MEDIUM CHAIN ACYL CoA DEHYDROGENASE DEFICIENCY (MCAD)

Orderable - MD MCAD Screen

Turnaround Time: 4-6 weeks STAT: 4 weeks

Alternate Name(s):
ACADM
MCAD

Specimen:
Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:
Sample may be transported at room temperature

Reference Ranges:
See report

Interpretive Comments:
Medium chain acyl CoA dehydrogenase (ACADM, a.k.a. MCAD) deficiency is a recessive trait associated with defective oxidation of fatty acids which may have serious clinical sequelae. In the Ontario population approximately 90% (PMID:20434380) of alleles associated with ACADM (MCAD) deficiency have a single A>G mutation at nucleotide #985. Thus approximately 81% of clinically affected members of this population would be expected to be homozygous for the 985A>G mutation, 18% would be compound heterozygotes while 13% of the alleles are expected to carry the 199C>T mutation with the remaining mutations being private mutations distributed throughout the ACADM gene.

Comments:
Full ACADM gene sequencing and del/dup analysis.
**Storage and Shipment:**

Store and ship at room temperature within 5 days of collection.

MLPA copy number analysis. All variants interpreted as either ACMG category 1, 2, or 3 (pathogenic, likely pathogenic, VUS; PMID: 25741868) are confirmed using Sanger sequencing, MLPA, or other assays. ACMG category 4 and 5 variants (likely benign, benign) are not reported, but are available upon request. This assay has been validated at a level of sensitivity equivalent to the Sanger sequencing and standard copy number analysis (>99%; PMID: 27376475).

**Test Schedule:**
As required, Monday to Friday 0800-1600 hours