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 peroxismal 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

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.

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.

If results are suggestive of hemizygosity for X-linked adrenoleukodystrophy, included will be the calculated value of a discriminating function used to more accurately segregate hemizygous individuals from normal controls.

Positive test results could be due to genetic or nongenetic condition. Additional confirmatory testing would be required.

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.