
Lab Dept: **Anatomic Pathology**

Test Name: **FATTY ACID OXIDATION PROBE,
FIBROBLASTS**

General Information

Lab Order Codes: FOAM

Synonyms: Fatty Acid Ox Probe Assay, Fibroblast Culture

CPT Codes: 82017 – Acylcarnitines; quantitative, each specimen
The following will be added on with additional charges, if appropriate:
88233 – Fibroblast culture
88240 – Cryopreservation for biochemical studies

Test Includes: An interpretive report of findings and recommendations for follow-up
and/or additional testing.

Logistics

Test Indications: In vitro confirmation of biochemical diagnoses of the following fatty acid
oxidation disorders:

- Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Long-chain 3-hydroxacyl-CoA dehydrogenase (LCHAD) deficiency
- Trifunctional protein deficiency
- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
- Carnitine palmitoyl transferase deficiency type II (CPT-II)
- Carnitine-acylcarnitine translocase (CACT) deficiency

In addition, the following organic acid disorders can be confirmed by
this assay:

- 2-Methylbutyryl-CoA dehydrogenase (SBCAD) deficiency
- Isobutyryl-CoA dehydrogenase (IBD) deficiency

This test is recommended only after appropriate anlyte testing
including acylcarnitines, organic acids, acylglycines, and/or fatty acids
(ACRN, OAU, ACULG, FAPCP, FAPM, FAPEP). Please provide clinical
information with patient specimens.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Medical Laboratories (MML Test: FAO/81927)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability:	Monday – Thursday ONLY
Turnaround Time:	15 - 71 days, test
Special Instructions:	Please provide clinical information with the specimen. Restricted collection days. Collect Monday – Thursday only. Cryopreserved specimens will be stored for 3 years or until specimen has been used up performing additional analyses.

Specimen

Specimen Type:	Tissue, skin
Container:	Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640O. The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin, Mayo Supply T115).
Draw Volume:	4-mm punch skin biopsy
Processed Volume:	Same as Draw Volume
Collection:	Determined by medical provider
Special Processing:	Lab Staff: Submit tissue in appropriate media. Store and ship at refrigerated temperatures. Forward promptly.
Patient Preparation:	Determined by medical provider
Sample Rejection:	Inappropriate specimens in formalin or fixative preservative; mislabeled or unlabeled specimens

Interpretive

An interpretive report will be provided

Interpretation: Abnormal results will include a description of the abnormal profile, in comparison to normal and abnormal co-run controls. In addition, the concentration of those acylcarnitine species that abnormally accumulated in the cell medium are provided and compared to the continuously updated reference range based on analysis of normal controls. Interpretations of abnormal acylcarnitine profiles also include information about the results' significance, a correlation to available clinical information, possible differential diagnoses, recommendations for additional biochemical testing and confirmatory studies if indicated, name and phone number of contacts who may provide these studies at the Mayo Clinic or elsewhere, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Critical Values: N/A

Limitations: This test is not available for prenatal testing. This assay is not informative if the deficient enzyme is physiologically not expressed in skin fibroblasts. Sometimes an abnormal acylcarnitine profile cannot differentiate between 2 disorders. In such instances, independent biochemical (eg, specific enzyme assay) or molecular genetic analyses are required.

Methodology: Fatty Acid Oxidation Probe: Fibroblasts incubated with enriched medium followed by Tandem Mass Spectrometry (MS/MS) for acylcarnitines
Fibroblast Culture: Cultivated from biopsy as monolayer
Cryopreservation: Fibroblast subculture followed by cryopreservation and storage.

References: [Mayo Medical Laboratories](#) October 2014