variantyx

Genomic Unity® Intellectual Disability Analysis

Overview

Genomic Unity® Intellectual Disability Analysis is an effective test for the genetic cause of both syndromic and non-syndromic intellectual disabilities.

It is an effective test for X-linked causes including: Alpha-thalassemia X-linked ID syndrome, CASK-related ID syndrome, Coffin-Lowry syndrome, **Fragile X syndrome**, Lujan syndrome, Monoamine oxidase deficiency, Partington syndrome and more. As well as autosomal causes including: ADNP syndrome, Cohen syndrome, Koolen-de Vries syndrome, PACS1 syndrome, SETBP1 disorder, Smith-Kingsmore syndrome, White-Sutton syndrome, Xia-Gibbs syndrome and more.

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 X-linked and autosomal intellectual disability associated genes
- ✓ Del/dup analysis of X-linked and autosomal intellectual disability associated genes
- ✓ Early-onset intellectual disability disorder 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

- 99.7% 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

Turnaround time

6-8 weeks after sample receipt

Reflex to Genomic Unity® Exome Plus Analysis

In the case that Genomic Unity® Intellectual Disability 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

ABCD1, ACSL4, ADNP, AFF2, AFF3, AGA, AHDC1, AIFM1, ALG13, AMMECR1, ANKRD11, AP1S2, AP4B1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARSE, ARX, ASXL1, ASXL3, ATP6AP2, ATP7A, ATRX, AUT22, BCAP31, BCOR, BCORL1, BDNF, BRWD3, CA8, CACNA1A, CASK, CC2D1A, CCDC22, CDKL5, CHD2, CHD7, CHD8, CLCN4, CNKSR2, CNTNAP2, CREBBP, CSNK2A1, CTCF, CTNNB1, CTNND2, CUL4B, DCX, DDX3X, DHCR7, DIP2B, DKC1, DLG3, DMD, DNM1, DNMT3A, DYNC1H1, DYRK1A, EBP, EFTUD2, EHMT1, EIF2S3, EP300, EZH2, FANCB, FGD1, FLNA, FMR1, FOLR1, FOXG1, FOXP1, FRMPD4, FTCD, FTSJ1, GAMT, GATAD2B, GATM, GDI1, GK, GNAO1, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HAL, HCCS, HCFC1, HCN1, HDAC6, HDAC8, HIVEP, HMGB3, HNRNPH2, HNRNPU, HOXA1, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IKBKG, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KCNB1, KCNJ10, KDM5C, KDM6A, KIF1A, KIF4A, KLF8, KLHL15, KMT2A, KMT2D, L1CAM, LAMP2, LAS1L, LINS1, MAGT1, MAN1B1, MAOA, MAP2K1, MBD5, MBTPS2, MECP2, MED12, MED13L, MED23, MEF2C, MID1, MID2, MTM1, MTOR, MYT1L, NAA10, NALCN, NDP, NDUFA1, NEXMIF, NF1, NHS, NIPBL, NLGN3, NLGN4X, NONO, NR2F1, NRXN1, NSD1, NSDHL, NSF5, NSUN2, OCRL, OFD1, OGT, OPHN1, OTC, PACS1, PAK3, PAX6, PCDH19, PDHA1, PGAP2, PGK1, PHF6, PHF8, PIGA, PIGO, PIGV, PLA2G6, PLP1, PNKP, POGZ, POLA1, PORCN, PPP2R5D, PPT1, PQBP1, PRPS1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAI1, RBM10, RIT1, RLIM, RNF113A, RPL10, RPS6KA3, SATB2, SCN1A, SCN2A, SCN8A, SETBP1, SETD5, SHANK3, SHROOM4, SLC16A2, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMC1A, SMC3, SMS, SOS1, SOX3, SRPX2, SSR4, ST3GAL3, STAG2, STXBP1, SYN1, SYNGAP1, SYP, TAF1, TBC1D24, TBL1XR1, TBR1, TCF4, THOC2, TIMM8A, TMLHE, TRAPPC9, TRIO, TSC1, TSC2, TSPAN7, TUSC3, UBE2A, UBE3A, UPF3B, USP27X, USP9X, VPS13B, WAC, WDR45, WT1, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZMYM3, ZNF41, ZNF711, ZNF81