The Croucher Laboratory for Human Genomics (CLHG)

The Chinese University of Hong Kong

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The Whole Exome Sequencing (WES) Evaluation Study – Circle PREMIUM

Aim

CLHG evaluates the accuracy of Prenetics' the Whole Exome Sequencing (WES) test on Circle PREMIUM by single-blind research method and using genomic DNA samples from the HapMap human diversity panels with the reference genotypes obtained by querying the 1000 Genomes database for tested positions.

Result

CLHG has analyzed a total of 5,580 genes related to 500+ reported categories that cover 33,817 variant sites. Out of 1,600,801 variant calls/ SNP in which reference genotype information was available, the WES test could correctly call 1,598,676 variants/ SNP. The evaluation result indicated that the WES test is 99.9% accurate analytically on those target genes.

Analyzed Genes

ABCB11, ABCC8, ACADM, ACADS, ACADVL, ADGRV1, AGA, AGL, AGXT, AIP, AIRE, ALDH3A2, ALDOB, ALK, ALPL, APC, ARSA, ASPA, ASS1, ATF6, ATM, ATP7B, AXIN2, BAP1, BARD1, BBS1, BBS10, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BTD, BUB1B, CAPN3, CAV3, CBL, CBS, CCND1, CDC73, CDH1, CDH23, CDK4, CDKN1B, CDKN2A, CEBPA, CFTR, CHEK2, CHM, CIB2, CLN3, CLN5, CLN8, CLRN1, CNGA3, CNGB3, COL7A1, COQ8B, CPT1A, CPT2, CTNS, CTSK, CUBN, CYLD, CYP17A1, DBT, DDB2, DHCR7, DIS3L2, DKC1, DLD, DMD, DOCK8, DPYD, DYSF, EGFR, ELAC2, ELANE, ELP1, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, F11, FAH, FANCA, FANCC, FANCD2, FANCF, FANCG, FAS, FH, FKTN, FLCN, G6PC, G6PD, GAA, GALC, GALT, GATA2, GBA, GCDH, GCH1, GCHFR, GJB2, GJB3, GNAT2, GNE, GPC3, GREM1, GRHPR, H19, HADH, HADHA, HBA1, HBA2, HBB, HEXA, HFE, HGD, HMBS, HNF1A, HOGA1, HOXB13, HRAS, HSD17B4, IDUA, ITK, IVD, KCNJ11, KCNQ1OT1, KIT, KLHL40, KRAS, LAMA3, LAMB2, LAMB3, LAMC2, LMX1B, MAN2B1, MAX, MC1R, MCOLN1, MEFV, MEN1, MET, MITF, MLC1, MLH1, MLH3, MN1, MPI, MSH2, MSH6, MTHFR, MUTYH, MYO7A, NBN, NDUFA13, NEB, NF1, NF2, NPC1, NPHS1, NPHS2, NRAS, OPA3, PAH, PALB2, PAX5, PCBD1, PCDH15, PDE6C, PDE6H, PDGFB, PDGFRA, PDZD7, PEX1, PEX7, PHOX2B, PKHD1, PLCE1, PMM2, PMS2, POLD1, POLE, POLH, POMGNT1, POT1, PPT1, PRKAR1A, PROP1, PRSS1, PTCH1, PTEN, PTPN11, PTS, PYGM, QDPR, RAD51C, RAD51D, RB1, RECQL, RECQL4, RET, RHBDF2, RMRP, RNASEL, RS1, RUNX1, SACS, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SGCA, SGCB, SH2D1A, SLC12A6, SLC17A5, SLC22A5, SLC25A13, SLC26A2, SLC26A4, SLC37A4, SMAD4, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMPD1, SOS1, SRD5A2, SRGAP1, STAG1, STAT3, STK11, SUFU, TERT, TGFBR1, TGFBR2, TH, TMEM127, TMEM216, TP53, TPP1, TRIM37, TSC1, TSC2, TTPA, UROD, USH1C, USH1G, USH2A, VHL, VPS13B, WAS, WHRN, WRN, WT1, XPA, XPC, XRCC3, ZNF783 and 5,415 analyzed genes.

Conclusion

In this evaluation study, the result showed that Prenetics' Whole Exome Sequencing (WES) test on Circle PREMIUM with 99.9% analytical accuracy.

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