variantyx

Genomic Unity® Neurology Analysis

Overview

Neurological disorders are particularly difficult to diagnose due to their heterogeneous nature. An early diagnosis can be instrumental in determining likely prognosis and guiding clinical management to delay onset of additional symptoms.

Genomic Unity® Neurology Analysis is an effective test for the genetic cause of many neurological disorders including: brain malformations, epilepsy disorders, motor neuron disorders, movement disorders, neurodevelopmental disorders, neuromuscular disorders and neuropathies.

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 more 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 neurology associated genes
- ✓ Del/dup analysis of neurology associated genes
- ✓ Adult-onset movement disorder STR analysis: *AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, C9ORF72, CACNA1A, CNBP, CSTB, DMPK, FMR1, FXN, NOP56, NOTCH2NLC, PPP2R2B, TBP.* Optionally includes *HTT* and *JPH3*
- ✓ Additional STR analysis: *AFF2, AFF3, DIP2B, FMR1*

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

Turnaround time

6-8 weeks after sample receipt

Reflex option

In the case that Genomic Unity® Neurology 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

A2M, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA7, ABCB7, ABCC6, ABCC8, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACAT1, ACE, ACER3, ACKR1, ACO2, ACOX1, ACSF3, ACSL4, ACTA2, ACTB, ACTG1, ACVRL1, ACY1, ADAM10, ADAR, ADCK3, ADCY5, ADGRG1, ADGRV1, ADK, ADNP, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGRN, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AKT1, AKT3, ALAD, ALAS2, ALDH18I, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH5A2, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX5AP, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMMECR1, AMN, AMPD1, AMPD2, AMT, ANG, ANK3, ANKLE2, ANKO5, ANKRD11, ANKTRX1, ANO10, ANO3, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOE, APP, APTX, AR, ARFGEF2, ARG1, ARHGAP31, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARK6IP1, ARL13B, ARL6, ARSA, ARSB, ARSE, ARSI, ARV1, ARX, ASAH1, ASCL1, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL3, ATAD3A, ATCAY, ATIC, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B3, ATP2B4, ATP5E, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, AUT22, AUTS2, B3GALNT2, B3GLCT, B4GALNT1, B4GAT1, B4GLAT1, B9D1, B9D2, BAG3, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCORL1, BCS1L, BDNF, BEAN1, BEST1, BICD2, BIN1, BLOC1S3, BLOC1S6, BMP4, BOLA3, BRAF, BRAT1, BRWD3, BSCL2, BSND, BTD, BVES, C12ORF57, C12ORF65, C19ORF12, C9ORF72, CA8, CACNA1A, CACNA1B, CACNA1D, CACNA1G, CACNA1H, CACNA1S, CACNA2D2, CACNB4, CACNG2, CAMTA1, CAPN1, CAPN3, CARS2, CASC5, CASK, CASR, CAV3, CBL, CC2D1A, CC2D2A, CCDC22, CCDC28B, CCDC78, CCDC88A, CCDC88C, CCM2, CCND2, CCT5, CD207, CD320, CD36, CD59, CD96, CDH15, CDK11A, CDK5RAP2, CDKL5, CDKN1C, CDKON, CEADD, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP63, CERS1, CFL2, CHAT, CHCHD10, CHCHD2, CHD2, CHD7, CHD8, CHKB, CHMP1A, CHMP2B, CHN1, CHRM3, CHRNA1, CHRNA2, CHRNA4, CHRNA7, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHSY1, CISD2, CISH, CIT, CIZ1, CLCN1, CLCN2, CLCN4, CLCNKA, CLCNKB, CLIC2, CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNBP, CNKSR2, CNOT3, CNTN1, CNTN6, CNTNAP2, CNTNAP4, COA5, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL12A1, COL18A1, COL4A1, COL4A2, COL4A3BP, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, COX7B, CP, CPA6, CPLANE1, CPLX1, CPOX, CPT2, CR1, CRBN, CREBBP, CRYAB, CSF1R, CSF2RB, CSNK2A1, CSPP1, CST3, CSTB, CTBP1, CTC1, CTCF, CTDP1, CTFD, CTNNB1, CTNND2, CTSD, CTSF, CUL4B, CWF19L1, CYP11B2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCAF8, DCHS1, DCTN1, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DHFR, DHH, DHTKD1, DIAPH1, DIAPH3, DIS3L2, DISP1, DKC1, DLAT, DLD, DLG3, DLL1, DLL4, DMD, DNAH9, DNAJB2, DNAJB6, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DRD2, DRD3, DSC3, DST, DTNBP1, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECHS1, ECM1, EDN3, EDNRB, EED, EEF1A2, EEF2, EFHC1, EFTUD2, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4E, EIF4G1, ELK1, ELOVL4, ELOVL5, ELP1, EMD, EMX2, EN2, ENO3, ENTPD1, EOGT, EOMES, EP300, EPB41L1, EPM2A, ERBB4, ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOC8, EXOSC3, EXOSC8, EZH2, F2, F5, FA2H, FADD, FAM126A, FAM134B, FANCB, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBXL4, FBXO38, FBXO7, FCGR2B, FECH, FGA, FGD1, FGD4, FGF10, FGF12, FGF14, FGF8, FGFR1, FGFR2, FGFR3, FGFRL1, FH, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNC, FLRT1, FLVCR1, FLVCR2, FMR1, FOLR1, FOXC1, FOXG1, FOXH1, FOXP1, FOXP2, FOXP3, FOXRED1, FREM1, FRG1, FRMD7, FRMPD4, FRRS1L, FTCD, FTL, FTO, FTSJ1, FUS, FXN, FZD9, GAA, GABBR2, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GAMT, GAN, GARS, GAS1, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCSH, GDAP1, GDI1, GDNF, GFAP, GFER, GFM1, GFPT1, GGSN, GIGYF2, GJA1, GJB1, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLI2, GLI3, GLRA1, GLRB, GLUL, GLYCTK, GM2A, GMPPB, GNAL, GNAO1, GNB1, GNB4, GNE, GNPAT, GNS, GOSR2, GP1BA, GPC3, GPHN, GPR143, GPSM2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, GRM1, GRM7, GRN, GSS, GTPBP3, GUF1, GUSB, GYG1, GYS1, HACE1, HADHA, HADHB, HAL, HARS1, HARS2, HCCS, HCFC1, HCN1, HCN4, HDAC4, HDAC6, HDAC8, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HFE, HGSNAT, HIBCH, HINT1, HIVEP, HK1, HMBS, HMGB3, HMGCS2, HNRNP2B1, HNRNPA1, HNRNPDL, HNRNPH2, HNRNPU, HOXA1, HOXD10, HPCA, HPD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HRAS, HSD17B10, HSD17B4, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, HTRA1, HTRA2, HTT, HUWE1, HYAL1, IBA57, ICAM1, ICK, IDS, IDUA, IER3IP1, IFIH1, IFRD1, IFT140, IFT172, IFT27, IGBP1, IGF1, IGHMBP2, IKBKAP, IKBKG, IL11RA, IL1RAPL1, IL1RN, INF2, INPP5E, INS, IQSEC2, IRX5, ISCA2, ISCU, ISPD, ITGA7, ITM2B, ITN2B, ITPA, ITPR1, JMJD1C, JRK, KANK1, KANSL1, KARS, KAT6A, KAT6B, KATNB1, KBTBD13, KCNA1, KCNA2, KCNB1, KCNC1, KCNC3, KCND3, KCNE1L, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ2, KCNK19, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD17, KCTD7, KDM5C, KDM6A, KIAA0586, KIDINS220, KIF11, KIF1A, KIF1B, KIF1BP, KIF1C, KIF21A, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KLF8, KLHL15, KLHL40, KLHL41, KLHL9, KMT2A, KMT2B, KMT2D, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT5, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMB2, LAMC3, LAMP2, LARGE1, LARS2, LAS1L, LBR, LDB3, LDHA, LETM1, LGI1, LHX4, LIAS, LIG4, LIMS2, LINS1, LIPT1, LITAF, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LPIN1, LPIN2, LRP2, LRP4, LRPPRC, LRRK2, LRSAM1, LYRM7, LYZ, LZTFL1, MAG, MAGEL2, MAGI2, MAGT1, MAMLD1, MAN1B1, MAOA, MAP2K1, MAPK10, MAPT, MARS1, MARS2, MASP1, MAT1A, MATR3, MBD5, MBTPS2, MCEE, MCM3AP, MCOLN1, MCPH1, MECP2, MECR, MED12, MED13L, MED17,

MED23, MED25, MEF2C, MEGF10, MET, MFN2, MFSD2A, MFSD8, MGAT2, MICU1, MID1, MID2, MKKS, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MME, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPDZ, MPI, MPV17, MPZ, MRE11, MRPL44, MRPS22, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTMR14, MTMR2, MTO1, MTOR, MTPAP, MTR, MTRR, MTTP, MUSK, MUT, MVK, MYCN, MYF6, MYH14, MYH2, MYH3, MYH7, MYH9, MYL2, MYO5A, MYO9A, MYOT, MYPN, MYT1L, NAA10, NACC1, NADK2, NAGA, NAGLU, NALCN, NAT8L, NBN, NDE1, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NEB, NECAP1, NEDD4L, NEFH, NEFL, NEMO, NEU1, NEXMIF, NF1, NFIA, NFIX, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHS, NIPA1, NIPBL, NKX2-1, NLGN3, NLGN4X, NLRP12, NLRP3, NOD2, NODAL, NOG, NOL3, NONO, NOP56, NOS2, NOS3, NOTCH1, NOTCH2, NOTCH2NLC, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPRL3, NR2F1, NRAS, NRG1, NRXN1, NSD1, NSD2, NSDHL, NSF5, NSUN2, NT5C2, NTRK1, NTRK2, NUBPL, NXF5, OCLN, OCRL, OFD1, OGT, OPA1, OPA3, OPHN1, OPTN, ORC1, OTC, OTX2, PABPN1, PACS1, PAFAH1B1, PAH, PAK3, PANK2, PARK2, PARK7, PAX6, PC, PCBD1, PCDH12, PCDH19, PCK2, PCNT, PDCD10, PDE6D, PDE8B, PDGFB, PDGFRB, PDHA1, PDK3, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGAP2, PGK1, PGM1, PHF6, PHF8, PHGDH, PHKA1, PHOX2A, PHOX2B, PHYH, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIK3CA, PIK3R2, PIK3R5, PINK1, PLA2G6, PLCB1, PLEC, PLEKHG2, PLEKHG4, PLEKHG5, PLK4, PLP1, PMM2, PMP22, PNKD, PNKP, PNPL86, PNPLA2, PNPLA6, PNPO, POGLUT1, POGZ, POLA1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, PPOX, PPP2R1A, PPP2R2B, PPP2R5D, PPT1, PQBP1, PRDM12, PREPL, PRF1, PRICKLE1, PRICKLE2, PRIMA1, PRKAG2, PRKCG, PRKCH, PRKDC, PRKN, PRKRA, PRNP, PRODH, PROP1, PRPH, PRPH2, PRPS1, PRRT2, PRRX1, PRSS12, PRX, PSAP, PSEN1, PSEN2, 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SLC25A12, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC38A8, SLC39A8, SLC3A1, SLC46A1, SLC4A1, SLC4A4, SLC52A2, SLC52A3, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC9A6, SLC9A9, SMAD2, SMAD3, SMARCA2, SMARCA4, SMARCB1, SMC1A, SMC3, SMCHD1, SMN1, SMN2, SMPD1, SMS, SNAP25, SNAP29, SNCA, SNCAIP, SNCB, SNIP1, SNORD118, SNX14, SNX3, SOBP, SOD1, SORL1, SOS1, SOX10, SOX2, SOX3, SPART, SPAST, SPATA5, SPEG, SPG11, SPG20, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SQSTM1, SRD5A3, SRPX2, SSR4, ST3GAL3, ST3GAL5, STAC3, STAG2, STAMBP, STAT1, STIL, STIM1, STRADA, STUB1, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUN1, SUN2, SUOX, SURF1, SYN1, SYNE1, SYNE2, SYNGAP1, SYNJ1, SYP, SYT14, SYT2, SZT2, TACO1, TAF1, TAF15, TAF2, TANGO2, TARDBP, TARS2, TAS2R38, TAZ, TBC1D20, TBC1D24, TBC1D7, TBCD, TBCE, TBCK, TBK1, TBL1XR1, TBP, TBR1, TBX1, TCAP, TCF20, TCF4, TCTN1, TCTN2, TCTN3, TDGF1, TDP1, TECPR2, TECR, TFAP2A, TFAP2B, TFG, TG, TGFB1, TGFB2, TGFBR1, TGFBR2, TGIF1, TGM6, TH, THAP1, THOC2, TIA1, TICAM1, TIMM8A, TIRAP, TK2, TLR3, TLR5, TM4SF20, TMCO1, TMEM126A, TMEM126B, TMEM138, TMEM165, TMEM187, TMEM216, TMEM231, TMEM237, TMEM240, TMEM43, TMEM5, TMEM67, TMEM70, TMLHE, TNF, TNFSF4, TNNI2, TNNT1, TNPO3, TOR1A, TOR1AIP1, TP63, TPK1, TPM2, TPM3, TPP1, TRAF3, TRAPPC11, TRAPPC9, TREM2, TREX1, TRIM2, TRIM32, TRIO, TRIP12, TRIP4, TRMT10A, TRMT5, TRPM6, TRPM7, TRPS1, TRPV4, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFN, TSHB, TSHR, TSPAN7, TTBK2, TTC19, TTC21B, TTC8, TTI2, TTN, TTP1, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWNK, TYMP, TYROBP, UBA1, UBA5, UBE2A, UBE3A, UBQLN2, UCHL1, UMPS, UNC80, UNC93B1, UPAP1, UPB1, UPF3B, UQCRB, UQCRQ, UROD, UROS, USP27X, USP8, USP9X, VAMP1, VANGL1, VAPB, VARS2, VAX1, VCP, VDAC1, VEGFA, VHL, VIPAS39, VLDLR, VMA21, VPS11, VPS13A, VPS13B, VPS35, VPS37A, VPS53, VRK1, WAC, WASHC5, WDPCP, WDR26, WDR45, WDR48, WDR62, WDR73, WDR81, WFS1, WNK1, WNT10A, WNT3, WNT5A, WNT7A, WT1, WWOX, XBP1, XK, XPR1, XRCC4, YAP1, YARS1, YWHAE, YY1, ZBTB16, ZBTB18, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZFR, ZFYVE26, ZFYVE27, ZIC2, ZIC3, ZMYM3, ZNF335, ZNF41, ZNF423, ZNF674, ZNF711, ZNF81

The *PRODH, NEB*, and *SIK1* genes are not fully covered by this test, therefore pathogenic variants may not be detected in these genes.