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MAYO CLINIC LABORATORIES

Test Definition: FAPCP

Fatty Acid Profile, Comprehensive (C8-C26), Serum

Overview

Useful For

Monitoring patients undergoing diet therapy for mitochondrial or peroxisomal disorders (possibly inducing essential fatty acid deficiency in response to restricted fat intake) using serum specimens

Monitoring treatment of essential fatty acid deficiency

Monitoring the response to provocative tests (fasting tests, loading tests)

Genetics Test Information

This test is a comprehensive profile that provides information regarding mitochondrial and peroxisomal fatty acid metabolism as well as the patient's nutritional status.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Serum

Ordering Guidance

This test is **not** the recommended initial screening test for evaluating patients with possible peroxisomal disorders, single-enzyme defects of peroxisomal metabolism such as X-linked adrenoleukodystrophy, or peroxisomal biogenesis disorders (Zellweger syndrome spectrum). For these purposes, the preferred tests are either POXP / Fatty Acid Profile, Peroxisomal (C22-C26), Plasma or POX / Fatty Acid Profile, Peroxisomal (C22-C26), Serum.

Necessary Information

1. Patient's age is required.

2. Include information regarding treatment, family history, and tentative diagnosis.

Specimen Required

Patient Preparation:

1. Fasting:
   a. **For nutritional assessment: 12 hours, required**
   b. For patients with a suspected fatty acid oxidation disorder, prolonged fasting is contraindicated. Collect as close to

the patient's next scheduled meal/feeding as possible.
2. Patient **must not** consume any alcohol for 24 hours before the specimen collection.
Supplies: Sarstedt Aliquot Tube, 5 mL (T914)
Collection Container/Tube:
Preferred: Serum gel
Acceptable: Red top
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
0.15 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	Reject
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Frozen (preferred)	92 days	
	Refrigerated	72 hours	

Clinical & Interpretive

Clinical Information
Fatty Acid Deficiency/Excess:
Fats are important sources of energy for tissues and for the function and integrity of cellular membranes. Deficiencies are commonly caused by inadequate dietary intake of lipids due to an unbalanced diet, long-term parenteral nutrition, or by intestinal malabsorption. Linoleic acid, an omega-6 fatty acid, and alpha-linolenic acid, an omega-3 fatty acid, are considered essential fatty acids in that they cannot be made by the body and are essential components of the diet.

The major clinical manifestations associated with essential fatty acid deficiency (EFAD) include dermatitis, increased water permeability of the skin, increased susceptibility to infection, and impaired wound healing. Biochemical abnormalities may be detected before the onset of recognizable clinical manifestations. EFAD can be detected by diminished levels of the essential fatty acids, linoleic and alpha-linolenic acid, as well as by increases in the triene:tetraene ratio.

Excess dietary fatty acids have been linked to the onset of cardiovascular disease. Elevated levels of linoleic acid can contribute to overproduction of the proinflammatory 2-series local hormones. The Academy of Nutrition and Dietetics recommends that dietary fat for the healthy adult population should provide 20% to 35% of energy, with an increased consumption of n-3 polyunsaturated fatty acids and limited intake of saturated and trans fats.(1)

Fatty Acid Oxidation Disorders:
Mitochondrial beta-oxidation is the main source of energy to skeletal and heart muscle during periods of fasting. When the body's supply of glucose is depleted, fatty acids are mobilized from adipose tissue and converted to ketone bodies through a series of steps providing an alternate source of energy. Deficient enzymes at any step in this pathway prevent the production of energy during periods of physiologic stress such as fasting or intercurrent illness.

The major clinical manifestations associated with fatty acid oxidation (FAO) disorders include hypoketotic hypoglycemia, liver disease and failure, skeletal myopathy, dilated/hypertrophic cardiomyopathy, and sudden unexpected death in early life. Signs and symptoms may vary greatly in severity, combination, and age of presentation. Life-threatening episodes of metabolic decompensation frequently occur after periods of inadequate calorie intake or intercurrent illness. When properly diagnosed, patients with FAO disorders respond favorably to fasting avoidance, diet therapy, and aggressive treatment of intercurrent illnesses, with significant reduction of morbidity and mortality.

Disease-specific characteristic patterns of metabolites from FAO disorders are detectable in blood, bile, urine, and cultured fibroblasts of living and many deceased individuals. Quantitative determination of C8-C18 fatty acids is an important element of the work-up and differential diagnosis of candidate patients. Fatty acid profiling can detect quantitatively modest, but nevertheless significant, abnormalities even when patients are asymptomatic and under dietary treatment. Confirmatory testing for many of the FAO disorders is also available. For more information see FAO / Fatty Acid Oxidation Probe Assay, Fibroblast Culture and HFAOP / Fatty Acid Oxidation Gene Panel, Varies.

Peroxisomal Disorders:
Peroxisomes are organelles present in all human cells except mature erythrocytes. They carry out essential metabolic functions including beta-oxidation of very long-chain fatty acids (VLCFA), alpha-oxidation of phytanic acid, and biosynthesis of plasmalogen and bile acids. Peroxisomal disorders include disorders of peroxisomal biogenesis with defective assembly of the entire organelle, and single peroxisomal enzyme/transporter defects where the organelle is intact but a specific function is disrupted. Peroxisomal beta-oxidation of VLCFA is impaired in all disorders of peroxisomal biogenesis and in selected single enzyme deficiencies, particularly X-linked adrenoleukodystrophy, resulting in elevated concentrations of VLCFA in serum or plasma. POXP / Fatty Acid Profile, Peroxisomal (C22-C26), Plasma or POX / Fatty Acid Profile, Peroxisomal (C22-C26), Serum is the preferred screening test for evaluating patients with possible peroxisomal disorders, single-enzyme defects of peroxisomal metabolism such as X-linked adrenoleukodystrophy, or peroxisomal biogenesis disorders (Zellweger syndrome spectrum). Confirmatory testing for X-linked adrenoleukodystrophy via molecular genetic analysis is available; see ABCD1 / X-Linked Adrenoleukodystrophy (XALD), *ABCD1* Gene Sequencing with Deletion/Duplication, Varies.

Reference Values

Octanoic Acid, C8:0
<1 year: 7-63 nmol/mL
1-17 years: 9-41 nmol/mL

> or =18 years: 8-47 nmol/mL

Decenoic Acid, C10:1
<1 year: 0.8-4.8 nmol/mL
1-17 years: 1.6-6.6 nmol/mL
> or =18 years: 1.8-5.0 nmol/mL

Decanoic Acid, C10:0
<1 year: 2-62 nmol/mL
1-17 years: 3-25 nmol/mL
> or =18 years: 2-18 nmol/mL

Lauroleic Acid, C12:1
<1 year: 0.6-4.8 nmol/mL
1-17 years: 1.3-5.8 nmol/mL
> or =18 years: 1.4-6.6 nmol/mL

Lauric Acid, C12:0
<1 year: 6-190 nmol/mL
1-17 years: 5-80 nmol/mL
> or =18 years: 6-90 nmol/mL

Tetradecadienoic Acid, C14:2
<1 year: 0.3-6.5 nmol/mL
1-17 years: 0.2-5.8 nmol/mL
> or =18 years: 0.8-5.0 nmol/mL

Myristoleic Acid, C14:1
<1 year: 1-46 nmol/mL
1-17 years: 1-31 nmol/mL
> or =18 years: 3-64 nmol/mL

Myristic Acid, C14:0
<1 year: 30-320 nmol/mL
1-17 years: 40-290 nmol/mL
> or =18 years: 30-450 nmol/mL

Hexadecadienoic Acid, C16:2
<1 year: 4-27 nmol/mL
1-17 years: 3-29 nmol/mL
> or =18 years: 10-48 nmol/mL

Hexadecenoic Acid, C16:1w9
<1 year: 21-69 nmol/mL

1-17 years: 24-82 nmol/mL
> or =18 years: 25-105 nmol/mL

Palmitoleic Acid, C16:1w7
<1 year: 20-1,020 nmol/mL
1-17 years: 100-670 nmol/mL
> or =18 years: 110-1,130 nmol/mL

Palmitic Acid, C16:0
<1 year: 720-3,120 nmol/mL
1-17 years: 960-3,460 nmol/mL
> or =18 years: 1,480-3,730 nmol/mL

Gamma-Linolenic Acid, C18:3w6
<1 year: 6-110 nmol/mL
1-17 years: 9-130 nmol/mL
> or =18 years: 16-150 nmol/mL

Alpha-Linolenic Acid, C18:3w3
<1 year: 10-190 nmol/mL
1-17 years: 20-120 nmol/mL
> or =18 years: 50-130 nmol/mL

Linoleic Acid, C18:2w6
< or =31 days: 350-2,660 nmol/mL
32 days-11 months: 1,000-3,300 nmol/mL
1-17 years: 1,600-3,500 nmol/mL
> or =18 years: 2,270-3,850 nmol/mL

Oleic Acid, C18:1w9
<1 year: 250-3,500 nmol/mL
1-17 years: 350-3,500 nmol/mL
> or =18 years: 650-3,500 nmol/mL

Vaccenic Acid, C18:1w7
<1 year: 140-720 nmol/mL
1-17 years: 320-900 nmol/mL
> or =18 years: 280-740 nmol/mL

Stearic Acid, C18:0
<1 year: 270-1,140 nmol/mL
1-17 years: 280-1,170 nmol/mL
> or =18 years: 590-1,170 nmol/mL

EPA, C20:5w3
<1 year: 2-60 nmol/mL
1-17 years: 8-90 nmol/mL
> or =18 years: 14-100 nmol/mL

Arachidonic Acid, C20:4w6
<1 year: 110-1,110 nmol/mL
1-17 years: 350-1,030 nmol/mL
> or =18 years: 520-1,490 nmol/mL

Mead Acid, C20:3w9
< or =31 days: 8-60 nmol/mL
32 days-11 months: 3-24 nmol/mL
> or =1 year: 7-30 nmol/mL

Homo-Gamma-Linolenic Acid, C20:3w6
<1 year: 30-170 nmol/mL
1-17 years: 60-220 nmol/mL
> or =18 years: 50-250 nmol/mL

Arachidic Acid, C20:0
<1 year: 30-120 nmol/mL
1-17 years: 30-90 nmol/mL
> or =18 years: 50-90 nmol/mL

DHA, C22:6w3
<1 year: 10-220 nmol/mL
1-17 years: 30-160 nmol/mL
> or =18 years: 30-250 nmol/mL

DPA, C22:5w6
<1 year: 3-70 nmol/mL
1-17 years: 10-50 nmol/mL
> or =18 years: 10-70 nmol/mL

DPA, C22:5w3
<1 year: 6-110 nmol/mL
1-17 years: 30-270 nmol/mL
> or =18 years: 20-210 nmol/mL

DTA, C22:4w6
<1 year: 2-50 nmol/mL
1-17 years: 10-40 nmol/mL
> or =18 years: 10-80 nmol/mL

Docosenoic Acid, C22:1
<1 year: 2-20 nmol/mL
> or =1 year: 4-13 nmol/mL

Docosanoic Acid, C22:0
0.0-96.3 nmol/mL

Nervonic Acid, C24:1
<1 year: 30-150 nmol/mL
1-17 years: 50-130 nmol/mL
> or =18 years: 60-100 nmol/mL

Tetracosanoic Acid, C24:0
0.0-91.4 nmol/mL

Hexacosenoic Acid, C26:1
<1 year: 0.2-2.1 nmol/mL
> or =1 year: 0.3-0.7 nmol/mL

Hexacosanoic Acid, C26:0
0.00-1.30 nmol/mL

Pristanic Acid, C15:0(CH3)4
< or =4 months: 0.00-0.60 nmol/mL
5-8 months: 0.00-0.84 nmol/mL
9-12 months: 0.00-0.77 nmol/mL
13-23 months: 0.00-1.47 nmol/mL
> or =2 years: 0.00-2.98 nmol/mL

Phytanic Acid, C16:0(CH3)4
< or =4 months: 0.00-5.28 nmol/mL
5-8 months: 0.00-5.70 nmol/mL
9-12 months: 0.00-4.40 nmol/mL
13-23 months: 0.00-8.62 nmol/mL
> or =2 years: 0.00-9.88 nmol/mL

Triene/Tetraene Ratio
< or =31 days: 0.017-0.083
32 days-17 years: 0.013-0.050
> or =18 years: 0.010-0.038

Total Saturated Acid
<1 year: 1.2-4.6 mmol/L

1-17 years: 1.4-4.9 mmol/L
> or =18 years: 2.5-5.5 mmol/L

Total Monounsaturated Acid
<1 year: 0.3-4.6 mmol/L
1-17 years: 0.5-4.4 mmol/L
> or =18 years: 1.3-5.8 mmol/L

Total Polyunsaturated Acid
<1 year: 1.1-4.9 mmol/L
1-17 years: 1.7-5.3 mmol/L
> or =18 years: 3.2-5.8 mmol/L

Total w3
<1 year: 0.0-0.4 mmol/L
1-17 years: 0.1-0.5 mmol/L
> or =18 years: 0.2-0.5 mmol/L

Total w6
<1 year: 0.9-4.4 mmol/L
1-17 years: 1.6-4.7 mmol/L
> or =18 years: 3.0-5.4 mmol/L

Total Fatty Acids
<1 year: 3.3-14.0 mmol/L
1-17 years: 4.4-14.3 mmol/L
> or =18 years: 7.3-16.8 mmol/L

Interpretation

An increased triene:tetraene ratio is consistent with essential fatty acid deficiency.

Fatty acid oxidation disorders are recognized on the basis of disease-specific patterns that are correlated to the results of other investigations in plasma (carnitine, acylcarnitines) and urine (organic acids, acylglycines).

Increased concentrations of serum very long-chain fatty acids (VLCFA) C24:0 and C26:0 are seen in peroxisomal disorders, X-linked adrenoleukodystrophy, adrenomyeloneuropathy, and Zellweger syndrome (cerebrohepatorenal syndrome).

Increased concentrations of serum phytanic acid (along with normal pristanic acid concentrations) are seen in Refsum disease (phytanase deficiency). Serum phytanic acid concentration also may be increased in other peroxisomal disorders and, when combined with the VLCFA, pristanic acid and pipecolic acid allow differential diagnosis of peroxisomal disorders.

Cautions

For nutritional assessment, a 12- to 14-hour fast is required; however, infants or persons suspected of having a fatty acid oxidation disorder should not fast before testing due to the possibility of acute metabolic decompensation. Instead, collect the specimen after the longest fast possible, just before feeding. In the case of a patient on total parenteral nutrition, specimen can be collected as normal.

Clinical Reference

1. Vannice G, Rasmussen H. Position of the Academy of Nutrition and Dietetics: Dietary fatty acids for healthy adults. J Acad Nutr Diet. 2014;114(1):136-153. doi:10.1016/j.jand.2013.11.001
2. Rinaldo P, Matern D, Bennett MJ. Fatty acid oxidation disorders. Ann Rev Physiol. 2002;64:477-502
3. Jeppesen PB, Christensen MS, Hoy CE, Mortensen PB. Essential fatty acid deficiency in patients with severe fat malabsorption. Am J Clin Nutr. 1997;65(3):837-843
4. Spector AA, Kim HY. Discovery of essential fatty acids. J. Lipid Res. 2015;56(1):11-215. VerHoeven NM, Jakobs C. Human metabolism of phytanic acid and pristanic acid. Prog in Lipid Res. 2001;40(6):453-466
6. Luszczki E, Boakye F, Zielinska M, Deren K, et al. Vegan diet: nutritional components, implementation, and effects on adults' health. Front Nutr. 2023 9;10:1294497. doi:10.3389/fnut.2023.1294497

Performance

Method Description

Quantitation of fatty acids of specific chain lengths is performed as follows: a 2-step, acid-base hydrolysis is followed by hexane extraction and derivatization with pentafluorobenzyl bromide. Separation and detection are accomplished by capillary gas chromatography electron-capture negative ion-mass spectrometry. Quantitation is based on analysis in the selected ion-monitoring mode by using 13 stable isotope-labeled internal standards.(Lagerstedt SA, Hinrichs DR, Batt SM, Magera MJ, Rinaldo P, McConnell JP. Quantitative determination of plasma C8-C26 total fatty acids for the biochemical diagnosis of nutritional and metabolic disorders. Mol Genet Metab. 2001;73[1]:38-45, Gramlich L, Ireton-Jones C, Miles JM, Morrison M, Pontes-Arruda A. Essential fatty acid requirements and intravenous lipid emulsions. JPEN J Parenter Enteral Nutr. 2019;43[6]:697-707)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

3 to 5 days

Specimen Retention Time

2 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82725

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
FAPCP	Fatty Acid Profile, Comprehensive,S	43674-1

Result ID	Test Result Name	Result LOINC® Value
16965	Octanoic Acid, C8:0	35145-2
16966	Decenoic Acid, C10:1	35147-8
16967	Decanoic Acid, C10:0	35146-0
16968	Lauroleic Acid, C12:1	35151-0
16969	Lauric Acid, C12:0	35150-2
16970	Tetradecadienoic Acid, C14:2	35148-6
16971	Myristoleic Acid, C14:1	35158-5
16972	Myristic Acid, C14:0	35157-7
16973	Hexadecadienoic Acid, C16:2	35154-4
16974	Hexadecenoic Acid, C16:1w9	35155-1
16975	Palmitoleic Acid, C16:1w7	35162-7
16976	Palmitic Acid, C16:0	35161-9
16977	g-Linolenic Acid, C18:3w6	35163-5
16978	a-Linolenic Acid, C18:3w3	35164-3
16979	Linoleic Acid, C18:2w6	35165-0
16980	Oleic Acid, C18:1w9	35166-8
16981	Vaccenic Acid, C18:1w7	35167-6
16982	Stearic Acid, C18:0	35149-4
16983	EPA, C20:5w3	35173-4
16984	Arachidonic Acid, C20:4w6	35168-4
16985	Mead Acid, C20:3w9	35172-6
16986	h-g-Linolenic Acid, C20:3w6	35171-8

16987	Arachidic Acid, C20:0	35169-2
16988	DHA, C22:6w3	35174-2
16989	DPA, C22:5w6	35181-7
16990	DPA, C22:5w3	35180-9
16991	DTA, C22:4w6	35182-5
16992	Docosenoic Acid, C22:1	35160-1
16993	Docosanoic Acid, C22:0	30194-5
16994	Nervonic Acid, C24:1w9	35170-0
16995	Tetracosanoic Acid, C24:0	30195-2
16996	Hexacosenoic Acid, C26:1	33036-5
16997	Hexacosanoic Acid, C26:0	30197-8
16998	Pristanic Acid, C15:0(CH3)4	22761-1
16999	Phytanic Acid, C16:0(CH3)4	22671-2
17000	Triene Tetraene Ratio	35411-8
17001	Total Saturated	35175-9
17002	Total Monounsaturated	35176-7
17003	Total Polyunsaturated	35177-5
17004	Total w3	35178-3
17005	Total w6	35179-1
17006	Total Fatty Acids	24461-6
17056	Interpretation	59462-2