

次世代定序全外顯子定序檢測-單基因遺傳疾病-I

基因檢驗套組-I

112年3月修訂

<p>皮膚科- Epidermolysis bullosa (EB)</p> <p><i>CARM1, CD151, CDSN, CHST8, COL17A1, COL4A1, COL7A1, CSTA, CTSB, CTSL, CTSV, DSG1, DSG1-AS1, DSG2, DSG2-AS1, DSG3, DSG4, DSP, DST, EXPH5, FBLN1, FBLN2, FERMT1, GIPC1, GRIK2, GRIP1, GRIPAP1, HDAC11, ITGA2, ITGA3, ITGA6, ITGB1, ITGB2, ITGB4, ITGB7, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMA5, LAMB1, LAMB2, LAMB3, LAMC1, LAMC2, MIR1268A, MMP1, NID1, PKP1, PKP4, PKP4-AS1, PLEC, PLOD3, RAB27A, SNORA93, SPTAN1, SYN1, TGM5, THRA, TRIB1, TSPAN4, VIM, WTAPP1</i></p>
<p>皮膚科- Palmoplantar keratoderma, pachyonychia congenita (PPK and PC)</p> <p><i>AAGAB, AQP5, CFTR, COL14A1, COL20A1, CTSC, DSC2, DSG1, DSP, ENPPI, FAM83G, GJB2, GJB3, GJB4, GJB6, JUP, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, NLRP1, RHBDF2, RSP01, SERPINB7, SLURP1, TRPV3</i></p>
<p>皮膚科- Ichthyosis</p> <p><i>ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ARSA, ARSB, ARSL, BRAF, CASP14, CDSN, CERS3, CHST8, CLDN1, CSTA, CYP4F22, EBP, ELOVL4, ERCC2, ERCC3, FLG, FLG2, GBA, GJB2, GJB3, GJB4, GJB6, GTF2H5, KDSR, KRAS, KRT1, KRT10, KRT2, KRT9, LIPN, MAP2K1, MAP2K2, MBTPS2, MPLKIP, NIPAL4, NSDHL, PEX7, PHGDH, PHYH, PNPLA1, PNPLA2, POMP, PSAT1, SDR9C7, SERPINB8, SLC27A4, SNAP29, SPINK5, ST14, STIM1, STS, SUMF1, TGM1, TGM5, VPS33B, ZMPSTE24</i></p>
<p>皮膚科- Inflammatory genodermatoses (Psoriasis, Autoinflammatory Keratinization Diseases, Hidradenitis Suppurativa, Netherton, PAPA, Majeed, Muckle-Wells, Omenn, Cryopyrin-associated periodic)</p> <p><i>ABCA12, ABHD5, ADAM17, ADAMTSL5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AP1S3, APH1A, APH1B, AQP5, ARSL, B3GNT2, C1orf68, CARD14, CARM1, CASP14, CD1A, CDSN, CERS3, CHST8, CLDN1, CSTA, CTSC, CYP4F22, DCLRE1C, DDX58, DSP, EBP, EGFR, ELANE, ELMO1, ELOVL4, ERAP1, ETS1, EXOC2, FBXL19, FGFR2, FLG, FLG2, FUT2, GJB2, GJB3, GJB4, GJB6, HSD3B7, IFIH1, IFNL1, IL12B, IL12RB1, IL13, IL23A, IL23R, IL36RN, IL4, KDSR, KLF4, KLK5, KLK7, KRT1, KRT10, KRT16, KRT17, KRT2, KRT9, LCE3B, LCE3C, LIPN, LPIN2, LTA, MBD2, MBTPS2, MEFV, MICA, MSMO1, MVK, MYD88, NCSTN, NFKBIA, NIPAL4, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOS2, NSDHL, OCRL, OTULIN, PDE4A, PEX7, PHGDH, PHYH, PLA2G4D, PLCG2, PNPLA1, POMP, PSAT1, PSEN1, PSEN2, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RAG1, RAG2, REL, REV3L, RNF114, RUNX3, SDR9C7, SERPINB7, SERPINB8, SLC27A4, SNAI1, SNAP29, SOCS1, SPINK5, ST14, STAT2, STAT3, STAT5A, STAT5B, STS, TAGAP, TEK, TGM1, TGM5, TNF, TNFAIP3, TNFRSF1A, TNFRSF9, TNIP1, TRAF3IP2, TYK2, UBE2L3, VPS33B, ZMPSTE24, ZNF750</i></p>
<p>皮膚科- Skin tumoral genodermatoses (Nevoid BCC, Osler-Weber- Rendu, Epidermal nevus, Peutz-Jeghers, neurofibromatosis, Legius, Brooke-Spiegler, Schöpf-Schulz-Passarge, Goltz-Gorlin, Birt-Hogg-Dubé, Gardner, Myotonic muscular dystrophy, Cowden, Keratitis, ichthyosis and deafness, Muir-Torre, Rubinstein-Taybi)</p> <p><i>ACVRL1, CNBP, CREBBP, CTNNA1, CYLD, DMPK, ENG, EP300, FGFR3, FLCN, GDF2, GJB2, HRAS, KRAS, MLH1, MSH2, MSH6, NF1, NF2, PIK3CA, PMS2, PORCN, PTCH1, PTCH2, PTEN, RASA1, SHH, SMAD4, SMARCB1, SPRED1, STK11, SUFU, TP53, WNT10A</i></p>
<p>皮膚科- Miscellaneous genodermatoses (Ehlers-Danlos, Pseudoxanthoma elasticum, Cutis laxa, early-onset sarcoidosis, Familial chilblain lupus, STING-associated vasculopathy with onset in infancy, hyper-IgE, Rothmund Thomson, Generalized Verrucosis, Keratosis Follicularis Spinulosa Decalvans)</p> <p><i>ABCC6, ACLY, ACO1, ACO2, ACTA2, ADA, ADAMTS2, ALDH18A1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ASXL1, ATM, ATP6V0A2, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BACH2, BGN, BHLHA9, BMP2, BMPRI1, BTRC, C1QTNF6, CARMIL2, CASP7, CBS, CC2D2A, CCR6, CD19, CD40, CD40LG, CD44, CD80, CDH3, CEP290, CHST14, CHSY1, CIB1, CLNK, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CORO1A, CS, CTLA4, CXCR4, DCLRE1C, DLAT, DLD, DLL4, DLST, DLX5, DOCK6, DOCK8, DPCD, DSE, DVL1, DVL3, DYNC1H1, ECM1, EFEMP2, ELN, EOGT, ESCO2, FBLN5, FBN1, FBN2, FBXW4, FGF10, FGF16, FGFR1, FGFR2, FGFR3, FH, FKBP14, FLNA, FOXD3, FOXP1, FOXP3, GATA2, GDF5, GLI3, GNAS, GZMB, HDAC4, HDAC8, HOXD13, ICOS, IDH1, IDH2, IDH3A, IDH3B, IDH3G, IFIH1, IHH, IKBKG, IKZF4, IL2, IL2RA, IL7, ITGB2, JAK3, KIF7, KMT2A, LBX1, LCK, LMBR1, LOX, LPP, LRP4, LTBP4, MAT2A, MBTPS2, MC1R, MDH1, MDH2, MED12, MFAP5, MGP, MKS1, MSH5, MST1, MYCN, MYH11, MYLK, NIPBL, NLRP1, NOD2, NOG, NOTCH1, NSDHL, OCA2, OGDH, OGDHL, PC, PCK1, PCK2, PDHA1, PDHA2, PDHB, PHF6, PIGV, PLOD1, POLL, PRDM5, PRKG1, PTHLH, PTPN22, PYCR1, RAD21, RASGRP1, RBM8A, RBPJ, RECQL4, RERE, RHOH, RIN2, ROR2, RPGRIP1L, SALL1, SALL4, SAMHD1, SDHA, SDHB, SDHC, SDHD, SH2B3, SHH, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC2, SOX11, SOX9, SPINK5, STAT3, STING1, STK4, SUCLA2, SUCLG1, SUCLG2, TAOK2, TBX15, TBX3, TBX5, TGFB2, TGFB3, TGFBRI1, TGFBRI2, THPO, TICAM1, TMC6, TMC8, TNFRSF13B, TNFRSF13C, TNXB, TOB2, TP63, TPP2, TREX1, TSLP, TYK2, TYR, UBASH3A, WAS, WNT10B, WNT3, WNT5A, WNT7A, XBP1, ZNF469</i></p>

眼科- Lens_disease

ABCA3, ABCD1, ABCD3, ABHD12, ACE, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADIPOQ, AFF1, AGK, AGPS, AIPL1, AIRE, AKR1B1, AKR1E2, ALDH18A1, ALDH1A1, ALG12, ANK2, AP4B1, APOA1, APOE, APP, ARID1B, ARL2, ARSL, ARTN, ASPH, ATAD3A, ATM, ATOH7, B3GALT6, B3GLCT, B3GNT4, B4GALT7, BAP1, BCAR3, BCOR, BEST1, BFSP1, BFSP2, BIN3, BMP4, BRD4, BUB1B, C12orf57, CACNA1C, CASR, CAT, CAV1, CBS, CC2D2A, CCNP, CDK5RAP2, CDKN2A, CHMP4B, CHRDL1, CLOCK, CLPB, CNBP, CNGB3, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, COL4A5, COL4A6, COL5A1, COL7A1, CPAMD8, CPOX, CRIM1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CRYZ, CTDP1, CTNND2, CYP1B1, CYP27A1, CYP46A1, CYP51A1, DCXR, DHCR7, DICER1, DMPK, DNASE2B, DNMBP, DOCK5, DPAGT1, DST, DYNCH1, EBP, EFNA5, EPG5, EPHA2, ERCC2, ERCC6, ERCC8, ESCO2, ESR1, ETFDH, EXOC6B, EYA1, EZR, FAM126A, FBN1, FBN2, FDFT1, FIGN, FKR, FKTN, FLNB, FOXC1, FOXE3, FTL, FTO, FYCO1, FZD4, GALE, GALK1, GALT, GBA2, GCM2, GCNT2, GEMIN4, GFER, GJAI, GJA3, GJA8, GJB6, GJC3, GJE1, GLA, GLB1, GNAI1, GNAQ, GNAS, GNPAT, GPR161, GPX1, GSTM1, GSTP1, GSTT1, GTF2IRD1, GUCY2D, HCCS, HIP1, HMX1, HRAS, HSF4, HSPG2, IARS2, IDO1, IFNGR1, IGF1R, IKBKG, INPP5K, INS, INTS1, IPO13, ITGB1, ITM2B, ITPA, JAM3, KCNJ13, KIAA1109, KLC1, KLRG1, LAMB2, LAMC1, LAMP2, LARGE1, LCA5, LCT, LCTL, LEMD2, LGR4, LGSN, LIM2, LMX1B, LONP1, LOXLI, LRP2, LRP5, LRP5L, LSS, LTBP2, LTBP3, MAB21L2, MAF, MAFA, MAFIP, MAG11, MAN2B1, MED12, MIP, MIPEP, MIR184, MMP1, MMP2, MOCS1, MOCS2, MTHFR, MUC16, MVK, MYH9, MYO7A, MYOC, NAT2, NAT8, NBN, NCOA6, NDP, NECTIN3, NEIL2, NEU1, NF2, NHS, NID1, NOD2, NOG, NPHP1, NR2E3, NRCAM, NSUN2, NUDT19, NUP188, OAT, OCRL, OGG1, OPA1, OPA3, OSGEP, OTX2, P2RY2, P3H2, PANK4, PARP1, PAX2, PAX6, PCBD1, PDE6B, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PITX2, PITX3, PNPT1, POLG, POLR3B, POMGNT1, POMT1, POMT2, PON2, PORCN, PQBP1, PRDX6, PROX1, PRX, PSMC3, PTCH1, PTEN, PTH, PXDN, PYCRI, RAB3GAP1, RAB3GAP2, RECQL4, REST, RGS6, RIC1, RIN2, RNF149, RNF2, RNLS, RNPC3, RP2, RPE65, RPGR, RRAGA, SALL2, SALL4, SC5D, SCFD2, SCHIP1, SEC23A, SF3B1, SH3D19, SIL1, SIPA1L3, SIX5, SIX6, SLC11A2, SLC16A12, SLC23A1, SLC23A2, SLC25A15, SLC33A1, SLC38A1, SLC40A1, SLC4A4, SLC7A8, SMAD4, SMARCA4, SMO, SMOX, SORD, SOX2, SPARC, SPAST, SRD5A3, SUOX, TAF1A, TAPI, TAP2, TAPBP, TAPT1, TBC1D20, TDRD7, TELO2, TENM3, TFAP2A, TGFB3, TGFB1, TMC03, TMEM114, TNPO1, TOR1AIP1, TP53, TRAPPC11, TRNT1, TRPM3, TSPAN12, TSR1, TUBA1A, UBE2A, UBR1, UNC45B, USH1C, USH2A, VCAN, VIM, VLDLR, VSX2, WDR36, WDR87, WFS1, WNT3, WRN, WTI, XPC, XRCC1, XYLT2, YAP1, YWHAE, ZNF350

眼科- Retina_disease

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AH11, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMS2, ARSG, ASRGL1, ATF6, ATG5, ATOH7, ATXN7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C12orf65, C1QTNF5, C2, C3, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP78, CERKL, CFAP410, CFB, CFH, CHM, CIB2, CLCC1, CLN3, CLRN1, CLRN3, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL9A1, CRB1, CRX, CSPPI, CTNNA1, CYP4V2, DHDDS, DHX38, DMD, DRAM2, DTHD1, EFEMP1, ELOVL1, ELOVL4, EMCI, ERCC6, ESPN, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FSCN2, FZD4, GARI, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS1, HBM, HGSNAT, HK1, HMCN1, HMX1, HTRA1, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSDB, MIR204, MKKS, MKS1, MTPP, MVK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OR2W3, OTX2, PAF1, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX26, PEX7, PGK1, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBI, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLB1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, SLC7A14, SNRNP200, SPARC, SPATA7, SPP2, TEAD1, TIMM8A, TIMP3, TLR3, TLR4, TMEM126A, TMEM216, TMEM237, TMEM67, TOPORS, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, VSX2, WDPCP, WDR19, WFS1, WHRN, ZNF408, ZNF423, ZNF513

耳鼻喉科- Non-syndromic hearing loss

ACTG1, ADCY1, AIFM1, BDPI, BSND, CABP2, CCDC50, CD164, CDH23, CEACAM16, CIB2, CLDN14, CLDN9, CLIC5, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRAP, GRHL2, GRXCRI, GRXCR2, GSDME, HGF, HOMER2, IFNLRI, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAP1B, MARVELD2, MCM2, MET, MIR96, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PCDH9, PDE1C, PDZD7, PJVK, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, WBP2, WFS1, WHRN

耳鼻喉科- Syndromic hearing loss
ADGRV1, ATP6V1B2, CDH23, CHD7, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, EDN3, EDNRB, ERAL1, EYA1, FGFR2, FOXI1, GATA3, HARS1, HARS2, HSD17B4, KCNE1, KCNJ10, KCNQ1, LARS2, MITF, MYO7A, NDP, PAX3, PCDH15, POLR1C, POLR1D, SEMA3E, SIX1, SIX5, SLC26A4, SNAI2, SOX10, TCOF1, TWNK, USH1C, USH1G, USH2A, WHRN
婦兒科-先天性別發育異常疾病
AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARL6, ARX, ATRX, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BMP15, BMP4, BMP7, BNC2, BUB1, BUB1B, CBX2, CC2D2A, CCNQ, CDKN1C, CDON, CEP120, CEP290, CEP41, CEP55, CEP57, CHD4, CHD7, CILK1, CKAP2L, COX7B, CREBBP, CSPP1, CTU2, CUL7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHCR24, DHCR7, DHH, DHX37, DLL1, DMRT1, DMRT2, DNMT3B, DYNC2H1, DYNC2I1, DYNC2I2, EFNBI, EPG5, ESCO2, EVC, EVC2, FAT4, FBXL4, FDXR, FEZF1, FGF10, FGF8, FGFRI, FGFR2, FGFR3, FIG4, FLNA, FOXL2, FRAS1, FREM2, FSHR, FUZ, FZD2, GATA4, GLI2, GLI3, GNRH1, GNRHR, GPC3, GRIP1, HCCS, HESX1, HFE, HHAT, HNF1B, HOXA13, HOXA4, HOXB6, HOXD13, HSD17B3, HSD17B4, HSD3B2, IFT80, IFT81, IGF2, IL17RD, INSL3, IRF6, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LMNA, MAMLD1, MAP3K1, MCM9, MED12, MID1, MKKS, MKS1, MYRF, NDUFB11, NEK1, NODAL, NR0B1, NR2F2, NR3C1, NR5A1, NSMF, OPHN1, PAX6, PBX1, PCNT, PCSK1, PDE4D, PEX1, PHGDH, PITX2, POR, PROK2, PROP1, PSAT1, PSMC3IP, PTCH1, PTDSS1, PTPN11, RBBP8, RIPK4, ROR2, RPGRIP1, RPGRIP1L, RSP01, SALL1, SC5D, SEMA3A, SETBP1, SHH, SIX3, SOS1, SOX10, SOX2, SOX3, SOX9, SPECC1L, SRD5A2, SRY, STAR, SUFU, TAC3, TACR3, TBC1D24, TBX15, TCTN2, TGIF1, TMEM107, TMEM216, TMEM231, TMEM67, TMEM70, TNXB, TOE1, TP63, TRAIP, TRIM32, TRIP13, TSPYL1, TTC9, TWIST2, UBR1, VANGL1, WDPCC, WDR35, WNT4, WNT5A, WNT7A, WT1, WWOX, ZEB2, ZFPM2, ZIC2
婦兒科-先天神經肌肉疾病
ABHD5, ACAD9, ACADL, ACADM, ACADVL, ACAT1, ACTA1, ACTB, ACVRI, AGRN, AIFM1, ALDH7A1, ALG13, ALG14, ALG2, AMPD1, ANO3, ANO5, ARHGEF10, ATM, ATP1A1, ATP1A3, B3GALNT2, BICD2, BIN1, BSCL2, BVES, CAPN3, CAV3, CFL2, CHAT, CHKB, CHRNA1, CHRN1, CHRND, CHRNE, CHRNG, CIZ1, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPT1A, CPT1B, CPT2, CTDPI, DAG1, DBH, DCTN1, DDC, DES, DHTKD1, DMPK, DNAJB2, DNAJB6, DNM2, DOK7, DYNC1H1, DYSF, EGR2, EMD, ETFA, ETFB, ETFDH, FBLN5, FGD4, FHL1, FIG4, FKRP, FKTN, FLNC, GAA, GAN, GBE1, GCDH, GCH1, GDAP1, GJB1, GMPPB, GNAL, GNB4, GNE, GYG1, GYS1, HADHA, HADHB, HARS1, HINT1, HK1, HNRNPDL, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, ISCU, ITGA7, JPH1, KARS1, KBTBD13, KIF1B, KIF5A, KLHL40, KLHL41, KLHL9, LAMA2, LAMB2, LARGE1, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPINI, LRSAM1, MATR3, MED25, MEGF10, MFN2, MICALI, MME, MORC2, MPZ, MSTN, MTM1, MTMR2, MYOT, NDRG1, NEB, NEFH, NEFL, PABPN1, PAH, PDK3, PFKM, PGAM2, PGM1, PHKA1, PLEC, PLEKHG5, PMP2, PMP22, PNKD, PNPLA2, PNPO, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKRA, PRPS1, PRRT2, PRX, PTS, PYGM, QDPR, RAB7A, RAPSN, RBCK1, RYR1, SBF1, SBF2, SGCA, SGCB, SGCD, SGCE, SGGC, SH3TC2, SLC12A6, SLC18A2, SLC22A5, SLC25A20, SLC2A1, SLC6A3, SMCHD1, SPEG, SPG11, SPR, STAC3, STIM1, SURF1, SYNE1, SYNE2, TAF1, TCAP, TDP1, TFG, TH, THAP1, TIA1, TMEM43, TNNT1, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM3, TRIM32, TRIM63, TRPV4, TTN, TUBA8, TUBB4A, TYMP, VCP
婦兒科-先天骨骼及結締組織疾病
ACTA2, ADAMTS2, ALX4, B3GALT6, B4GALT7, BMP1, CBS, CHST14, CLCN7, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, CREB3L1, CRTAP, DSE, ERF, FBN1, FBN2, FGFRI, FGFR2, FGFR3, FKBP10, FKBP14, FLNA, FLNB, IFITM5, IKBKG, LRP5, MBTPS2, MSX2, OSTM1, P4HB, PLOD1, PLOD2, PLS3, PPIB, PRDM5, RIN2, SEC24D, SERPINF1, SERPINH1, SLC39A13, SMAD3, SMAD6, SP7, SPARC, TCF12, TCIRG1, TGFB2, TGFB3, TGFBRI, TGFBRI2, TMEM38B, TNFRSF11A, TNFRSF11, TNXB, TRAPPC2, TRPV4, TWIST1, WNT1, XYLT2, ZIC1, ZNF469
婦兒科-先天新陳代謝疾病
ABCD1, ACAD9, ACADL, ACADM, ACADVL, ACAT1, ACOX1, ARG1, ARSA, ARSB, ARSL, ASAH1, ASL, ASS1, ATP13A2, AUH, BTD, CLN3, CLN5, CLN6, CLN8, CPS1, CPT1A, CPT1B, CPT2, CSF1R, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, DDC, DNAJC5, DPM1, DPM2, DPM3, DTNBP1, EBP, ELN, ETFA, ETFB, ETFDH, FUCA1, GAA, GALC, GALNS, GBA, GJB2, GLA, GLB1, GM2A, GNE, GNPTAB, GNPTG, GNS, GRN, GUSB, GYS1, HADHA, HADHB, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HSD17B10, HYAL1, IDS, IDUA, IVD, LAMP2, LAMP3, LIPA, LMNB1, LPINI, LPIN2, MAN2B1, MANBA, MCCC1, MCCC2, MCOLN1, MFSD8, MMAA, MMAB, MMACHC, MMADHC, MMUT, NAGA, NAGLU, NAGS, NEU1, NPC1, NPC2, OTC, PC, PCCA, PCCB, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGM1, PHKA1, PHYH, PPARA, PPT1, PRRT2, PSAP, PYGM, SGSH, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SMPD1, SUMF1, TPP1, TYMP
婦兒科-遺傳性腎臟疾病
ABCC8, ACE, ACTN4, ADAMTS13, ADCY10, AGTR1, AGXT, AH1, ALAD, ALAS2, ALG1, ALG8, ALMS1, ANKS6, ANLN, ANOS1, AP2S1, APOE, APOL1, APRT, AQP2, ARHGAP24, ARHGAP24, ARL13B, ARL6, ARMC5, ATG5, ATP1A1, ATP2B3, ATP6V0A4, ATP6V1B1, ATP7B, ATXN10, AVPR2, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICCI, BMP4, BMP7, BSND, C1S, C2, C3, C3AR1, C4BPA, C7, C8A, C9, CA2, CACNAID, CACNAIH, CACNAIS, CAPN12, CASR, CC2D2A, CCL2, CD151, CD2AP, CD46, CDC5L, CDK20, CEP164, CEP290, CEP41, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN16, CLDN19, CLU, CNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ7, COQ9, CPOX, CRI, CR2, CRB2, CTNS, CUBN, CUL3, CYP11B2, CYP1B1, CYP24A1, DCDC2,

DGKE, DLC1, DLX5, DLX6, DMP1, DNASE1, DSTYK, DYNC2H1, DYNC2H11, DZIP1L, EGF, EHHADH, EMP2, ENPP1, EYA1, F12, FAM20A, FAN1, FAT1, FBXW7, FCGR2A, FCGR3B, FCN1, FECH, FGF23, FN1, FOXC1, FRAS1, FREM1, FREM2, FXYD2, GALNT3, GANAB, GAPVD1, GATA3, GATM, GLA, GLB1, GLIS2, GLIS3, GNA11, GRHRP, GSN, HFE, HMBS, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, HSD3B2, IFT122, IFT140, IFT172, IFT43, INF2, INPP5B, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITGB4, ITSN1, ITSN2, JAG1, KANK2, KANK4, KAT2B, KCNA1, KCNJ1, KCNJ10, KCNJ12, KCNJ2, KCNJ5, KCNK5, KCNMB1, KIF7, KL, KLHL3, LAGE3, LAMA5, LAMB2, LCAT, LCORL, LMNA, LMX1B, LPL, LRP2, LRP4, LRP5, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MASP1, MASP2, MEJV, MET, MKKS, MKS1, MMACHC, MMP14, MYH9, MYO1E, MYOG, NEK1, NEK8, NF1, NOS3, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NUP107, NUP205, NUP93, NXF5, OCRL, OFD1, OSGEP, PAX2, PCBD1, PDSS2, PHB, PHEX, PIEZO2, PKD1, PKD2, PKHD1, PLCE1, PLG, PMM2, PODXL, PPOX, PTH1R, PTPRO, REN, RET, RMND1, ROBO2, RPGRIPL, RRM2B, SALL1, SALL4, SARS2, SCARB1, SCARB2, SCN4A, SCNN1A, SCNN1B, SCNN1D, SCNN1G, SDCCAG8, SEC61A1, SGPL1, SIAH1, SIX1, SIX2, SIX5, SLC12A1, SLC12A2, SLC12A3, SLC12A5, SLC12A7, SLC16A12, SLC22A12, SLC26A3, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC4A5, SLC5A1, SLC5A2, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC9A1, SLC9A3, SLC9A3R1, SLIT2, SMARCAL1, SOX17, STAR, STRA6, STS, STX16, SYNPO, TCTN1, TCTN2, TCTN3, THBD, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TNXB, TP53RK, TP63, TPRKB, TRIM32, TRNT1, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, UMOD, UPK3A, UROD, UROS, USF2, VDR, VHL, VIPAS39, VPS33B, VWF, WDR19, WDR35, WDR73, WHAMM, WNK1, WNK4, WNT4, WT1, WT1-AS, XDH, XPNPEP3, ZNF423

神經科- Young stroke

ABCA1, ABCC6, ABCG5, ABCG8, ABHD2, ABO, ACAD9, ACTA2, ACTB, ACTC1, ACTG1, ACVRL1, ADA2, ADAMTS13, ADAMTS7, AGXT, ALOX5AP, ANGPTL4, ANKRD31, ANKS1A, APOA1, APOA4, APOA5, APOB, APOC4, APOE, APOL1, APP, ASS1, ATP2B1, ATP7A, B3GALT6, BCAS3, CALR, CBS, CCM2, CD59, CFTR, CITED2, CNM2, CNTNAP2, COL3A1, COL4A1, COL4A2, CPS1, CRELD1, CST3, CTSA, CXCL12, CYP11B1, CYP17A1, DPM3, DYRK1B, EDNRA, ELN, ENG, ERCC6, ERCC8, F2, F5, FAM20A, FBN1, FES, FGA, FGB, FGG, FLNA, FLNC, FLT1, FOXE3, FREM1, FREM2, FURIN, FZD2, GATA4, GATA6, GDF2, GGCX, GIP, GJB2, GLA, GNAQ, GUCY1A1, GYS1, HBB, HDAC9, HHIPL1, HMGCR, HSD11B2, HTRA1, ITGA7, JAG1, JAK2, KCNE2, KCNK5, KCNQ1, KRAS, KSR2, LAMA3, LAMA5, LCAT, LDLR, LDLRAP1, LIPA, LMNA, LOX, LPA, LPL, MBOAT7, MC4R, MECP2, MFAP5, MFGE8, MIA3, MLXIPL, MMACHC, MMUT, MPL, MRAS, MRPS6, MTHFR, MTR, MTRR, MYBPC3, MYD88, MYH11, MYH6, MYLK, MYPN, NAGS, NF1, NKX2-5, NOS3, NOTCH3, NPPA, NR2F2, NR3C1, NT5C2, OTC, PCNT, PCSK9, PDGFD, PDK2, PEMT, PHACTR1, PHGDH, PIGA, PIK3C2A, PKD1, PLG, PMAIP1, PMM2, PNP, POM121L9P, PRDX6, PRKAG2, PRKCH, PRKG1, PRNP, PROC, PROS1, RASD1, REST, RFT1, RNF213, SCIMP, SCN5A, SEMA4A, SERPINC1, SERPINH1, SH2B3, SLC19A2, SLC22A4, SLC22A5, SLC2A10, SLC5A3, SMAD3, SMAD4, SMARCAL1, SMG6, SNAP29, SNF8, SORT1, SRR, STIM1, SVEP1, SWAP70, TANGO2, TBX20, TCF21, TET2, TFPI, TGFB1, TGFB2, TGFB3, TGFBRI, TGFBRI2, THPO, THSD1, TLL1, TNNT2, TNXB, TP53, TP53INP1, TREX1, TRIB1, TTR, UBE2Z, VAMP5, VAMP7, VAMP8, VHL, VWF, WDR12, WFS1, XYLT1, XYLT2, YAP1, ZAP70, ZC3HC1, ZEB2, ZMPSTE24, ZNF507

腫瘤醫學-遺傳癌症

ABC11, ALK, APC, ATM, AXIN2, BAP1, BLM, BMPRIA, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, COL7A1, CYLD, DDB2, DICER1, DIS3L2, DKC1, DOCK8, EGFR, ELANE, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, FAH, FANCA, FANCC, FANCG, FH, FLCN, GATA2, GBA, GJB2, GPC3, HFE, HMBS, HRAS, ITK, KIT, MAX, MEN1, MET, MLH1, MSH2, MSH6, MTAP, MUTYH, NBN, NF1, NF2, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POLH, PRKARIA, PRSS1, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RMRP, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SH2D1A, SLC25A13, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SRY, STAT3, STK11, SUFU, TERT, TGFBRI, TMEM127, TP53, TRIM37, TSC1, TSC2, UROD, VHL, WAS, WRN, WT1, XPA, XPC