Congenital Myasthenic Syndromes: Deletion/Duplication Panel

Test Code: MD110  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1

**Condition Description**

Congenital myasthenic syndromes (CMS) are a highly variable group of diseases characterized by fatigable weakness of skeletal muscle. Symptoms range from mild to progressive disabling weakness. The age of onset is also variable ranging from birth to early childhood. Infections, fever, or excitement may precipitate severe exacerbations of weakness or episodes of respiratory insufficiency. Additional features of the neonatal onset subtypes include feeding difficulties, choking spells, poor suck and cry, eyelid ptosis, and weakness. Additional features of the childhood onset subtypes include delayed motor milestones, fluctuating eyelid ptosis, and fluctuating extraocular muscle weakness. CMS can be inherited in an autosomal recessive or an autosomal dominant manner; however, the autosomal recessive manner is more common.

References:
- GeneReviews
- OMIM

**Genes**

- ALG2
- CHAT
- CHRNA1
- CHRNB1
- CHRNDE
- COLQ
- DOK7
- MUSK
- RAPSN
- SCN4A

**Indications**

This test is indicated for:
- Confirmation of a clinical diagnosis of congenital myasthenic syndromes (CMS).
- Carrier testing in adults with a family history of congenital myasthenic syndromes (CMS).

**Methodology**

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (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: 10 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.

**Related Tests**

- Congenital Myasthenic Syndromes: Sequencing Panel