variantyx

Genomic Unity® Epilepsy Analysis

Overview

Genomic Unity® Epilepsy Analysis is an effective test for the genetic cause of epilepsy in patients with clinical symptoms consistent with the three general classes of epilepsies: genetic generalized epilepsies (GGE), focal epilepsies, epileptic encephalopathy (EE). As well as their specific syndromes and epilepsies associated or co-morbid with neurodevelopmental conditions including intellectual disability.

Genomic Unity® Epilepsy Analysis also tests for metabolic conditions that present with seizures.

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

- ✓ Sequence analysis of seizure associated genes
- ✓ Del/dup analysis of seizure associated genes
- ✓ STR analysis of: *AFF2, AFF3, CSTB, 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 to Genomic Unity® Exome Plus Analysis

In the case that Genomic Unity® Epilepsy Analysis does not identify causal variants, the option is given to reflex up to Genomic Unity® Exome Plus 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

AARS1, ABAT, ABCD1, ACY1, ADAR, ADGRG1, ADSL, AFF2, AFF3, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A2, ALDH7A2, ALG13, ALG9, ALPL, AMACR, AMT, ANKRD11, AP4B1, AP4E1, AP4M1, AP4S1, ARFGEF2, ARG1, ARHGEF9, ARSA, ARV1, ARX, ASAH1, ASNS, ASPA, ASPM, ATP13A2, ATP1A2, ATP1A3, ATP2A2, ATP6AP2, ATP6V0A2, ATRX, BCKDK, BRAT1, BTD, C12ORF57, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CARS2, CASK, CASR, CC2D1A, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COA8, COL4A1, COX15, COX6B1, CP2, CPA6, CSF1R, CSTB, CTC1, CTFD, CTNNB1, CTSF, CUL4B, CYP27A1, D2HGDH, DARS1, DARS2, DCX, DDX3X, DEAF1, DEPDC5, DHFR, DIP2B, DNAJC5, DNM1, DOCK7, DPYD, DPYS, DYNC1H1, DYRK1A, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMX2, EPM2A, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FGD1, FGF12, FGFR3, FH, FKRP, FKTN, FLNA, FMR1, FOLR1, FOXG1, FOXRED1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GALC, GAMT, GATM, GCDH, GCH1, GCSH, GFAP, GFM1, GJC2, GLB1, GLDC, GLRA1, GNAO (GNOA1), GNB1, GNE, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRN, GTPBP3, GUF1, HACE1, HCN1, HCN4, HECW2, HEPACAM, HIBCH, HNRNPU, HPRT1, HSD17B10, HSPB1, HTRA1, HTT, IBA57, IER3IP1, IQSEC2, ITPA, JMJD1C, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD17, KDM5C, KDM6A, KIF1A, KIFBP, KMT2D, L2HGDH, LAMA2, LARGE1, LGI1, LIAS, LMNB1, LMNB2, LRPPRC, MAGI2, MBD5, MCPH1, MCPH2, MED12, MEF2C, MFSD8, MLC1, MOCS1, MRPL44, MTFMT, MTOR, NACC1, NALCN, NDE1, NDUFA1, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEU1, NEXMIF, NFU1, NGLY1, NHLRC1, NIPBL, NOTCH3, NPRL3, NR2F1, NRXN1, NUBPL, OFD1, OPHN1, PAFAH1B1, PAK3, PCDH19, PEX7, PGK1, PHF6, PHGDH, PIGN, PIGO, PIGT, PIGV, PLCB1, PLP1, PNKD, PNKP, PNPO, POLG, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PPP2R5D, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRIMA1, PRODH, PRRT2, PSAP, PTS, PURA, PYCR2, QARS1, QDPR, RAB39B, RAB3GAP1, RAI1, RARS1, RELN, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPINI1, SETBP1, SETD2, SGCE, SHH, SIK1, SIX3, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNORD118, SOX10, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STIL, STRADA, STX1B, STXBP1, SUCLA2, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBX1, TCF4, TPK1, TPP1, TREX1, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2B, TUBB4A, UBA5, UBE2A, UBE3A, UNC80, VPS13A, VPS13B, WDR26, WDR45, WDR62, WWOX, YY1, ZEB2, ZFYVE26

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