

Appendix 1

Choice A

Tier 1

ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ATP2B2, ATP6V1B1, BCS1L, BDP1, BSND, CABP2, CACNA1D, CATSPER2, CCDC50, CDH23, CEACAM16, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FGF3, FOXI1, GATA3, GIPC3, GJB1, GJB2, GJB3, GJB4, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS, HARS2, HGF, HOMER2, HOXA2, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, LARS2, LHFPL5, LHX3, LOXHD1, LRTOMT, MARVELD2, MASP1, MIR96, MITF, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NR2F1, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX2, PAX3, PCDH15, PDZD7, PMP22, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SALL1, SALL4, SERPINB6, SIX1, SIX5, SLC17A8, SLC26A4, SLC4A11, SLITRK6, SMPX, SNAI2, SOX10, SOX2, STRC, SYNE4, TBC1D24, TECTA, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, USH1G, USH2A, WFS1

Tier 2

ABHD5, ABR, ACAN, ALDH1A2, ANKH, AP3D1, APAF1, APOPT1, AQP4, ARSB, ATF2, ATOH1, ATP1A2, ATP6V0A4, ATP8B1, AXIN1, BARHL1, BBS1, BBS2, BBS4, BBS10, BBS12, BCAP31, BCL2, BCL2L1, BCR, BDNF, BLOC1S5, BLOC1S6, BMP4, BSN, BTD, CACNB2, CACNG2, CASP3, CD151, CDKN1B, CDKN1C, CDKN2D, CELSR1, CHRNA9, CHSY1, CKB, CLDN11, CLDN9, CLNS1A, COL4A3, COL4A4, CPLX1, CRYL1, DDB2, DDR1, DIO2, DIO3, DLX2, DLX5, DMD, DVL1, DVL2, DVL3, EPHB1, EPHB2, EPHB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ESR2, FABP4, FAM65B, FAS, FBXO10, FBXO2, FGFR1, FGFR2, FGFR3, FIGN, FKBP14, FLNA, FOXC1, FOXG1, FZD3, FZD6, GBX2, GFER, GFI1, GJA1, GJA1P1, GJB5, GLI3, GPX1, GRID1, GSTM1, GSTP1, GSTT1, GTPBP3, GUSB, HAL, HES1, HES5, HMX2, HMX3, HOXA1, HOXB1, IFT88, IGF1, ITGA8, JAG1, JAG2, KCNMA1, KIT, KITLG, LAMA2, LARGE, LFNG, LMO4, LMX1A, LRIG3, LRP2, MAFB, MANBA, MAP1A, MCL1, MCOLN3, MIR182, MIR183, MKKS, MOS, MPV17, MSX2, MTAP, MTO1, MYO1A, MYO1C, MYO1F, NAV2, NDP, NEU1, NEUROD1, NEUROG1, NF1, NLRP3, NOTCH1, NOX3, NOXO1, NR4A3, NTF3, NTN1, NTRK2, NTRK3, OC90, OTOP1, OTOR, OTX1, OTX2, PCDH20, PET100, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHEX, PITX2, PNOC, POLD1, POLH, POLR1C, POLR1D, POU1F1, PROP1, PRRX1, PRRX2, PSIP1, PTK7, RARA, RARB, RARG, RASA1, RPGR, RPS6KA3, S1PR2, SCARB2, SCRIB, SDHD, SEMA3E, SERAC1, SLC12A2, SLC12A6, SLC12A7, SLC19A2, SLC1A3, SLC26A5, SLC28A3, SLC29A3, SLC30A4, SLC33A1, SLC4A7, SLC9A1, SMS, SOBP, SOD1, SOX9, SPINK5, SPRY2, ST3GAL5, SYNJ2, TBL1X, TBX1, TBX10, TCF21, TCOF1, TFAP2A, TFB1M, TGFA, TGFB2, THRA, THRB, TMC2, TMPRSS5, TNFRSF11B, TRMU, TRPV4, TSHR, TUB, TYR, TYRP1, UCN, VANGL2, XPA, XPC, YAP1.

Choice B

APC, ACVRL1, KRIT1, LHX3, VWF, TAZ, NKX2-1, F2, ATP7A, PTPN11, COL1A2, CTNS, GBA, GCH1, CPT2, CBS, HPS1, HPS4, PYGM, P2RY12, DNMT3B, PIK3CD, NPC1, PHKA2, MEFV, RET, GIF, SLC19A3, TTPA, PALB2, RAF1, SLC19A2, DSP, ALDOB, F8, F9, TG, TPO, GAA, ZAP70, SLC37A4, F10, F7, IL7R, GALT, BTD, PROP1, OTC, TSHB, MMACHC, HBB, FAH, MMAA, MMAB, MUT, HADH, G6PC, PAH, ACAT1, F5, PTPRC, SPR, POU1F1, SLC7A7, CPT1A, TSHR, F13A1, PAX8, CFTR, MTR, MTRR, GATA2, ACADM, RB1, ASS1, RAG1, UNC13D, CACNA1C, SLC46A1, CYP27B1, IYD, SLC5A5, PHKG2, RAG2, CD3D, HADHA, IL2RG, JAK3, NAGS, OTC, HSD3B2, CYP21A2, STAR, DUOX2, DUOXA2, IVD, PCCA, PCCB, CPS1, SLC25A20, HADHB, HLCS, FGFR3, ASL, F13B, ETFA, ETFB, ETFDH, BCKDHA, BCKDHB, DBT, INS, ALDH7A1, PDX1,

DCLRE1C, GG CX, FBP1, ACADVL, MPI, GCDH, MTHFR, ATP7B, VWF, ELN, KCNH2, SCN5A, LDLR, JUP, RYR1, TGFBR1, TGFBR2, MYH7, LMNA, BCHE, ENG, BMPR1A, APOB, KCNJ2, AGL, CARD11

Choice C

ABCA12, ABCB11, ABCC2, ABCC6, ABCC8, ABCD1, ABCG5, ACADS, ACVR1, ADA, ADAMTS13, AHI1, ALAS2, ALDH3A2, ALDH5, A1ALS2, ANKH, APRT, APTX, AR, ARG1, ARSB, ARX, ASPA, ATM, ATP8B1, ATRX, AVPR2, BLM, BRAF, BSCL2, BTK, CA2, CAPN3, CAV3, CD40LG, CDAN1, CDKL5, CDKN1C, CHD7, CHM, CLCN1, CLCN5, CLCN7, CLN3, CLN5, CLN6, CNGB3, COL17A1, COL1A1, COL7A1, COLQ, CREBBP, CRLF1, CTSK, CUL7, CYBA, CYBB, CYP27A1, DCX, DGUOK, DHCR7, DMD, DNAH11, DNAH5, DOCK8, DOK7, DSC2, DSG2, DSP, EDA, EDAR, EIF2AK3, ELANE, ELN, EMD, EPM2A, EVC, EVC2, EXT1, EXT2, F11, FANCA, FANCC, FANCG, FBN1, FGA, FGB, FGD1, FH, FKRP, FKTN, G6PD, GABRG2, GALC, GALK1, GALNS, GAN, GATA4, GBE1, GCK, GDAP1, GLA, GLB1, GLDC, GNAS, GNPTAB, GPC3, GPR143, GPR56 (ADGRG1), GSS, GYS2, HBA1, HBA2, HEXA, HEXB, HGSNAT, HMGCL, HNF4A, HPRT1, HRAS, HSD17B3, IDS, IDUA, IKBKAP, IKBKG, INSR, IRF6, ITGB4, KCNJ11, KCNQ2, KCNQ3, KMT2D, KRAS, KRT14, KRT5, L1CAM, LAMA3, LAMB2, LAMB3, LAMC2, LMX1B, LRP5, MAP2K1, MAP2K2, MCCC1, MCCC2, MCOLN1, MLC1, MTM1, MYH3, NAGLU, NCF1, NCF2, NDP, NIPBL, NKX2-5, NOTCH3, NPHS1, NROB1, NSD1, NTRK1, OCRL, OFD1, OPA1, PAX6, PC, PCNT, PDHA1, PEX7, PKHD1, PLP1, PMM2, POMT1, POR, PORCN, PPT1, RAI1, RECQL4, RPS6KA3, RUNX2, SCN1A, SCNN1A, SCNN1B, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SHH, SIX3, SLC12A1, SLC25A38, SLC26A3, SLC2A1, SLC2A10, SLC34A3, SLC39A4, SLC3A1, SLC45A2, SLC5A2, SLC6A19, SLC6A8, SLC7A9, SMAD3, SMARCAL1, SMC1A, SMPD1, STAT3, STS, SURF1, TAT, TBX5, TCIRG1, TNNI2, TSC1, TSC2, TWIST1, UGT1A1, VDR, WAS