



檢體資訊

報告編號: JB23_185 檢測方法: NGS 採集日期: 2023.04.26 聯 絡 人: 宋岳峰

採檢單號: 100361 委託醫師: 宋岳峰 收檢日期: 2023.04.27 聯絡人電話: 0920859835

檢 體 類 別: 血液檢體 委 託 單 位: 三總神經內科 報 告 日 期: 2023.05.17 聯 絡 人 信 箱: sungyf@hotmail.com.tw

檢測報告總結

1. 意外發現一個與隱性疾病血色素沉著症 (Hemochromatosis) 相關基因 HFE 的致病性變異位點。受檢者在此位點為異型合子,因此僅為帶因者。

樣本品質備註

樣本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。

報告簽署

報告簽署人: **美** 2023-05-2

報告簽署人註解:

覆核醫師:

2023-05-24



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檢測結果

編號 染色體

基因 / 外顯子

變異位點

AAF / TAAF

遺傳合子

致病力

本檢測未偵測到神經系統疾病相關基因的致病性或可能致病性變異位點

借註

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) 與 TAAF (Taiwan alternative allele frequency) 分別紀錄該變異位點在東亞族群與台灣族群的等位基因頻率。遺傳合子包含 Homo (Homozygous 同型合子)、Het (Heterozygous 異型合子)、Hemi (Hemizygous 半合子)。詳細說明請參考檢測結果注釋。

檢測位點注釋

本次檢測無神經系統疾病相關變異位點注釋



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產品介紹

本『神經系統疾病基因檢測』是為個人化預防保健所開發的基因檢測服務,檢測對象為神經系統疾病患者或疑似有的人群。本檢測提供 20 種神經系統疾病檢測,涵蓋認知障礙、癲癇等各種常見神經系統疾病,透過 1915 個相關基因的變異位點檢測,可以了解受檢者的神經系統疾病遺傳資訊。藉由生活習慣及飲食的改善,可以減低各類神經系統疾病發生的機率,以達到「預防醫學」之目的並進行全方位的「健康管理」規劃。

檢驗項目

威爾森氏症	Wilson's disease
癲癇	Epilepsy
腦小血管疾病	Brain 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 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,



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基因列表

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, CYP2TA1, 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, 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, GRIN2A, GRIN2B, GRM1, GRM7, GRN, GSN, GSS, GSX2, GTF2E2, GTF2H5, GTPBP2, GTPBP3, GUF1, GUSB, HACE1, HADH, HADHA, HADHB, HARS1, HCCS, HCN1, HCN4, HDAC4, HDAC8, HDC, HEPACAM, HERC1, HESX1, HEXA, HEXB, HFE, HGSNAT, HIBCH, HIC1, HIKESHI, HINT1, HIVEP2, HK1, HLA-DQA1, HLCS, HMBS, HMGCL, HNMT, HNRNPA1, HNRNPA2B1, HNRNPH2, HNRNPU, HOXD10, HPCA, HPDL, HPRT1, HS6ST1, HSD17B10, HSD17B4, HSD3B7, HSPB1, HSPB3, HSPB8, HSPD1, HTRA2, HTT, HYAL1, HYLS1, IARS2, IBA57, IDS, IER3IP1, IFIH1, IFNG, IFNGR1, IFRD1, IFT140, IGHMBP2, IL10, IL12A, IL17RD, IL23R, IL31RA, IMMP2L, IMPDH2, INF2, INPP5E, INPP5K, INS, INS-IGF2, INSR, INTS8, INVS, IQCB1, IQGAP3, IQSEC1, IQSEC2, IRAK1, IREB2, IRF4, IRF6, ISCA1, ISCA2, ITGA7, ITM2B, ITPA, ITPR1, JAG1, JAK3, JAM2, JMJD1C, JPH1, JPH3, JRK, KARS1, KAT6A, KAT6B, KATNAL2, KATNIP, KCNA1, KCNA2, KCNA4, KCNB1, KCNC1, KCND3, KCNH5, KCNIP1, KCNJ10, KCNJ11, KCNK4, KCNK9, KCNMA1, KCNN2, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD17, KCTD7, KDM3B, KDM4B, KDM5B, KDM5C, KIAA0586, KIAA0753, KIDINS220, KIF11, KIF1A, KIF1B, KIF1C, KIF24, KIF26B, KIF5A, KIF7, KIFBP, KIRREL2, KIRREL3, KIT, KLC2, KLHL9, KLLN, KLRC4, KMT2B, KMT2E, KNSTRN, KPNA3, KRAS, KRIT1, KRT1, KRT83, KY, L1CAM, LAMA1, LAMA2, LAMA4, LAMB1, LAMP2, LARGE1, LARP7, LARS2, LDB3, LDLR, LDLRAP1, LETM1, LGI1, LGI4, LIAS, LIG3, LIG4, LINS1, LIPC, LIPN, LIPT1, LITAF, LMAN2L, LMF1, LMNA, LMNB1, LMNB2, LNP1, LNPK, LONP1, LPL, LRCH2, LRP12, LRPPRC, LRRK2, LRSAM1, LSM7, LTBP4, LYRM7, LYST, LYZ, LZTFL1, MAB21L1, MAG, MAGEL2, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K2, MAPK10, MAPK8IP3, MARCHF4, MARCHF6, MARS1, MARS2, MASP2, MAST1, MAST4, MAT1A, MATR3, MBD5, MBOAT7, MBTPS2, MCM3AP, MCOLN1, MDH1, MDH2, MECP2, MECR, MED12L, MED13L, MED23, MED25, MED27, MEF2C, MEFV, MEN1, MEOX1, MFF, MFN2, MFSD8, MICAL1, MICOS13, MICU1, MINPP1, MKS1, MLC1, MMACHC, MMADHC, MME, MMP1, MMP10, MMUT, MOBP, MOGS, MOK, MORC2, MPC1, MPDU1, MPLKIP, MPO, MPV17, MPZ, MRE11, MRI1, MRM2, MRNIP, MRPL12, MRPS22, MRPS25, MRPS34, MS4A14, MSH6, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, 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, MT-TW, 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, NMNAT2, NODAL, NOL3, NONO, NOP10, NOP56, NOS3, NOTCH2, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPPA, NPR2, NPRL2, NPRL3, NR4A2, NR5A1, NRG1, NRXN1, NRXN2, NSD1, NSD2, NSUN2, NT5C2, NTNG1, NTRK1, NTRK2, NTSR1, NUBPL, NUDT2, NUP107, NUP133, NUP214, NUP62, NUP85, OCA2, OCRL, ODC1, OFD1, OGDH, OGDHL, OPA1, OPA3, OPHN1, OPLAH, OPTN, OR5K3, OR5K4, OR6C1, OSGEP, OSMR, OSTM1, OTC, OKA, OTUBEB, OXA, P4HA2, P4HTM, PACS2, PAFAH1B1, PAK1, PANK2, PARK7, PARN, PARS2, PAX3, PAX6, PC, PCCA, PCCB, PCDH11, PCDH12, PCDH12, PCDH12, PCDH12, PCDH12, PCSK9, PCYT2, PDCD10, PDE2A, PDE4D, PDE6D, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PDXK, PDYN, PDZD7, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PGAP1, PGAP2, PGAP3, PGK1, PGM3, PHACTR1, PHAX,



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基因列表

PHF6, PHGDH, PHKA1, PHOX2B, PHYH, PI4KA, PIBF1, PIEZO2, PIGA, PIGB, PIGC, PIGG, PIGK, PIGL, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGW, PIGY, PIK3CA, PIK3CD, PIK3R5, PINK1, PITRM1, 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, PNPLAT, 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, 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, TP11, 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



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檢測流程

gDNA 進行文庫製備後使用探針系統捕獲完整的待定序區域,確保其符合品質管控需求後將其注入流道槽中。透過NGS 平台進行雙端定序,平均定序深度為 100 倍以上。定序片段與人類參考基因組 (hg19) 進行序列比對後,篩選相關基因變異位點,並排除定序深度小於 20 倍或等位基因頻率小於 30% 的變異位點。最後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) 判定後將報導位點列於報告中。

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 2023-04-28
HPO 2023-03-20
rmsk 2020-02-20
HGVS 20.05
Human reference genome hg19

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):

本檢測報導位點僅包含致病性 (Pathogenic) 與可能致病性 (Likely pathogenic) 等變異位點·若檢測出前述變異位點表示受檢者帶有可能影響疾病發生的變異·但並不代表受檢者將來必然會罹患該病症·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·也並不表示將來沒有罹患疾病的可能。

- (1) 致病性變異 (Pathogenic): 致病性變異位點可能直接影響疾病發生,這類的變異位點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): 可能致病性變異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 (Uncertain significance): 此變異位點在疾病上的影響目前未確定。
- (4) 藥物反應變異 (Drug response): 此變異位點可能影響用藥效果。
- (5) 可能良性變異 (Likely benign): 可能良性變異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): 良性變異位點不影響疾病發生,這類的變異位點具有充足的證據顯示與疾病的發生無關。

遺傳合子 (Genetic Zygosity):

- (1) 異型合子 (Heterozygous): 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 (Homozygous): 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 (Hemizygous): 變異位點出現在僅有一條不成對的染色體基因上。



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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 / TAAF	遺傳合子	致病力
2	chr6	HFE /2	c.187C>G p.(His63Asp)	2.70% /2.93%	Het	ClinVar: Conflicting interpretation of pathogenicity*

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) 與 TAAF (Taiwan alternative allele frequency) 分別紀錄該變異位點在東亞族群與台灣族群的等位基因頻率。遺傳合子包含 Homo (Homozygous 同型合子)、Het (Heterozygous 異型合子)、Hemi (Hemizygous 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

本檢測意外發現一個與隱性疾病血色素沉著症 (Hemochromatosis) 相關的可能致病性變異位點 *HFE* c.187C>G·受檢者在此位點為異型合子 (Heterozygous)·因此僅為帶因者 (Carrier)。此外·*HFE* c.187C>G 位點過去被報導為低外顯率 (Penetrance) 的變異·帶有此變異同型合子 (Homozygous) 或是另一股染色體帶有其他 *HFE* 基因變異 (Compound heterozygous) 的人·只有小於 2% 的比例會有血色素沉著症相關症狀 ^{1,2}。

HFE c.187C>G p.(His63Asp) 在全球的等位基因頻率為 10.82%·在東亞的等位基因頻率為 2.70%·在台灣族群等位基因頻率為 2.93%³。此檢測位點位於 HFE 基因的第 2 外顯子·並在核苷酸編碼區第 187 位置發生 C 轉 G 的錯義突變·造成第 63 個胺基酸從組胺酸 (Histidine; His) 轉變為天門冬胺酸 (Aspartic acid; Asp)。過去研究發現血色素沉著症患者帶有此檢測位點 4.5。ClinVar 資料庫報導此位點為血色素沉著症的致病性變異 6。綜上所述,判斷此位點為血色素沉著症的致病性變異 7。

- 1 https://pubmed.ncbi.nlm.nih.gov/11874997/
- 2 https://www.ncbi.nlm.nih.gov/books/NBK1440/
- 3 https://gnomad.broadinstitute.org/variant/chr6-26091179-C-G?dataset=gnomad_r2_1
- 4 https://pubmed.ncbi.nlm.nih.gov/8696333/
- 5 https://pubmed.ncbi.nlm.nih.gov/24729993/
- 6 https://www.ncbi.nlm.nih.gov/clinvar/variation/10/
- 7 https://pubmed.ncbi.nlm.nih.gov/25741868/

^{*}Conflicting interpretations of pathogenicity: Pathogenic(24); Pathogenic, low penetrance(1); Uncertain significance(2); Benign(2)。括號內數字代表 ClinVar 資料庫提交者判定該致病性的數量。



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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 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 (Translocation)、倒位 (Inversion) 與 DNA 結構變異等相關變異型資訊。

本報告僅呈現致病性、可能致病性等變異位點,未確定臨床意義變異 (Variant of uncertain significance)、可能良性 (Likely benign) 與良性 (Benign) 變異位點與不會被報導。本報告不提供不具致病性之同義突變 (Synonymous mutation)。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。

陰性檢測結果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。

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附註

檢測結果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。本基因檢測提供的資訊僅協助醫 師進行疾病診斷,醫師為病患進行診斷與治療時,不應只基於本報告之資訊,須將病患過往病歷資料共同納入考量。

本基因檢測旨在提供醫師額外的遺傳訊息,隨著醫學知識迅速發展,報告內容僅提供檢測當下最新的遺傳資訊,從報告發放到閱讀期間,可能會出現新的證據。此外,相同的變異位點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。