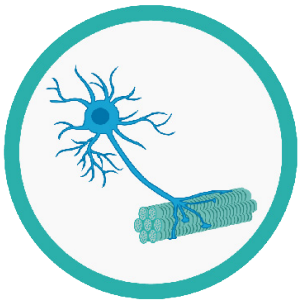
variantyx



Genomic Unity® Motor Neuron Disorders Analysis

NR005

Overview

Motor neuron disorders affect the nerves that control the body's voluntary muscles. They are characterized by progressive muscle weakness and atrophy, spasticity and overactive tendon reflexes.

Genomic Unity® Motor Neuron Disorders Analysis is an effective test for the genetic cause of muscle weakness in patients with clinical symptoms consistent with the following inherited motor neuron disorders: Amyotrophic lateral sclerosis (ALS), Hereditary spastic paraplegia (HSP), motor neuropathies, Spinal muscular atrophy (SMA) as well as related disorders.

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 motor neuron disorder associated genes
- ✓ Del/dup analysis of motor neuron disorder associated genes
- ✓ Frontotemporal dementia and Amyotrophic lateral sclerosis (FTD-ALS) STR analysis: *C9ORF72*
- ✓ Spinal and bulbar muscular atrophy STR analysis: *AR*

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

- 99.7% 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® Motor Neuron 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

ALDH18A1, ALS2, AMPD2, ANG, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ASAH1, ATL1, ATP13A2, ATP7A, B4GALNT1, BICD2, BSCL2, C12ORF65, C19ORF12, C9ORF72, CAPN1, CCT5, CHCHD10, CHMP2B, CPT1C, CYP2U1, CYP7B1, DCTN1, DDHD1, DDHD2, DNAJB2, DYNC1H1, ENTPD1, ERBB4, ERLIN1, ERLIN2, EXOSC3, EXOSC8, FA2H, FARS2, FBXO38, FIG4, FUS, GARS1, GBA2, GJC2, GRN, HACE1, HINT1, HNRNPA1, HSPB1, HSPB3, HSPD1, IBA57, IGHMBP2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, LAS1L, MAG, MAPT, MARS1, MATR3, NEFH, NIPA1, NT5C2, OPTN, PFN1, PLEKHG5, PLP1, PNPLA6, PRPH, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SETX, SIGMAR1, SLC16A2, SLC1A4, SLC33A1, SLC52A3, SLC5A7, SMN1, SMN2, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SQSTM1, TAF15, TARDBP, TBK1, TECPR2, TFG, TRPV4, TUBA4A, UBA1, UBAP1, UBQLN2, USP8, VAMP1, VAPB, VCP, VPS37A, VRK1, WASHC5, ZFYVE26, ZFYVE27

The *SMN2* gene is only analyzed for determination of copy number for assessment of severity of spinal muscular atrophy (SMA).

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Controlled document #: DCMMPRC850026

Updated 09-01-2021

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