
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: 81369/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 venipuncture

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](#) August 2016

Updates:

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