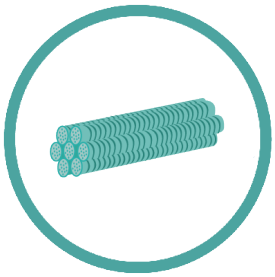
variantyx



Genomic Unity® Muscular Dystrophy Analysis
NR008

Overview

Muscular dystrophies are a group of genetic disorders characterized by progressive muscle degeneration and weakness.

Genomic Unity® Muscular Dystrophy Analysis is an effective test for the genetic cause of muscle weakness in patients suspected of having one of the following muscular dystrophies: Becker muscular dystrophy, Duchenne muscular dystrophy, Emery-Dreifuss muscular dystrophy, Limb-Girdle muscular dystrophy, Ullrich congenital muscular dystrophy, Walker-Warburg syndrome as well as other congenital muscular dystrophies.

Method

PCR free whole genome sequencing (WGS) is used as the underlying NGS technology. Its consistent read depth across >98% of the genome enables identification of multiple variant types from a single patient sample.

Proprietary algorithms optimized for each variant type are used to perform discrete in-silico analyses of the data which are brought together for collective interpretation, providing a complete genetic picture.

Rigorously trained variant scientists interpret all variant types in the context of the patient's phenotype and generate a unified clinical report.

Included analyses

- ✓ Sequencing analysis of muscular dystrophy associated genes
- ✓ Del/dup analysis of muscular dystrophy associated genes

Test performance

Highly uniform sequencing depth

- 30X mean mappable coverage
- >98% of nucleotides covered at ⩾8x
- >99% of HGMD and ClinVar annotated variants covered at ⩾8x

Highly sensitive detection of SNVs and indels up to 50 bp

- 99.8% sensitivity
- 99.7% positive predictive value

Highly sensitive detection of structural variants

- 96% clinical sensitivity
- In most cases, the exact genomic coordinates (the breakpoints) of the structural variant can be determined

Accepted sample types

- Blood - optimally 5ml
- gDNA - 5µg minimum
- Saliva

Reflex option

In the case that Genomic Unity® Muscular Dystrophy Analysis does not identify causal variants, the option is given to reflex up to Genomic Unity® Whole Genome Analysis which looks more broadly for causal variants across all genes.

The reflex option is offered for a nominal patient pay price when not covered by the patient's insurance.

Genes analyzed

ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, ISPD, ITGA7, LAMA2, LARGE1, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SELENON, SGCA, SGCB, SGCD, SGCG, SMCHD1, SUN1, SUN2, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN

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