Test Definition: BAIPD

Bile Acids for Peroxisomal D/O, S

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 racemaces

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

Highlights

Bile acids for peroxisomal disorders is a serum test measuring C27 bile acids, which are 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.

Bile acids for peroxisomal disorders can also be used for monitoring of treatment efficacy.

Testing Algorithm

See Ordering Guide: Bile Acid-Associated Tests in Special Instructions.

See Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy in Special Instructions.

For more information, see Newborn Screening Act Sheet X-linked Adrenoleukodystrophy: Increased Very Long Chain Fatty Acids in Special Instructions.

Special Instructions

- Newborn Screening Act Sheet X-linked Adrenoleukodystrophy: Increased Very Long Chain Fatty Acids
- Ordering Guide: Bile Acid-Associated Tests
- Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Serum

Advisory Information

Do not use for assessment of general liver dysfunction in adults or diagnosis or monitoring of intrahepatic cholestasis of pregnancy (see BAFS / Bile Acids, Fractionated and Total, Serum).

Specimen Required

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

Collection Container/Tube:

Preferred: Red top

Acceptable: Serum gel

Submission Container/Tube: Plastic vial

Specimen Volume: 0.5 mL

Forms

If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume

0.3 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Other	Nonfasting specimen

Specimen Stability Information

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

Clinical and 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 levels 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

Dihydroxycholestanoic Acid < or =0.10 nmol/mL

Trihydroxycholestanoic 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 racemaces. 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, dihydroxycholestanoic acid and trihydroxycholestanoic 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 postmeal and due to bile acid therapy, such as cholic acid, deoxycholic acid, and ursodeoxycholic acid.

Clinical Reference

1. Johnson DW, Brink HJ, Schuit RC, et al: Rapid and quantitative analysis of unconjugated C27 bile acids in plasma and blood samples by tandem mass spectrometry. Journ Lipid Res 2001;42:9-16

2. Bootsma AH, Overmars H, Van Rooiji A, et al: Rapid analysis of conjugated bile acids in plasma using electrospray tandem mass spectrometry: Application for selective screening of peroxisomal disorders. J Inher Metab Dis 1999;22:307-310

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;24:361-370

4. Heubi J, Setchell KDR, Bove KE: Inborn Errors of Bile Acid Metabolism. Sem Liver Dis 2007;27:282-294

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: Inborn Errors of Peroxisome Biogenesis and Function. In Pediatric Endocrinology and Inborn Errors of Metabolism. Edited by K Sarafoglou, GF Hoffmann, KS Roth, New York, NY, McGraw-Hill Medical Division, 2009, pp 323-337

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) and Time(s) Test Performed

Monday, Wednesday, Thursday, Friday; 8 a.m.

Analytic Time

2 days (not reported on Saturday or Sunday)

Maximum Laboratory Time

7 days

Specimen Retention Time

1 Month

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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 U.S. 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	In Process

Result ID	Test Result Name	Result LOINC Value
41446	Dihydroxycholestanoic Acid	53479-2
41447	Trihydroxycholestanoic Acid	38188-9

Result ID	Test Result Name	Result LOINC Value
41448	Total Cholic Acid	30518-5
41449	Total Chenodeoxycholic Acid	30519-3
41450	Total Ursodeoxycholic Acid	55159-8
41451	Total Bile Acids	14628-2