variantyx

Genomic Unity® Movement Disorders Analysis

NR006

Overview

Genomic Unity® Movement Disorders Analysis is an effective test for the genetic cause of abnormal movement in patients with suspected ataxia and dystonia diagnoses. As well as suspected diagnoses of Parkinsons, Parkinsonism, Tyrosine hydroxylase deficiency and choreas including Hungtington related diseases.

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

- ✓ Sequence analysis of movement disorder associated genes
- ✓ Del/dup analysis of movement disorder associated genes
- ✓ Adult-onset movement disorder (with or without cognitive involvement) STR analysis: *ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, CACNA1A, FXN, NOP56, NOTCH2NLC, PPP2R2B, TBP.*
  Optionally includes *HTT* and *JPH3*

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
- 99.6% 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® Movement 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

ABCB7, ABHD12, ABHD5, ACO2, ADAR, ADCY5, AFG3L2, AHI1, ALDH5A1, ANO10, ANO3, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BEAN1, CA8, CACNA1A, CACNA1B, CACNA1G, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CIZ1, CLCN2, CLN5, CLPP, COASY, COL6A3, COQ8A, COX20, CP, CPLANE1, CWF19L1, CYP27A1, CYP2U1, DCAF17, DDC, DLAT, DNAJC19, DNAJC6, DNMT1, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FBXO7, FGF14, FLVCR1, FTL, FXN, GBA2, GCDH, GCH1, GFAP, GLRA1, GNAO (GNOA1), GOSR2, GRID2, GRM1, GSS, HARS2, HEXA, HIBCH, HPCA, INPP5E, ITN2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ1, KCNMA1, KCTD17, KIF1C, KIF7, KMT2B, LAMA1, LARS2, LMNB1, LRPPRC, LRRK2, MAPT, MARS2, MECR, MKKS, MKS1, MME, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX2-1, NOL3, NOP56, NOTCH2NLC, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PANK2, PARK7, PDGFB, PDGFRB, PDYN, PEX7, PEX10, PHYH, PINK1, PLA2G6, PNKD, PNKP, PNPL86, POLG, POLR3A, POLR3B, PPP2R2B, PRKCG, PRKN, PRKRA, PRRT2, RELN, RNF216, RPGRIP1L, RUBCN, SACS, SCN2A, SCP2, SERAC1, SETX, SGCE, SIL1, SLC16A2, SLC19A3, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC30A10, SLC52A2, SLC6A3, SLC9A6, SNCA, SNX14, SPG7, SPR, SPTBN2, STUB1, SYNE1, SYNJ1, SYT14, TAF1, TBP, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TH, THAP1, TIMM8A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TOR1A, TOR1AIP1, TPK1, TPP1, TRAPPC11, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, VPS13A, VPS35, WDPCP, WDR45, WDR81, WFS1, WWOX, XPR1, ZFYVE26, ZNF423

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