acid synthesis such as

D-bifunctional protein deficiency and alpha methyl-CoA racemase deficiency. Elevated levels of C27 bile acids may enable diagnosis of peroxisomal biogenesis disorders and bile acid synthesis defects in children with liver cholestasis. Treatment for peroxisomal biogenesis disorders and bile acid synthesis defects with cholic acid is available. Measurement of C27 bile acids before and during treatment with bile acid therapy such as cholic acid can assist with monitoring of treatment efficacy.

Reference Values

Dihydroxycholestanic acid: < or =0.10 nmol/mL

Trihydroxycholestanic acid: < or =1.30 nmol/mL

Total cholic acid: < or =5.00 nmol/mL

Total chenodeoxycholic acid: < or =6.00 nmol/mL

Total ursodeoxycholic acid: < or =2.00 nmol/mL

Total bile acids: < or =19.00 nmol/mL

Interpretation

Increases in serum C27 bile acids are seen in patients with peroxisomal biogenesis disorders such as Zellweger syndrome or single enzyme defects of bile acid synthesis such as D-bifunctional protein deficiency and alpha methyl CoA racemases. Total bile acids are metabolized in the liver and can serve as a marker for normal liver function. The values of 2 bile acid precursors, dihydroxycholestanic acid and trihydroxycholestanic acid, will be reported, along with total cholic acid, total chenodeoxycholic acid, total ursodeoxycholic acid, and total bile acids. No interpretive report will be provided.

Cautions

Bile acid concentrations in serum may be elevated post meal or due to bile acid therapy, such as cholic acid, deoxycholic acid, and ursodeoxycholic acid.

Clinical Reference

1. Johnson DW, ten Brink HJ, Schuit RC, Jakobs C. Rapid and quantitative analysis of unconjugated C(27) bile acids in plasma and blood samples by tandem mass spectrometry. *J Lipid Res.* 2001;42(1):9-16
2. Bootsma AH, Overmars H, van Rooij A, et al. Rapid analysis of conjugated bile acids in plasma using electrospray tandem mass spectrometry: application for selective screening of peroxisomal disorders. *J Inherit Metab Dis.* 1999;22(3):307-310
3. Ferdinandusse S, Jimenez-Sanchez G, Koster J, et al. A novel bile acid biosynthesis defect due to a deficiency of peroxisomal ABCD3. *Hum Mol Genet.* 2015;24(2):361-370
4. Heubi JE, Setchell KDR, Bove KE. Inborn errors of bile acid metabolism. *Clin Liver Dis.* 2018;22(4):671-687. doi: 10.1016/j.cld.2018.06.006
5. Sundaram SS, Bove KE, Lovell MA, Sokol RJ. Mechanisms of disease: inborn errors of bile acid synthesis. *Nat Clin Pract Gastroenterol Hepatol.* 2008;5(8):456-468
6. Wanders RJA, Rizzo WB. Inborn errors of peroxisome biogenesis and function. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. *Pediatric Endocrinology and Inborn Errors of Metabolism.* 2nd ed. McGraw-Hill Medical Division; 2017:427-446
7. Fischler B, Eggersten G, Bjorkhem I. Genetic defects in synthesis and transport of bile acids. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. *Pediatric Endocrinology and Inborn Errors of Metabolism.* McGraw-Hill Medical Division; 2017:447-460

Performance

Method Description

Bile acid concentrations in serum are measured by liquid chromatography tandem mass spectrometry stable isotope dilution analysis. Serum is mixed with isotopically labeled internal standards of selected bile acids and then subjected to protein precipitation. Sample preparation is semiautomated using a liquid handler. Reverse-phase liquid chromatography is performed using mobile phases to separate free bile acids, their respective tauro- and glyco-conjugates, and 2 bile acid precursors.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

3 to 5 days

Specimen Retention Time

1 Month

Performing Laboratory Location

Rochester

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. This test 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 |
|---------|-----------------------------------|--------------------|
| BAIPD | Bile Acids for Peroxisomal D/O, S | 43130-4 |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|----------------------------|---------------------|
| 41446 | Dihydroxycholestanic Acid | 53479-2 |
| 41447 | Trihydroxycholestanic Acid | 38188-9 |

| | | |
|-------|-----------------------------|---------|
| 41448 | Total Cholic Acid | 30518-5 |
| 41449 | Total Chenodeoxycholic Acid | 30519-3 |
| 41450 | Total Ursodeoxycholic Acid | 55159-8 |
| 41451 | Total Bile Acids | 14628-2 |