



Genomic Unity[®] Neuromuscular Disorders Analysis

Overview

Neuromuscular disorders encompass a wide spectrum of nerve-related diseases and conditions that affect the body's muscles. Patients experience progressive muscle weakness and atrophy due to disrupted communication between the nervous system and the muscles it controls.

Genomic Unity[®] Neuromuscular Disorders Analysis is an effective test for the genetic cause of muscle weakness in patients with clinical symptoms consistent with the following inherited neuromuscular disorders: muscular dystrophies, congenital myopathies and congenital myasthenic syndromes.

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 neuromuscular disorder associated genes
- ✓ Del/dup analysis of neuromuscular disorder associated genes
- ✓ Myotonic dystrophy STR analysis: *CNBP*, *DMPK*

Test performance

Highly uniform sequencing depth

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

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 - 5 μ g minimum
- Saliva

Turnaround time

6-8 weeks after sample receipt

Reflex option

In the case that Genomic Unity[®] Neuromuscular Disorders 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

ACTA1, AGRN, ALG14, ALG2, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRYAB, DAG1, DES, DMD, DMPK, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKBP14, FKR, FKTN, FLNC, GAA, GFPT1, GMPPB, GNE, GYS1, HINT1, HNRNPA2B1, HNRNPDL, HSPG2, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, KLHL9, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LIMS2, LMNA, LMOD3, LRP4, MATR3, MEGF10, MTM1, MTMR14, MUSK, MYF6, MYH2, MYH7, MYO9A, MYOT, MYPN, NEB, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, RAPSN, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC5A7, SMCHD1, SNAP25, STAC3, STIM1, SUN1, SUN2, SYNE1, SYNE2, TCAP, TIA1, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VCP, VMA21

The *NEB* gene is not fully covered by this test, therefore pathogenic variants may not be detected in this gene.