

次世代定序全外顯子定序檢測-單基因遺傳疾病-II

基因檢驗套組-II (限基因醫學部醫師開立)

112 年 3 月修訂

皮膚科- Ectodermal dysplasia (ED)
AKT1, BCS1L, BRAF, CDH3, COL11A1, CYP26C1, DICER1, DLX5, DOCK8, DSP, EDA, EDAR, EDARADD, ERCC2, EVC, EVC2, FGF10, FGFR2, FGFR3, FZD6, GJB2, GJB6, GRHL2, HOXC13, HR, IFT122, IFT43, IKBKG, JUP, KDF1, KLLN, KRAS, KRT14, KRT16, KRT17, KRT74, KRT85, LRP6, MAP2K1, MAP2K2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKBIA, NLRP1, ORAI1, PADI3, PIK3CA, PKP1, PORCN, PRKD1, PTEN, RMRP, RSPO4, SDHB, SDHD, SMARCB1, TP53, TP63, TWIST2, WDR19, WDR35, WNT10A, WNT10B
皮膚科- Pigmentary genodermatoses (Vitiligo, Incontinentia Pigmenti, Dowling Degos Disease, Cutis marmorata telangiectatica congenita, dyschromatosis symmetrica hereditaria, Aicardi-Goutières)
ADAR, ARL6IP6, BACH2, BTNL2, C1QTNF6, CASP7, CCR6, CD44, CD80, CDH23, CLNK, CXCR5, DCT, DDX6, GNA11, GZMB, HCG9, HERC2, IFIH1, IKBKG, IKZF4, IL2RA, KRT5, LPP, MC1R, MLANA, OCA2, PMEL, POFUT1, POGLUT1, PSENEN, PTPN22, RERE, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIPI1, SAMHD1, SH2B3, SLA, SLC29A3, SMOC2, SOX10, TICAM1, TOB2, TREX1, TSBP1, TYR, TYRP1, UBASH3A, ZMIZ1
皮膚科- Epidermal genodermatoses (Darier, Hailey-Hailey, Porokeratosis, Acral Peeling Skin, Erythrokeratoderma variabilis)
ATP2A2, ATP2C1, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FDPS, FERMT1, FLG2, GJB3, GJB4, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MVD, MVK, NFKB2, PKP1, PLEC, PMVK, SART3, SERPINB8, SLC17A9, SSH1, TERC, TGM5
眼科- Corneal dystrophy
AGBL1, AGK, CHRDL1, CHST6, COL8A2, CRYAA, CRYBA4, CRYBB1, CRYBB2, CRYGC, CRYGD, CYP4V2, DCN, EXOSC2, GJA8, GLA, GNAQ, GSN, KRT12, MAF, MBTPS2, OPN1LW, OPN1MW, OSMR, PIKFYVE, PRDM5, SLC25A4, SLC4A11, TACSTD2, TGFB1, TKFC, UBIAD1, ZEB1, ZNF469
耳鼻喉科- Otosclerosis
BMP2, BMP4, RELN, TGFB1
婦兒科-遺傳性心臟疾病
A2ML1, AARS2, ABCB4, ABCC9, ACAD8, ACAD9, ACADS, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, ACVRL1, AGK, AGL, AGPAT2, AGTR1, AHCY, AIP, AKAP9, ALG10, ALMS1, ALPK3, AMN, ANK2, ANKRD1, ANO5, APC2, APOA1, APOB, ARSB, ASPH, ATP1B1, ATP2A2, ATPAF2, AVIL, BAG3, BIN1, BMPR1A, BMPR1B, BMPR2, BOLA3, BRAF, BSCL2, C1QBP, CACNA1C, CACNA1D, CACNA1S, CACNA2D1, CACNA2D4, CACNB2, CALM1, CALM2, CALM3, CALR, CALR3, CAPN3, CASQ2, CAV1, CAV3, CAVIN4, CBL, CCDC78, CDKN1C, CHD7, CHKB, CHRM2, CIDEC, CNGA1, CNGA2, CNGA3, CNGA4, CNOT1, COA5, COA7, COA8, COL4A1, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CORIN, COX10, COX14, COX15, COX20, COX6B1, CPT1A, CPT2, CRELD1, CRPPA, CRYAB, CSRP3, CTC1, CTF1, CTNNA3, CUBN, CXADR, D2HGDH, DAG1, DBH, DCAF8, DCP1B, DES, DMD, DNAJC19, DOLK, DPH2, DPP10, DPP6, DSC2, DSC3, DSG2, DSP, DTNA, DYSF, EDN1, EDNRA, EIF2AK4, ELAC2, ELN, EMD, ENG, EPG5, ETFA, ETFB, ETFDH, EYA4, FASTKD2, FBNI, FBXL4, FBXO32, FGA, FGF12, FHL1, FHL2, FIG4, FKRP, FKTN, FLNA, FLNC, FOXC1, FOXF1, FOXH1, FOXRED1, FXN, G6PC3, GAA, GATA4, GATA5, GATA6, GATAD1, GBE1, GDF2, GFM1, GINS3, GJA1, GJA5, GLA, GLB1, GMPPB, GNAS, GNB5, GNE, GPC3, GPC4, GPD1L, GSN, GTPBP3, GUSB, GYG1, GYS1, GYS2, H19, HACD1, HADHA, HADHB, HAMP, HCCS, HCN4, HEY2, HFE, HJV, HOOK3, HPS1, HRAS, HSD17B10, IBA57, IDH2, IDUA, IGF2, IGHMBP2, IL31RA, ILK, ITGA6, ITGB4, JAG1, JPH2, JUP, KANK2, KCNA5, KCNAB2, KCNB2, KCND3, KCNE1, KCNE2, KCNE3, KCNE4, KCNE5, KCNH2, KCNJ16, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KCNT1, KRAS, KRT17, L2HGDH, LAMA2, LAMA4, LAMP2, LARGE1, LDB3, LDLR, LDLRAP1, LIG3, LIMS2, LIPE, LITAF, LMNA, LMNB2, LPAR6, LYZ, LZTR1, MAP2K1, MAP2K2, MEFV, MIB1, MLYCD, MMUT, MRPL3, MRPL44, MTO1, MTSS1, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOT, MYOZ1, MYOZ2, MYPN, NDRG4, NDUFA1, NDUFA11, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFV1, NDUFV2, NEBL, NEU1, NEXN, NF1, NF2, NFIX, NKX2-5, NODAL, NOS1AP, NOS2, NOTCH1, NOTCH2, NOTCH3, NPPA, NRAS, NSD1, NSUN2, NUBPL, OSMR, PCCA, PCCB, PCSK9, PDLIM3, PDSS1, PDSS2, PERP, PET100, PEX19, PEX7, PHYH, PIK3CA, PKP2, PKP4, PLEC, PLEKHM2, PLN, PMM2, PNN, PNPLA2, POLG, POMGNT1, POMT1, POMT2, PPARG, PPP1R3A, PRDM16, PRKAG2, PRKARIA, PSEN1, PSEN2, PTPN11, PXDNL, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBCK1, RBM20, RFFL, RIT1, RMND1, RNF207, RRAS, RYR2, SAA1, SCN10A, SCN1B, SCN2B, SCN3B, SCN4A, SCN4B, SCN5A, SCN9A, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA, SELENON, SEMA3A, SERPINE1, SETD6, SGCA, SGCB, SGCD, SGCG, SGO1, SHOC2, SIRT3, SLC19A2, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC8A1, SLMAP, SMAD1, SMAD3, SMAD4, SMAD5, SMAD9, SMCHD1, SNTA1, SOS1, SOX17, SPEG, SPRED1, SRI, SUCLG1, SYNE2, TACO1, TAZ, TBX1, TBX20, TBX4, TBX5, TCAP, TECRL, TFAP2B, TGFB3, TGFBRI, TGFBRII, THBS1, TIMMDC1, TLL1, TMEM126B, TMEM43, TMEM70, TMPO, TNNC1, TNNT3, TNNT3K, TNNT2, TOPBP1, TPM1, TRDN, TRIM32, TRIM37, TRIM63, TRPC6, TRPM4, TRPM7, TSFM, TTN, TTR, TWNK, TXNRD2, UQCC2, UQCRB, UQCRC2, UQCRCQ, VCL, VCP, WFS1, XK, YARS2, ZFPM2, ZIC3

NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK9, NEUROD2, NGLY1, NHLRC2, NKX2-1, NKX6-2, NODAL, NOS3, NOTCH3, NPC1, NPC2, NR4A2, NSUN2, NUP62, OFD1, PANK2, PARK7, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCGF2, PDE10A, PDGFB, PDGFRB, PDHA1, PDHX, PET100, PGAP1, PIGC, PIGP, PIGQ, PIK3R5, PINK1, PLA2G6, PLAA, PLAU, PLEKHG2, PLP1, PMPCB, PNKD, PNKP, PNPLA8, PNPT1, PODXL, POLG, POLRIC, POLRID, POLR3A, POLR3B, PPP2R2B, PPP3CA, PRKAR1B, PRKCG, PRKN, PRKRA, PRNP, PRRT2, PRSS12, PSAP, PSEN1, PSEN2, PTCH1, PTPRH, PTS, PURA, QDPR, RAB11B, RAB39B, RARS1, RHOBTB2, RIC3, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RSRC1, SALL4, SAMHD1, SARS1, SCN1A, SCN1B, SCN2A, SCN4A, SCN8A, SCN9A, SCO2, SCP2, SDHA, SEPSECS, SERAC1, SET, SETX, SGCE, SHH, SIGMARI, SIK1, SIX3, SLC18A2, SLC19A3, SLC20A2, SLC25A42, SLC2A1, SLC30A10, SLC30A9, SLC39A14, SLC39A8, SLC44A1, SLC45A1, SLC6A3, SLC6A8, SLC9A6, SMC1A, SMPD1, SNCA, SNCAIP, SNCB, SNORD118, SPATA5, SPG11, SPOP, SPR, SQSTM1, ST3GAL3, STAG2, STARD7, STN1, STX16, STXBP1, SUCLA2, SUOX, SURF1, SYNE1, SYNGAP1, SYNJ1, SYT1, TACO1, TAF1, TANGO2, TARDBP, TBC1D24, TBCD, TBK1, TBP, TCF4, TCOF1, TECR, TGFB2, TGIF1, TGM6, TH, THAP1, TIMM8A, TK2, TMEM106B, TMEM230, TMEM240, TNIK, TORIA, TPII, TPKE1, TRAPPC11, TRAPPC12, TRAPPC9, TREM2, TREX1, TRITI, TRPM7, TRPV4, TSEN15, TSEN2, TSEN34, TSEN54, TSMF, TTBK2, TTC19, TTPA, TUBB3, TUBB4A, TUSC3, TWIST1, TWNK, TXN2, UBA5, UBQLN2, UBTF, UCHL1, UFM1, UGDH, UHRF1BP1L, UPB1, UQCRC1, UQCRCQ, VAC14, VAMP1, VAMP2, VCP, VPS13A, VPS13C, VPS13D, VPS35, VPS37A, WARS2, WASHC4, WDR45, WDR73, XPR1, YIF1B, YY1, ZC3H14, ZC4H2, ZIC2, ZMYND11, ZNF142

腫瘤醫學-心臟毒性

ABCC9, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, AKAP9, ANK2, ANKRD1, ARHGAP31, ATM, B3GAT3, BAG3, BCOR, BMPR2, BRAF, CACNA1C, CACNB2, CALM1, CALM2, CASQ2, CAV3, CAVIN4, CBL, CDH2, CFAP53, CFC1, CHD7, CITED2, CLDN16, CLDN19, CNNM2, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, CREBBP, CRELD1, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EFEMP2, EGF, EHMT1, ELN, EMD, ENG, EP300, EVC, EVC2, EYA4, FBN1, FBN2, FHLL1, FKTN, FLNA, FLNC, FOXC1, FOXF1, FOXH1, FXYD2, GAA, GATA4, GATA5, GATA6, GDF1, GDF2, GJA1, GJA5, GLA, GPC3, GPD1L, HCCS, HCN4, HFE, HRAS, HTRA1, ILK, JAG1, JPH2, JUP, KCNA1, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNK3, KCNQ1, KDM6A, KMT2D, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K2, MED12, MED13L, MEIS2, MFAP5, MIB1, MMP21, MMP3, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOZ2, MYPN, NEBL, NEXN, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NOTCH3, NPPA, NR2F2, NRAS, NSD1, PDLIM3, PKD1L1, PKD2, PKP2, PLN, PRDM16, PRKAG2, PRKG1, PSEN1, PSEN2, PTPN11, RAF1, RASA1, RBM10, RBM20, RIT1, RYR2, SALL1, SALL4, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, SKI, SLC12A3, SLC22A5, SLC25A4, SLC2A10, SLMAP, SMAD3, SMAD4, SMAD6, SMC3, SNTA1, SOS1, SOS2, SOX2, STRA6, SYNE1, SYNE2, TAB2, TAZ, TBX1, TBX20, TBX5, TCAP, TFAP2B, TGFB2, TGFB3, TGFBRI, TGFBRII, TLL1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TREX1, TRIM63, TRPM4, TRPM6, TTN, TTR, VCL, ZEB2, ZFPM2, ZIC3