variantyx

Genomic Unity® Endocrinology Analysis

EA001

Overview

Patients with endocrine disorders often present with heterogeneous phenotypes given that one bodily function can be regulated by multiple hormones and that one hormone can regulate multiple bodily functions.

Genomic Unity® Endocrinology Analysis is an effective test for the genetic cause of suspected endocrine disorders including: Adrenal hypoplasia congenita; Adrenoleukodystrophy; Glucocorticoid deficiency; Familial hypocalciuric hypercalcemia; Hypothyroidism and resistance to thyroid hormone; Kallmann syndrome; Liddle's syndrome; Monogenic diabetes (MODY) 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 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 genes associated with endocrine disorders
- ✓ Del/dup analysis of genes associated with endocrine disorders

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

Reflex option

In the case that Genomic Unity® Endocrinology 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

ABCA1, ABCC8, ABCD1, ABCG5, ABCG8, ACAT1, ACSF3, ACTN4, ADCY3, AFF3, AGL, AIRE, ALDOA, ALDOB, ALMS1, AMH, AMHR2, ANOS1, AP2S1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, AQP2, AR, ARL6, ARMC5, ARX, ATRX, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BLK, BMP15, BSND, CASR, CCDC28B, CDC73, CDKN1A, CDKN1B, CDKN1C, CDKN2B, CDKN2C, CEL, CEP290, CEP41, CHD7, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, COL1A1, COL1A2, CREB3L3, CREBBP, CUL4B, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DUOX2, DUOXA2, DUSP6, DYNC2H1, DYRK1B, EGF, EIF2AK3, ENO3, EPM2A, ERCC3, ETFA, ETFB, ETFDH, FBP1, FEZF1, FGF17, FGF23, FGF8, FGFR1, FIG4, FLRT3, FOXE1, FOXL2, FOXP3, FRAS1, FSHB, FSHR, FXYD2, G6PC, G6PC2, GAA, GALT, GATA4, GATA6, GBE1, GCK, GCM2, GH1, GHR, GLIS3, GLUD1, GNA11, GNAS, GNRH1, GNRHR, GPIHBP1, GYG1, GYS1, GYS2, HADH, HESX1, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HS6ST1, HSD11B2, HSD17B3, HSD3B2, IFT27, IGSF1, IL17RD, INF2, INS, INSR, IPF1, IRF6, KAL1, KCNA1, KCNJ10, KCNJ11, KISS1, KISS1R, KLF11, KRAS, KSR2, LAMB2, LAMP2, LDHA, LDLR, LDLRAP1, LEPR, LEPR, LHB, LHCGR, LIPA, LMF1, LMNA, LPL, LRP5, LZTFL1, MAGEL2, MAGT1, MAMLD1, MAP3K1, MC2R, MC3R, MEN1, MEN2, MKKS, MKRN3, MKS1, MPV17, MRAP, NEUROD1, NEUROG3, NF1, NHLRC1, NIPA2, NKX2-1, NKX2-2, NKX2-5, NNT, NOBOX, NPHS1, NPHS2, NPSH2, NR0B1, NR0B2, NR3C1, NR5A1, NROB1, NSMF, NTRK2, OXCT1, PAX4, PAX8, PC, PCBD1, PCK1, PCSK1, PCSK9, PDX1, PFKM, PGAM2, PGK1, PGM1, PHEX, PHF6, PHKA1, PHKA2, PHKB, PKD1, PKD2, PLCE1, POLG, POMC, PORCN, POU1F1, PPARG, PRKAG2, PRKAG3, PRKAR1A, PROK2, PROKR2, PROP1, PTF1A, PTH, PTPN11, PYGL, PYGM, RAF1, RBCK1, RET, RFX6, RSPO1, SARS2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SDHB, SECISBP2, SEMA3A, SEMA3E, SHOX, SIM1, SLC12A3, SLC16A1, SLC16A2, SLC26A4, SLC2A2, SLC37A4, SLC5A5, SOS1, SOX9, SPRY4, SRD5A2, SRY, STAR, TAC3, TACR3, TCF1, TCF2, TG, THRA, THRB, TPO, TRIM32, TRPC6, TRPM6, TSHB, TSHR, TTC8, UCP2, UCP3, VHL, VPS13B, WDPCP, WDR11, WFS1, WT1, ZFP57, ZFPM2

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