



# Test Definition: FAPM

Fatty Acid Profile, Mitochondrial (C8-C18),  
Serum

## Overview

### Useful For

Biochemical diagnosis of inborn errors of mitochondrial fatty acid oxidation, including deficiencies of medium-chain acyl-Co-A dehydrogenase, long-chain 3-hydroxyacyl-Co-A dehydrogenase, very long-chain acyl-Co-A dehydrogenase, and glutaric acidemia type 2

### Genetics Test Information

This test can be ordered to screen patients with a suspected mitochondrial fatty acid oxidation (FAO) disorder.

### Testing Algorithm

For more information see [Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)

### Special Instructions

- [Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)

### Method Name

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

### NY State Available

Yes

## Specimen

### Specimen Type

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

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 workup and differential diagnosis of candidate patients. Fatty acid profiling can detect

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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

**Reference Values**

Octanoic Acid, C8:0

&lt;1 year: 7-63 nmol/mL

1-17 years: 9-41 nmol/mL

&gt; or =18 years: 8-47 nmol/mL

Decenoic Acid, C10:1

&lt;1 year: 0.8-4.8 nmol/mL

1-17 years: 1.6-6.6 nmol/mL

&gt; or =18 years: 1.8-5.0 nmol/mL

Decanoic Acid, C10:0

&lt;1 year: 2-62 nmol/mL

1-17 years: 3-25 nmol/mL

&gt; or =18 years: 2-18 nmol/mL

Lauroleic Acid, C12:1

&lt;1 year: 0.6-4.8 nmol/mL

1-17 years: 1.3-5.8 nmol/mL

&gt; or =18 years: 1.4-6.6 nmol/mL

Lauric Acid, C12:0

&lt;1 year: 6-190 nmol/mL

1-17 years: 5-80 nmol/mL

&gt; or =18 years: 6-90 nmol/mL

Tetradecadienoic Acid, C14:2

&lt;1 year: 0.3-6.5 nmol/mL

1-17 years: 0.2-5.8 nmol/mL

&gt; or =18 years: 0.8-5.0 nmol/mL

Myristoleic Acid, C14:1

&lt;1 year: 1-46 nmol/mL

1-17 years: 1-31 nmol/mL

&gt; or =18 years: 3-64 nmol/mL

Myristic Acid, C14:0

&lt;1 year: 30-320 nmol/mL

1-17 years: 40-290 nmol/mL

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> 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

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

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

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

### Interpretation

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

### Cautions

For nutritional assessment, a 12- to 14-hour fast is required; however, patients 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. Rinaldo P, Matern D, Bennett MJ. Fatty acid oxidation disorders. *Ann Rev Physiol.* 2002;64:477-502

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2. Kang E, Kim YM, Kang, M, et al. Clinical and genetic characteristics of patients with fatty acid oxidation disorders identified by newborn screening. BMC Pediatr. 2018;18(1):103

## 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
FAPM	Fatty Acid Profile, Mitochondrial,S	43675-8

Result ID	Test Result Name	Result LOINC® Value
17007	Octanoic Acid, C8:0	35145-2
17008	Decenoic Acid, C10:1	35147-8
17009	Decanoic Acid, C10:0	35146-0
17010	Lauroleic Acid, C12:1	35151-0
17011	Lauric Acid, C12:0	35150-2
17012	Tetradecadienoic Acid, C14:2	35148-6
17013	Myristoleic Acid, C14:1	35158-5
17014	Myristic Acid, C14:0	35157-7
17015	Hexadecadienoic Acid, C16:2	35154-4
17016	Palmitoleic Acid, C16:1w7	35162-7
17017	Palmitic Acid, C16:0	35161-9
17018	Linoleic Acid, C18:2w6	35165-0
17019	Oleic Acid, C18:1w9	35166-8
17020	Stearic Acid, C18:0	35149-4
17055	Interpretation	59462-2