

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Fatty Acid Oxidation Disorders Gene Panel
- **Test Code:** 4695

Related Diseases & Genes:

• “Short-chain acyl-CoA dehydrogenase (SCAD) deficiency, Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, Trifunctional protein deficiency, Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency, infantile Carnitine palmitoyl transferase deficiency type II (CPT-II), Lethal Neonatal Carnitine palmitoyl transferase deficiency type II (CPT-II), Myopathic Carnitine palmitoyl transferase deficiency type II (CPT-II), Carnitine-acylcarnitine translocase (CACT) deficiency, 2-Methylbutyryl-CoA dehydrogenase (SBCAD) deficiency, Isobutyryl-CoA dehydrogenase (IBD) deficiency” (OMIM #: 201470 201450 609016 609015 609015 201475 600649 608836 255110 212138 610006 611283)|| Gene: “ACADS, ACADM, HADHA, HADHB, ACADVL, CPT2, CACT, ACADSB, ACAD8” (OMIM #:)

Synonyms:

-none-

Test Details:

- **Methodology:** Next Generation DNA Sequencing
- **Performed:** Everyday
- **Reported:** 6 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Inborn Errors of Metabolism

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