Congenital Muscular Dystrophy: Sequencing Panel

Test Code: MM211
Turnaround time: 6 weeks
CPT Codes: 81404 x1, 81405 x1, 81406 x1

Condition Description

The congenital muscular dystrophies are a group of genetically and clinically heterogeneous hereditary myopathies characterized by congenital hypotonia and muscle weakness, contractures, and delayed motor development. Muscle biopsy usually reveals a nonspecific dystrophic pattern. The clinical course is broadly variable and can involve the brain and eyes. Initial testing often includes clinical evaluation, muscle imaging, electromyography, and muscle biopsy, followed by targeted genetic testing.

Note: This test does not detect the retrotransposon insertion in the 3' UTR of the \textit{FKTN} gene common in some Asian populations. For patients with suspected Fukuyama congenital muscular dystrophy, testing for the \textit{FKTN} insertion is recommended. Analysis for the \textit{FKTN} insertion is available as a separate assay.

Reference:
- OMIM

Genes


Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of congenital muscular dystrophies (CMD).

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

\textit{Submit only 1 of the following specimen types}

\textbf{Type: Saliva}

\textbf{Specimen Requirements:}

Oragene™ Saliva Collection Kit
Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

\textbf{Specimen Collection and Shipping:}

Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

\textbf{Type: Whole Blood (EDTA)}

\textbf{Specimen Requirements:}

EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old): 3-5 ml
Older Children & Adults: 5-10 ml
Autopsy: 2-3 ml unclotted cord or cardiac blood

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Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
8µg
Isolation using the Perkin Elmer™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

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

Related Tests

- Single-gene tests.
- Neuromuscular Disorders Panel.
- Congenital Muscular Dystrophy: Deletion/Duplication Panel.