麗寶醫事檢驗所
LIHPAO Medical Laboratory

神經系統疾病基因檢測

Neurological Disease Genetic Testing

檢測報告

神經系統疾病基因檢測

檢體資訊

報告編號:	**檢體類別:**	**委託單位:**
檢體編號:	**採檢日期:**	**聯 絡 人:**
病歷編號:	**收檢日期:**	**聯絡人電話:**
採檢單號:	**報告日期:**	**聯絡人信箱:**
檢測方法:	**委託醫師:**	

檢測報告總結

檢體品質備註

檢體品質需符合品管標準方得執行後續檢測，除特別標註外，皆代表樣本通過品管要求。

報: 張本樺 2024-09-02

報告簽署人: **報告簽署人註解:**

檢測結果

編號	染色體	基因 / 外顯子	變異位點	AAF	合子型式	致病力

備註:
檢測結果呈現美國醫學遺傳學暨基因體學學會 (ACMG) 指引或 ClinVar 資料庫判定為致病性/可能致病性 (Pathogenic/Likely Pathogenic) 的變異位點。AAF (Alternative Allele Frequency) 紀錄該變異位點在東亞族群的等位基因頻率。合子型式包含 Homo (Homozygote 同型合子)、Het (Heterozygote 異型合子)、Hemi (Hemizygote 半合子)。詳細說明請參考檢測結果注釋。

檢測位點注釋

產品介紹

神經系統疾病成因複雜且診斷困難，不同的基因變異可能出現類似的症狀，在這個基因體醫學時代以全外顯子定序掃描為基礎，探討基因相關的神經遺傳疾病，有助於突破傳統上以臨床表徵歸納進行診斷所遇到的困境。全外顯子基因檢測可合併常規檢測，發展更完整的神經疾病診斷模式，避免患者接受冗長的檢查與不適合或不必要的治療方案，減少醫療資源的浪費。本檢測廣泛的分析 1915 個基因，找出相關的致病性變異 (Pathogenic Variant) 及可能致病性變異 (Likely Pathogenic Variant)，提供臨床醫師對相關疾病的評估與參考。

檢驗項目

威爾森氏症	Wilson's Disease
癲癇	Epilepsy
腦小血管疾病	Cerebral Small Vessel Disease
脊髓側索硬化	Amyotrophic Lateral Sclerosis
希佩爾 - 林道症候群	Von-Hippel-Lindau Disease
體顯性腦動脈血管病變合併皮質下腦梗塞及腦白質病變	Cerebral Autosomal Dominant Arteriopathy with

檢驗項目

	Subcortical Infarcts and Leukoencephalopathy
妥瑞症候群	Tourette's Syndrome
認知障礙	Cognitive Disorder
多發性系統退化症	Multiple System Atrophy
家族性澱粉樣多發性神經病變	Familial Amyloid Polyneuropathy
多發性神經纖維瘤	Neurofibromatosis
共濟失調	Ataxia
結節性硬化症	Tuberous Sclerosis
腦白質失養症	Leukodystrophy
夏柯 - 馬利 - 杜斯氏症	Charcot-Marie-Tooth Disease
溶小體儲積症	Lysosomal Storage Disease
MELAS症候群	MELAS Syndrome
肌張力不全症	Dystonia
原發性側索硬化	Primary Lateral Sclerosis
遺傳性痙攣性下身麻痺	Hereditary Spastic Paraplegia

基因列表

A2M, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA2, ABCA4, ABCA5, ABCA7, ABCB1, ABCB7, ABCC2, ABCC6, ABCC8, ABCD1, ABCG5, ABCG8, ABHD12, ABHD5, ABI3, ACAD11, ACADM, ACADS, ACAT1, ACBD5, ACD, ACER3, ACO2, ACOX1, ACSF3, ACTB, ACTG2, ACTL6B, ADA2, ADAM10, ADAM22, ADAM28, ADAMTS1, ADAMTS5, ADAMTSL4, ADAR, ADARB1, ADCY5, ADCY6, ADD3, ADGRG1, ADGRV1, ADH1C, ADORA2A, ADPRS, ADRA2B, ADSL, AFF3, AFF4, AFG3L2, AGL, AGRN, AGTPBP1, AGXT, AHCY, AHDC1, AHI1, AHNAK2, AIFM1, AIMP1, AIMP2, AKAP9, AKT1, AKT3, ALAD, ALAS2, ALDH18A1, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALOX12B, ALOXE3, ALS2, AMACR, AMER1, AMPD2, ANG, ANGPTL3, ANK1, ANK2, ANKRD11, ANO10, ANO3, ANOS1, ANXA11, AOPEP, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5B1, AP5Z1, APC, APCS, APOA1, APOA2, APOA5, APOB, APOC2, APOE, APOO, APP, APTX, AR, ARCN1, ARF1, ARHGEF10, ARHGEF15, ARHGEF9, ARID1B, ARL13B, ARL3, ARL6, ARL6IP1, ARL6IP6, ARMC9, ARSA, ARSB, ARSG, ARSI, ARSK, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASS1, ASXL1, ATAD1, ATCAY, ATG5, ATG7, ATL1, ATL3, ATM, ATN1, ATP10A, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B2, ATP2B3, ATP5F1E, ATP5MC3, ATP5MD, ATP5PF, ATP6V0A2, ATP6V1A, ATP6V1B2, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRIP, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B2M, B3GALNT2, B4GALNT1, B9D1, BAG3, BAP1, BBIP1, BBS1, BBS10, BBS12, BBS5, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL10, BCS1L, BDNF, BEAN1, BICD2, BIN1, BMP15, BNC1, BOLA3, BRAF, BRAT1, BRSK2, BSCL2, BTD, C11orf65, C12orf4, C12orf65, C19orf12, C1R, C9orf72, CA2, CA8, CABP4, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CACNB4, CACNG2, CAD, CADM3, CAMK2A, CAMK2B, CAMTA1, CAPN1, CARS1, CARS2, CASK, CASR, CAV1, CBY1, CC2D1A, CC2D2A, CCDC141, CCDC183, CCDC28B, CCDC40, CCDC78, CCDC88A, CCDC88C, CCM2, CCND1, CCNF, CCR1, CCT5, CD28, CD59, CDC40, CDC42, CDH15, CDH23, CDK19, CDKL5, CDKN1A, CDKN1B, CDKN2B, CDKN2C, CDON, CELF2, CELSR3, CENPE, CENPJ, CEP104, CEP120, CEP152, CEP164, CEP290, CEP41, CEP78, CFAP410, CFAP43, CFH, CFHR3, CHAMP1, CHAT, CHD2, CHD7, CHMP2B, CHP1, CHRM3, CHRNA1, CHRNA2, CHRNA4, CHRNB2, CHRNB4, CHRNG, CIB2, CIC, CIITA, CILK1, CISD2, CIZ1, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN4, CLCN6, CLCNKB, CLDN11, CLIP1, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLRN1, CLTC, CLTCL1, CLTRN, CLU, CNKSR2, CNNM2, CNP, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA3, COA7, COA8, COASY, COBLL1, COG4, COG5, COG8, COL12A1, COL13A1, COL18A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A3, COL7A1, COL9A3, COLGALT1, COMP, COPB1, COQ2, COQ4, COQ7, COQ8A, COQ9, COX10, COX15, COX20, COX4I1, COX6A1, COX6B1, COX7B, CP, CPA6, CPLANE1, CPOX, CPS1, CPT1C, CRADD, CRAT, CRBN, CRH, CRLF1, CSF1R, CSNK1E, CSPP1, CSRNP3, CST3, CSTB, CTBP1, CTC1, CTDP1, CTLA4, CTNNA2, CTNND2, CTNS, CTSD, CUBN, CUL4B, CUX1, CUX2, CWF19L1, CXCR4, CYB5A, CYB5R3, CYFIP2, CYLD, CYP1B1, CYP27A1, CYP2U1, CYP4F22, CYP7B1, DAB1, DAG1, DALRD3, DAO, DARS1, DARS2, DBT, DCAF17, DCAF8, DCC, DCHS1, DCLRE1B, DCPS, DCTN1, DCTN2, DCX, DDB2, DDC, DDHD1, DDHD2, DDX20, DEAF1, DECR1, DEGS1, DENND5A, DEPDC5, DGAT2, DGUOK, DHCR24, DHDDS, DHFR, DHH, DHTKD1, DHX30, DIAPH1, DIAPH3, DIS3L2, DISP1, DKC1, DKK1, DLAT, DLD, DLG3, DLL1, DLL3, DMD, DMXL2, DNAJB2, DNAJB5, DNAJC13, DNAJC16, DNAJC19, DNAJC3, DNAJC30, DNAJC5, DNAJC6, DNAJC7, DNASE1L3, DNM1, DNM1L, DNM2, DNMT1, DOCK3, DOCK7, DOCK8, DOLK, DPAGT1, DPM1, DPM2, DPYD, DPYSL5, DRD2, DRD3, DSG4, DSP, DST, DSTYK, DUSP6, DYNC1H1, DYRK1A, DYSF, DZIP1L, EARS2, EBF3, ECE1, ECHS1, ECM1, EDC3, EDN3, EDNRB, EEF1A2, EEF2, EFHC1, EGR2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4G1, ELF2, ELFN1, ELOVL4, ELOVL5, ELP1, ELP2, ELP4, EMC1, EMD, ENPP1, ENTPD1, EP300, EPB41L1, EPB41L4A, EPB42, EPHA4, EPHX2, EPM2A, EPRS1, ERAP1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ERMARD, ESR1, ETHE1, EWSR1, EXOSC5, EXOSC8, EZR, FA2H, FAH,

基因列表

FAM126A, FAM149B1, FAM228B, FANCG, FANCI, FAR1, FARS2, FAS, FASN, FAT1, FAT2, FAT4, FBLN1, FBLN5, FBN1, FBXL4, FBXO28, FBXO31, FBXO38, FBXO7, FCSK, FDXR, FEZF1, FGA, FGD1, FGD4, FGF12, FGF13, FGF14, FGF17, FGF8, FGFR1, FGFR3, FH, FIG4, FITM2, FKRP, FKTN, FLII, FLNA, FLNC, FLRT1, FLRT3, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOXG1, FOXH1, FOXI1, FOXRED1, FRMD4A, FRMPD4, FSHR, FTL, FUCA1, FUS, FUZ, FXN, FZR1, G6PC, GAA, GABBR2, GABPA, GABRA1, GABRA2, GABRA5, GABRA6, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GALC, GALNS, GALNT3, GALT, GAMT, GAN, GAS1, GATA1, GATM, GBA, GBA2, GBE1, GBF1, GCDH, GCH1, GCK, GCLC, GDAP1, GDAP2, GDF3, GDF5, GDF6, GDNF, GEMIN4, GEMIN5, GFAP, GFM2, GGCX, GHR, GIGYF2, GJA1, GJB1, GJB2, GJB3, GJB6, GJC2, GLA, GLB1, GLDC, GLE1, GLI2, GLI3, GLRA1, GLRB, GLRX5, GLS, GLT8D1, GLUD1, GLUD2, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNAS, GNB1, GNB2, GNB4, GNB5, GNE, GNPTAB, GNS, GOSR2, GOT2, GP1BA, GPAA1, GPC4, GPHN, GPI, GPNMB, GPT2, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, 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MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MTND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TC, MT-TE, MT-TF, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TN, MT-TP, MT-TQ, MT-TS1, MT-TS2, MT-TT, MT-TV, MTTW, MTCL1, MTFMT, MTHFR, MTHFS, MTM1, MTMR10, MTMR14, MTMR2, MTO1, MTOR, MTPAP, MTR, MTRR, MVK, MYBPC1, MYBPC3, MYD88, MYF6, MYH14, MYH3, MYL3, MYLK2, MYO1H, MYO5A, MYO6, MYO7A, MYO9A, MYOC, MYORG, MYOT, MYT1, MYT1L, NAA20, NACC1, NADK2, NAGA, NAGLU, NAGS, NALCN, NANS, NAPB, NARS1, NARS2, NAXD, NAXE, NBEA, NCDN, NCF2, NDNF, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NEFL, NEK1, NELFA, NEMF, NEUROD2, NEXMIF, NF1, NF2, NFIA, NFIX, NGF, NGLY1, NHLRC1, NHLRC2, NHSL2, NID1, NIPA1, NIPA2, NIPAL4, NIPBL, NKX2-1, NKX6-2, NLRP1, NLRP3, NMNAT1, 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PJVK, PKHD1, PKP2, PLA2G6, PLAA, PLAU, PLCB1, PLCD1, PLD3, PLEKHG2, PLEKHG4, PLEKHG5, PLK4, PLOD1, PLP1, PLPBP, PMM2, PMP2, PMP22, PMPCA, PMPCB, PNKD, PNKP, PNP, PNPLA6, PNPLA7, PNPLA8, PNPO, PNPT1, PODXL, POLA1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POLR3H, POLR3K, POMGNT1, POMK, POMT1, POMT2, PON1, PON2, PON3, PORCN, POU1F1, POU3F4, PPARG, PPARGC1A, PPDPF, PPIL1, PPOX, PPP1R15B, PPP1R17, PPP1R21, PPP2R1A, PPP2R2B, PPP2R5D, PPP3CA, PPP6R2, PPT1, PRDX1, PRDX3, PRF1, PRICKLE1, PRICKLE2, PRICKLE3, PRIMA1, PRKAR1A, PRKAR1B, PRKCG, PRKN, PRKRA, PRMT7, PRNP, PRODH, PROK2, PROKR2, PRPH, PRPS1, PRRT2, PRSS12, PRSS48, PRTN3, PRX, PSAP, PSAT1, PSEN1, PSEN2, PSMC3, PSMC3IP, PTCH1, PTEN, PTPN11, PTPN22, PTPN23, PTRH2, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB7A, RAD21, RAD50, RAI1, RAPGEF2, RARS1, RBBP8, RBFOX1, RBFOX3, RBM12, RBM20, REEP1, REEP2, RELN, RET, RETREG1, RFC1, RFT1, RFX5, RFXANK, RFXAP, RHO, RHOBTB2, RIMS2, RIN2, RIPK4, RMND1, RNASE4, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF168, RNF170, RNF216, RNF220, ROBO3, RORA, RORB, RPGRIP1L, RPIA, RPL10, RPL36A-HNRNPH2, RPS24, RRM2B, RRP1B, RS1, RSRC1, RTEL1, RTL9, RTN2, RTN4IP1, RTTN, RUBCN, RYR1, RYR3, S100PBP, SACS, SAMD12, SAMD9L, SAMHD1, SARDH, SARM1, SARS1, SATB1, SATB2, SBF1, SBF2, SCARB2, SCFD1, SCN10A, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCP2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHB, SDHC, SDHD, SDR9C7, SEC23B, SEC61A1, SELENOI, SEMA3A, SEMA6B, SEPSECS, SEPTIN11, SEPTIN9, SERAC1, SERPINC1, SERPING1, SERPINI1, SET, SETBP1, SETD1A, SETD1B, SETX, SGCE, SGCG, SGPL1, SGSH, SH2B1, SH2B3, SH3BP2, SH3TC2, SHANK3,

基因列表

SHFL, SHH, SHMT2, SIGMAR1, SIL1, SIN3A, SIX3, SLC12A2, SLC12A3, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC18A2, SLC18A3, SLC19A2, SLC19A3, SLC1A1, SLC1A2, SLC1A3, SLC1A5, SLC20A2, SLC25A10, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A24, SLC25A4, SLC25A42, SLC25A46, SLC26A1, SLC26A4, SLC2A1, SLC2A3, SLC30A10, SLC30A9, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC37A4, SLC38A11, SLC38A3, SLC39A14, SLC39A4, SLC39A8, SLC44A1, SLC45A1, SLC46A1, SLC4A1, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A6OS, SLC7A7, SLC9A1, SLC9A6, SLIRP, SLITRK1, SMAD4, SMARCA2, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMO, SMPD1, SNAI2, SNAP25, SNAP29, SNCA, SNCAIP, SNCB, SNIP1, SNRPN, SNX14, SNX27, SOD1, SON, SORCS1, SORL1, SOST, SOX10, SOX5, SP110, SP9, SPAG1, SPART, SPAST, SPATA5, SPATA5L1, SPEN, SPG11, SPG21, SPG7, SPIDR, SPOP, SPP1, SPR, SPRED1, SPRY4, SPTA1, SPTAN1, SPTB, SPTBN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SPTLC3, SQSTM1, SRCAP, SRD5A3, SRPX2, SRSF2, SS18L1, ST20-MTHFS, ST3GAL3, ST3GAL5, STAG1, STAG2, STARD7, STAT3, STAT4, STIL, STN1, STRADA, STS, STUB1, STX11, STX16, STX1B, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SULT2B1, SUMF1, SUOX, SURF1, SVBP, SYNE1, SYNE2, SYNJ1, SYT1, SYT14, SYT2, SZT2, TACO1, TACR3, TAF1, TAF15, TANC2, TANGO2, TAOK1, TARDBP, TARS1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBK1, TBL1XR1, TBP, TBR1, TCF20, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TDGF1, TDP1, TDP2, TECPR2, TECR, TELO2, TENM4, TERT, TET2, TEX43, TF, TFG, TGFB1, TGIF1, TGM1, TGM4, TGM6, TH, THAP1, THG1L, THSD1, TIA1, TICAM1, TIMM50, TIMM8A, TIMMDC1, TINF2, TK2, TLR3, TLR4, TMEM106B, TMEM107, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM43, TMEM53, TMEM63A, TMEM67, TMEM70, TMPO, TMPRSS6, TNFRSF1A, TNFRSF1B, TNIK, TNIP1, TNK1, TNK2, TNNI3, TNPO2, TNR, TNRC6A, TOE1, TOMM40, TOP3A, TOPORS, TOR1A, TP53, TP53RK, TPI1, TPK1, TPP1, TPRKB, TRAF3, TRAF3IP1, TRAF7, TRAIP, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2L, TRAPPC4, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM2, TRIM8, TRIO, TRIP4, TRIT1, TRMT5, TRNT1, TRPC3, TRPM1, TRPM3, TRPM7, TRPV4, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHR, TSPOAP1, TTBK2, TTC19, TTC21B, TTC37, TTC5, TTC8, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TXN2, UBA1, UBA5, UBAC2, UBAP1, UBE3A, UBE4A, UBQLN2, UBR7, UBTF, UCHL1, UCP2, UFM1, UFSP2, UGDH, UGP2, UNC13A, UNC13C, UNC13D, UNC80, UPB1, UQCRC1, UQCRQ, URB2, UROC1, USF3, USH1C, USH1G, USH2A, USP19, USP8, VAC14, VAMP1, VAMP2, VAPB, VCP, VHL, VLDLR, VPS11, VPS13A, VPS13C, VPS13D, VPS16, VPS33A, VPS35, VPS37A, VPS41, VPS4A, VPS51, VRK1, VWA1, VWA3B, WARS1, WARS2, WAS, WASF1, WASHC4, WASHC5, WDR11, WDR19, WDR26, WDR4, WDR45, WDR45B, WDR48, WDR73, WDR81, WFS1, WHRN, WIPF1, WNK1, WWOX, XK, XPA, XPC, XPR1, XRCC1, XRCC4, XYLT1, XYLT2, YARS1, YEATS2, YIF1B, YIPF5, YWHAE, YWHAG, YY1, ZBTB11, ZC3H14, ZC4H2, ZEB2, ZFR, ZFYVE26, ZFYVE27, ZIC2, ZMYND11, ZNF142, ZNF236, ZNF423, ZNF592

檢測流程

使用探針系統抓取基因外顯子進行全外顯子定序，平均定序深度為 100X 以上。定序片段會與人類參考基因組 (hg19) 進行序列比對以篩選相關基因變異位點，排除定序深度小於 20X 或等位基因頻率小於 30% 的變異位點後，針對本檢測相關的 1915 個基因進行分析，根據美國醫學遺傳學暨基因體學學會指引 (ACMG Guideline) 與多個資料庫 (gnomAD、ClinVar 等) 判定，致病性或可能致病性變異位點的結果會呈現於報告中。若變異位點與本檢測宣稱的檢驗疾病無關，結果內容會移至意外發現 (Incidental Finding) 變異位點區塊。

資料庫與分析工具版本

Minimap2	2.11-r797-v07
GATK	3.8
VEP	105
RENOVO	1.0
VCFpolyX	2ec35c82e
gnomAD	2.1.1
ClinVar	2024-03-26
rmsk	2020-02-20
HGVS	20.05
Human reference genome	hg19
Biobambam2	2.0.87
Vardict	1.8.2
Taiwan Biobank (TWB)	https://taiwanview.twbiobank.org.tw/browse38

檢測結果注釋

致病力 (Pathogenicity):

(1) 致病性變異 (Pathogenic): 具有充足的證據顯示與疾病的發生有高度相關。
(2) 可能致病性變異 (Likely Pathogenic): 被認為和疾病發生有關，但目前只有較少證據佐證。
(3) 未確定臨床意義變異 (Variant of Uncertain Significance): 在疾病上的影響目前未確定。
(4) 藥物反應變異 (Drug Response): 可能影響用藥效果。
(5) 可能良性變異 (Likely Benign): 被認為和疾病發生無關，但目前只有較少證據佐證。
(6) 良性變異 (Benign): 具有充足的證據顯示與疾病的發生無關。

合子型式 (Zygosity):

(1) 異型合子 (Heterozygote): 指一對染色體上位點的基因型是由不同鹼基所構成。
(2) 同型合子 (Homozygote): 指一對染色體上位點的基因型是由相同鹼基所構成。
(3) 半合子 (Hemizygote): 變異位點出現在僅有一條不成對的染色體基因上。

意外發現變異位點資訊

編號	染色體	基因 / 外顯子	變異位點	AAF	合子型式	致病力
本檢測無意外發現變異位點						

備註:
在本檢測項目涵蓋的基因區段中，發現與本檢測疾病不相關，但可能影響特定疾病發生的變異位點稱之為意外發現變異位點 (Incidental finding)。
檢測結果呈現美國醫學遺傳學暨基因體學學會 (ACMG) 指引或 ClinVar 資料庫判定為致病性/可能致病性 (Pathogenic/Likely Pathogenic) 的變異位點。AAF (Alternative Allele Frequency) 紀錄該變異位點在東亞族群的等位基因頻率。合子型式包含 Homo (Homozygote 同型合子)、Het (Heterozygote 異型合子)、Hemi (Hemizygote 半合子)。詳細說明請參考檢測結果注釋。

意外發現變異位點注釋

本次檢測無意外發現變異位點注釋。

檢測限制

本定序方式僅檢測單點核苷酸變異 (Single Nucleotide Variants, SNVs)、小片段缺失或插入的突變 (Small Insertions/Deletions, Small InDels)，無法提供基因序列具有大片段插入或缺失 (Large InDels)、拷貝數變異 (Copy Number Variants)、轉位 (Translocations)、倒位 (Inversions) 與 DNA 結構變異等相關變異型資訊。

本檢驗僅針對生殖系變異位點 (Germline Variants) 進行偵測，並無提供體細胞變異位點 (Somatic Variants) 資訊。本報告僅呈現致病性、可能致病性變異位點，未確定臨床意義變異 (Variant of Uncertain Significance, VUS)、可能良性 (Likely Benign) 與良性 (Benign) 變異位點不會被報導。本報告不提供不具致病性之同義突變 (Synonymous Mutations)。本報告僅呈現檢測基因的外顯子 (Exon) 區域與影響外顯子剪切的變異位點。

本檢測僅針對檢測標的進行偵測，不包含在檢測範圍的其他變異也可能會導致疾病的發生。相同的檢測結果在不同個體間可能會有表現型的差異，本檢測結果並無考慮受檢者間的個體差異，且不預測罹患疾病的嚴重程度或發病年齡。

雖然 DNA 基因檢測技術準確率很高，但少數錯誤仍可能發生，因此重大醫療決策應配合臨床症狀或其他相關檢測結果進行評估。本項檢測經確效驗證，整體準確度 99.99%。

本檢測偵測到的變異位點會透過 ACMG 指引進行致病力判定，當變異位點判讀所需要的佐證資料不足時，可能會被判為未確定臨床意義變異 (VUS)，導致該位點不會呈現在報告中。隨著醫學知識迅速發展，報告內容僅提供檢測當下最新的資訊，未來可能會出現新的證據並影響變異位點的致病力判讀結果，因此可能有偽陽性或偽陰性的風險。

本定序方式會先經過捕獲 (Capture) 和擴增 (Enrichment) 的步驟，由於定序區域的捕獲效率不一，少部分定序深度不足 20 X 的位置將無法進行後續分析。本次檢測的可偵測區間可洽詢實驗室提供。

附註

本檢測僅就委託者之委託事項提供檢測結果，檢測實驗室不進行臨床意義之判斷。本基因檢測結果僅提供給受檢者做參考或協助醫師進行疾病診斷，當醫師為病患進行診斷與治療時，不應只基於本報告之資訊，須將受檢者過往病歷資料共同納入考量。

疾病可能為多重因素綜合導致的結果，除了基因之外其他如生活、飲食、習慣等因素也會影響罹患疾病的風險。若檢測出致病性變異或可能致病性變異僅表示受檢者有較高風險罹患特定種類的疾病，但並不表示受檢者將來必然會罹患該項疾病，有些遺傳到致病性變異位點的人終其一生都未罹患相關的疾病。相反地，若未檢測出任何致病性變異位點時，表示在本檢測所涵蓋的基因中未找到可能影響疾病發生之變異位點，然而影響疾病生成的遺傳性原因有可能未在本檢測被驗出，因為目前並沒有任一檢測項目可以涵蓋所有導致疾病的遺傳變化，因此陰性結果不能完全排除受檢者帶有影響疾病發生之變異，也並不表示將來沒有罹患該項疾病的可能。

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附件

報告編號:
採檢日期:
報告日期:
委託單位:

附件資訊

本次檢測無相關附件資訊。