variantyx

Genomic Unity® Prenatal Analysis

Overview

Genomic Unity® Prenatal Analysis is a clinical diagnostic test designed to identify genetic variants that correlate with clinical symptoms manifested in a fetus or a pregnancy, or lead to severe early onset genetic disorders.

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 reported pregnancy findings and family history and generate a unified clinical report.

Included analyses

- ✓ Aneuploidy analysis of chromosomes 13, 18, 21, X and Y
- ✓ Uniparental disomy (UPD) and loss of heterozygosity (LOH)
- ✓ Sequence analysis of congenital or pediatric disease genes
- ✓ Genome-wide copy number variant (CNV) analysis
- ✓ STR analysis of: *AR* (Spinal bulbar muscular atrophy), *DMPK* (Myotonic dystrophy), *FMR1* (Fragile X), *FXN* (Friedreich's ataxia)

Maternal cell contamination (MCC) is determined for quality control purposes. Parental inheritance will be identified for reported variants.

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 35 bp

- 99.739% sensitivity
- 99.999% specificity
- 99.657% 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

Highly sensitive detection of pathogenic STRs

- >99% clinical sensitivity

Accepted sample types

- Amniotic fluid (20 ml), cells cultured from amniotic fluid, or fetal genomic DNA plus parental blood (5ml)
- Amniotic fluid samples should be collected between pregnancy weeks 15-18

Turnaround time after sample receipt

A preliminary report* based on FISH will be issued within 4 days for aneuploidies of 13, 18, 21, X or Y. A complete report will be issued as follows:

- 20 days for cutured cells or genomic DNA
- 30 days for amniotic fluid

*not available for genomic DNA samples

Genes analyzed

AAAS, ABCA12, ABCB11, ABCC6, ABCC8, ABCC9, ABCD1, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACTA1, ACTB, ACTG1, ADA, ADAMTS2, ADGRG1, AFF2, AGA, AGL, AGPS, AGXT, AHCY, AHI1, AIRE, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG6, ALMS1, ALPL, ALX4, AMER1, AMN, AMPD2, AMT, ANO5, ANOS1, APOPT1/COA8, AQP2, AR, ARG1, ARL13B, ARL3, ARMC9, ARSA, ARSB, ARSE, ARX, ASL, ASNS, ASPA, ASPM, ASS1, ATM, ATP5A1/ATP5F1A, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATR, ATRX, B3GALNT2, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BMP1, BMPR2, BRAF, BSND, BTD, BTK, C5ORF42/CPLANE1, CACNA1C, CANT1, CAPN3, CASK, CBL, CBS, CC2D2A, CDH23, CDKL5, CDKN1C, CECR1, CEP104, CEP120, CEP290, CEP41, CERKL, CFC1, CFTR, CHD2, CHD7, CHM, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST3, CIITA/MHC2TA, CIT, CLCN5, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, CNTN1, CNTNAP1, CNTNAP2, COL11A1, COL11A2, COL17A1, COL1A1, COL1A2, COL27A1, COL2A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COMP, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CRB1, CREBBP, CRTAP, CSPP1, CTNNB1, CTNS, CTSD, CTSK, CUL4B, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP27A1, CYP27B1, DAG1, DBT, DCLRE1C, DCX, DHCR7, DHDDS, DHH, DKC1, DLD, DLG3, DLL3, DMD, DMPK, DMXL2, DNAH5, DNAI1, DNAI2, DNM2, DOK7, DUOX2, DYM, DYNC2H1, DYRK1A, DYSF, EBP, ECEL1, EDA, EDNRB, EGR2, EHMT1, EIF2AK3, EIF2B5, ELN, EMD, ENG, EP300, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXT1, EXT2, EYA1, EYS, F11, F2, F8, F9, FAH, FAM149B1, FAM161A, FANCA, FANCB, FANCC, FANCD2, FANCG, FBN1, FGD1, FGD4, FGF8, FGFR1, FGFR2, FGFR3, FH, FIG4, FKBP10, FKRP, FKTN, FLNA, FLNB, FMR1, FOXC1, FOXC2, FOXF1, FOXG1, FOXH1, FOXP2, FOXP3, FOXRED1, FXN, FZD4, G6PC, G6PD, GAA, GABRB3, GABRD, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATA3, GATA4, GATA6, GBA, GBE1, GCDH, GCH1, GDAP1, GDF1, GDF5, GDI1, GFM1, GH1, GJA1, GJA8, GJB1, GJB2, GJB3, GJB6, GK, GLA, GLB1, GLDC, GLE1, GLI2, GLI3, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GP1BA, GP9, GPC3, GPC6, GRHPR, GRIA3, GRIP1, GUCY2C, GUCY2D, GUSB, H19, HADH, HADHA, HADHB, HAX1, HBA1, HBA2, HBB, HCCS, HCFC1, HDAC8, HEXA, HEXB, HFE2/HJV, HGD, HGSNAT, HK1, HLCS, HMGCL, HMOX1, HNF1B, HOGA1, HOXD13, HPD, HPRT1, HPS1, HPS3, HPS4, HPSE2, HRAS, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSPG2, HUWE1, HYLS1, IDS, IDUA, IFITM5, IFT172, IGF1R, IGF2, IGF2R, IGHMBP2, IKBKAP/ELP1, IKBKG, IL1RAPL1, IL2RG, INPP5E, INPPL1, INSR, IRF6, ISPD, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAK3, KATNB1, KCNJ1, KCNJ11, KCNJ2, KCNK9, KCNQ1, KCNQ1OT1, KDM5C, KDM6A, KIAA0556/KATNIP, KIAA0586, KIAA2022/NEXMIF, KIF7, KLHL40, KMT2D, KRAS, L1CAM, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LAMP2, LARGE1/LARGE, LBR, LCA5, LDLR, LDLRAP1, LEMD3, LGI4, LHX3, LIFR, LIPA, LMBRD1, LMNA, LMOD3, LMX1B, LOXHD1, LPL, LRPPRC, LSS, LYRM7, LYST, MAGEL2, MAN2B1, MAOA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MECP2, MED12, MED13L, MED17, MEFV, MEN1, MESP2, MFN2, MFSD8, MID1, MID2, MITF, MKRN3, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MPI, MPL, MPV17, MPZ, MRE11A, MSX1, MTFMT, MTHFR, MTM1, MTR, MTRR, MTTP, MUSK, MUT/MMUT, MVK, MYBPC1, MYCN, MYH3, MYH7, MYO15A, MYO7A, NAGLU, NAGS, NBN, NCF1, NDE1, NDN, NDP, NDRG1, NDUFA1, NDUFA10, NDUFA12, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NEK1, NEU1, NF1, NF2, NIPBL, NKX2-5, NOTCH1, NPC1, NPC2, NPHP1, NPHP3, NPHS1, NPHS2, NR0B1, NR2E3, NR5A1, NRAS, NRXN1, NSD1, NTRK1, NUBPL, OAT, OCA2, OCRL, OFD1, OPA3, OPHN1, OTC, OTX2, P3H1, PAFAH1B1, PAH, PANK2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PEPD, PET100, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PHF21A, PHF6, PHGDH, PHOX2B, PIBF1, PIEZO2, PIGN, PITX2, PKD1, PKD2, PKHD1, PLA2G6, PLEC/PLEC1, PLOD2, PLP1, PMM2, PMP22, PNPO, POLG, POLR1C, POLR1D, POMGNT1, POMGNT2, POMT1, POMT2, POP1, POR, PORCN, PPIB, PPT1, PQBP1, PREPL, PRODH, PROP1, PRPS1, PSAP, PSAT1, PTCH1, PTEN, PTPN11, PTS, PUS1, PYGM, QDPR, RAB23, RAD21, RAF1, RAG2, RAI1, RAPSN, RARS2, RB1, RBM10, RBM8A, RDH12, RELN, RET, RIPK4, RIT1, RLBP1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, ROR2, RP2, RPE65, RPGRIP1L, RPL10, RPS19, RPS6KA3, RS1, RTEL1, RUNX2, RYR1, SACS, SALL1, SALL4, SAMD9, SAMHD1, SATB2, SBDS, SBF2, SCN1A, SCN5A, SCO1, SDHA, SDHAF1, SDHD, SEPN1/SELENON, SEPSECS, SERAC1, SERPINA1, SERPINH1, SGCA, SGCB, SGCD, SGCE, SGCG, SGSH, SH2D1A, SH3TC2, SHANK3, SHH, SHOC2, SHOX, SHROOM4, SIL1, SIX1, SIX3, SIX5, SIX6, SKI, SLC12A1, SLC12A3, SLC12A6, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A24, SLC25A4, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC35A3, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A11, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMAD4, SMARCAL1, SMC1A, SMC3, SMN1, SMN2, SMPD1, SMS, SNRPN, SNTA1, SOS1, SOST, SOX10, SOX2, SOX3, SOX9, SPRED1, SRD5A2, SRPX2, SRY, ST3GAL5, STAR, SUCLA2, SUFU, SUMF1, SUOX, SURF1, SUZ12, SYN1, TAT, TAZ, TBCE, TBX1, TBX5, TCAP, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TECPR2, TERT, TG, TGFBR1, TGFBR2, TGIF1, TGM1, TH, THRA, TIMM8A, TMEM107, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM38B, TMEM5/RXYLT1, TMEM63A, TMEM67, TMEM70, TNNC1, TNNI2, TNNI3, TNNT1, TNNT2, TNNT3, TNXB, TP53, TP63, TPM1, TPM2, TPM3, TPO, TPP1, TREX1, TRIM32, TRIM37, TRIP11, TRMU, TRPS1, TRPV4, TSC1, TSC2, TSEN54, TSFM, TSHB, TSHR, TTC19, TTC21B, TTC37, TTN, TTPA, TUBA1A, TUBB2B, TUBB3, TWIST1, TYMP, TYR, TYRP1, UBE3A, UGT1A1, UNC13D, UPF3B, USH1C, USH1G, USH2A, VHL, VLDLR, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, VWF, WAS, WDR34, WDR35, WDR62, WISP3, WNT10A, WNT7A, WRN, WT1, XIAP, XPA, XPC, ZC4H2, ZDHHC9, ZEB2, ZFPM2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMPSTE24