Test Definition: VLCKM

Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Known Mutation

Reporting Title: VLCAD Deficiency, Known Mutation
Performing Location: Rochester

Specimen Requirements:

This test can only be performed if a mutation has previously been identified in a family member of this individual.

Forms:
1. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
3. If not ordering electronically, submit a Molecular Genetics Request Form (Supply T245) with the specimen.

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:
Specimen Type: Blood
Container/Tube: Lavender top (EDTA) or yellow top (ACD)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Acceptable:
Specimen Type: Blood spots
Container/Tube: Whatman Protein Saver 903 Paper
Specimen Volume: 5 blood spots
Collection Instructions:
1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
2. Do not expose specimen to heat or direct sunlight.
3. Do not stack wet specimens.
4. Keep specimen dry.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts
Container/Tube: T-75 or T-25 flask
Specimen Volume: 1 full T-75 or 2 full T-25 flasks
Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type	Temperature	Time
Varies	Varies	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
51025	Specimen	Alphanumeric		31208-2
51026	Specimen ID	Alphanumeric		N/A
51027	Source	Alphanumeric		N/A
51028	Order Date	Alphanumeric		N/A
51029	Reason For Referral	Alphanumeric		42349-1
51030	Method	Alphanumeric		In Process
51031	Result	Alphanumeric		In Process
51032	Interpretation	Alphanumeric		69047-9
51033	Extraction Performed?	Alphanumeric		N/A
51034	Amendment	Alphanumeric		In Process
51035	Reviewed By	Alphanumeric		N/A
51036	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81403

Reference Values:

An interpretive report will be provided.