
Lab Dept: Chemistry

Test Name: LONG CHAIN FATTY ACIDS

General Information

Lab Order Codes: LFAT

Synonyms: Fatty Acid Profile, Peroxisomal (C22-C26), Serum; Phytanic Acid; Pristanic Acid; Very Long Chain Fatty Acids

CPT Codes: 82726 – Very long chain fatty acids

Test Includes: C22:0, C24:0, C26:0, C24:0/C22:0 Ratio C26:0/C22:0 Ratio, Pristanic Acid, Phytanic Acid and Pristanic/Phytanic Acid Ratio

Logistics

Test Indications: Evaluating patients with possible peroxisomal disorders, including peroxisomal biogenesis disorders, X-linked adrenoleukodystrophy, and Refsum's disease. As an aid in the assessment of peroxisomal function.

Lab Testing Sections: Chemistry – Sendouts

Referred to: Mayo Medical Laboratories (MML Test: POX)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 4 - 7 days, test set up Monday - Friday

Special Instructions: Include information regarding treatment, family history and tentative diagnosis. Refer to [Patient Preparation](#).

Specimen

Specimen Type: Blood

Container: SST (Marble, gold or red top tube)

Draw Volume: 1.5 mL (Minimum: 0.5 mL) blood

Processed Volume: 0.5 mL (Minimum: 0.15 mL) serum

Collection: Routine blood collection

Special Processing: Lab staff: Centrifuge specimen within 45 minutes of collection, remove serum aliquot into a screw-capped round bottom plastic vial. Store and ship at frozen temperatures. Include information regarding treatment, family history, and tentative diagnosis. Must include patient's age.

Patient Preparation: Patient must not consume any alcohol for 24 hours before the specimen is drawn. Overnight (12 - 14 hour) fast recommended.

Sample Rejection: Gross lipemia; mislabeled or unlabeled specimens

Interpretive

Reference Range:

C22:0	≤96.3 nmol/mL
C24:0	≤91.4 nmol/mL
C26:0	≤1.30 nmol/mL
C24:0/C22:0 Ratio	≤1.39 ratio
C26:0/C22:0 Ratio	≤0.023 ratio
Pristanic Acid (nmol/mL):	
0 – 4 months	≤0.60
5 – 8 months	≤0.84
9 – 12 months	≤0.77
13 – 23 months	≤1.47
≥24 months	≤2.98
Phytanic Acid (nmol/mL):	
0 – 4 months	≤5.28
5 – 8 months	≤5.70
9 – 12 months	≤4.40
13 – 23 months	≤8.62
≥24 months	≤9.88

Pristanic/Phytanic Acid Ratio:	
0 – 4 months	≤0.35
5 –8 months	≤0.28
9 – 12 months	≤0.23
13 – 23 months	≤0.24
≥24 months	≤0.39
<p>Interpretation: Reports include concentrations of C22:0, C24:0, C26:0 species, phytanic acid and pristanic acid and calculated C24:0/C22:0, C26:0/C22:0, and phytanic acid/pristanic acid ratios. When no significant abnormalities are detected, a simple descriptive interpretation is provided.</p> <p>A profile of elevated phytanic acid, low-normal pristanic acid, and normal very long-chain fatty acids is suggestive of Refsum disease (phytanic acid oxidase deficiency); however, serum phytanic acid concentration may also be increased in disorders of peroxisomal biogenesis and should be considered in the differential diagnosis of peroxisomal disorders.</p> <p>If results are suggestive of hemizygoty for X-linked adrenoleukodystrophy, included will be the calculated value of a discriminating function used to more accurately segregate hemizygous individuals from normal controls.</p> <p>Positive test results could be due to genetic or nongenetic condition. Additional confirmatory testing would be required.</p>	

Critical Values:

N/A

Limitations:

In rare instances, patients with X-linked adrenoleukodystrophy (X-ALD) may have only minimally elevated values, 15-20% of women heterozygous for X-ALD have normal plasma very long-chain fatty acid levels.

False-positive results may occur with nonfasting specimens.

Methodology:

Gas Chromatography/Mass Spectrometry (GC-MS), Stable Isotope Dilution Analysis

References:

[Mayo Medical Laboratories](#) January 2018

Updates:

9/27/2012: EDTA and Heparin removed as alternated tube types.