The Croucher Laboratory for Human Genomics (CLHG)
The Chinese University of Hong Kong

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The Whole Exome Sequencing (WES) Evaluation Study – Cancer Screening

Aim

CLHG evaluates the accuracy of Prenetics' the Whole Exome Sequencing (WES) test on cancer screening 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 140 genes related to 36 hereditary cancers that cover 4,735 variant sites. Out of 232,015 variant calls in which reference genotype information was available, the WES test could correctly call 231,954 variants. The evaluation result indicated that the WES test is 99.97% accurate analytically on those target cancer genes.

Analyzed Genes

ABCB11, AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, COL7A1, CYLD, DDB2, DIS3L2, DKC1, DOCK8, EGFR, ELAC2, ELANE, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, FAH, FANCA, FANCC, FANCD2, FANCF, FANCG, FH, FLCN, GATA2, GBA, GPC3, GREM1, H19, HFE, HMBS, HNF1A, HOXB13, HRAS, ITK, KCNQ1OT1, KIT, KRAS, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MSH2, MSH6, MUTYH, NBN, NDUFA13, NF1, NF2, NRAS, PALB2, PAX5, PDGFB, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PRSS1, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RB1, RECQL, RECQL4, RET, RHBDF2, RMRP, RNASEL, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SH2D1A, SLC25A13, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SRD5A2, SRGAP1, STAG1, STAT3, STK11, SUFU, TERT, TGFBR1, TGFBR2, TMEM127, FAS, TP53, TRIM37, TSC1, TSC2, UROD, VHL, WAS, WRN, WT1, XPA, XPC, XRCC3, and ZNF783.

Conclusion

In this evaluation study, the result showed that Prenetics' Whole Exome Sequencing (WES) test on cancer screening with 99.97% analytical accuracy.

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