Rhabdomyolysis: Sequencing Panel

| Test Code: MM650                                                                 |
| Turnaround time: 6 weeks                                                        |
| CPT Codes: 81404 x1, 81405 x1, 81406 x1, 81407 x1                              |

Condition Description

Rhabdomyolysis is caused by the breakdown of muscle leading to an increase in serum and urine myoglobin, and creatine kinase levels (CK previously referred to as CPK). Rhabdomyolysis can be caused by a number of conditions including excessive exercise, drugs, congenital muscular dystrophies and inborn errors of metabolism. The most common inborn errors of metabolism include fatty acid oxidation disorders, glycogen storage diseases, and mitochondrial disorders.

Reference:

Genes

ACAD9, ACADL, ACADVL, AGL, AMPD1, CPT2, ETFB, GAA, GYS1, HADHA, HADHB, LPIN1, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, PYGM, RRM2B, SLC22A5, SUCL2, TK2, TYMP

Indications

This test is indicated for:

- Individuals with recurrent episodes of rhabdomyolysis.
- Individuals with an unexplained rise in CPK.

Methodology

**Next Generation Sequencing:** In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

**Next Generation Sequencing:** Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (Children >2 years): 3-5 ml.
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

- In microtainer: 60 ug.
- Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Special Instructions

Please indicate any medications or dietary changes on the test requisition.

Related Tests

- Acylcarnitine Profile
- Carnitine Concentration Profile
- Organic Acids Quantitative Analysis
- Coenzyme Q10 Profile, plasma
- Urine Amino Acids
- Urine Carnitine Profile
- Myophosphorylase Deficiency (McArdle Disease): PYGM Mutation Panel Test, Full Gene Sequencing and Deletion/Duplication Analysis
- Adenosine Monophosphate Deaminase 1 Deficiency: AMPD1 Mutation Panel Test and Full Gene Sequencing
- Medium Chain Acyl Co-A Dehydrogenase Deficiency: ACADM Mutation Panel Test, Full Gene Sequencing, and Deletion/Duplication Analysis
- CPT2: CPT2 Full Gene Sequencing and Deletion/Duplication Analysis
- Glutaric Aciduria Type 2 (GA II): ETFA, ETFB Full Gene Sequencing and Deletion/Duplication Analysis