

Basic Carrier Screen

Genes (3):

CFTR, SMN1, FMR1

Expanded Carrier Screen (v1.2)

Genes (1,424):

*AAAS, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCD1, ABCD4, ABHD5, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1,*ACE, ACO2, ACOX1, ACTA1, ADA, ADA2, ADAMTS13, ADAMTS2, ADAMTSL2, ADAR, ADAT3, ADGRG1, ADGRV1, ADK, ADPRS, ADSL, AFF2, AGA, AGBL5, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGXT, AHCY, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG3, ALG6, ALG8,*ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, AMH, AMHR2, AMN, AMPD2, AMT, ANKS6,*ANO10, ANO5,*ANTXR1, ANTXR2, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4M1, AP4S1, APTX, AQP2, AR, ARFGEF2, ARG1, ARHGEF9, ARL13B, ARL6, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX,*ASAH1, ASCC1, ASL, ASNS, ASPA, ASPM, ASS1, ATAD1, ATCAY, ATF6, ATM, ATOH7, ATP13A2, ATP6AP1, ATP6V0A2, ATP6V0A4, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATR, ATRX, AUH, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT7, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCHE, BCKDHA, BCKDHB, BCKDK, BCS1L, BGN, BIN1, BLM, BLOC1S3, BLOC1S6, BLTP1, BMP1, BMPER, BMPR1B, BOLA3, BRAT1, BRF1, BRIP1, BRWD3, BSCL2, BSND, BTB, BTK, BUB1B, C12orf57, C19orf12, C1QA, C1QB, C1QC, C2CD3, C3, C5, CA2, CABP4, CAD, CANT1, CAPN3, CARD11, CARS2, CASK, CASP14, CASQ2, CASR, CAVIN1, CC2D1A, CC2D2A, CCB1, CCDC103, CCDC115, CCDC39, CCDC40, CCDC8, CCDC88C, CCN6, CCNO, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD8A, CDAN1, CDC45, CDCA7, CDH11, CDH23,*CDH3, CDK10, CDK5RAP2, CDT1, CENPJ, CEP104, CEP120, CEP152, CEP290,*CEP41, CEP78, CERKL, CERS3, CFAP410, CFAP418, CFD, CFH,*CFI,*CFL2, CFP, CHAT, CHKB, CHM, CHMP1A, CHRNA1, CHRND, CHRNE, CHRNA1, CHST14, CHST3, CHST6, CHSY1,*CIB2, CIITA, CISD2, CIT, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKB, CLDN1, CLDN10, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3,*CNNM4, CNTNAP1, CNTNAP2, COA8, COASY, COG6, COG7, COL11A1, COL11A2,*COL17A1, COL18A1, COL27A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COLEC11, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ8B, COX10, COX15, COX20, COX6B1, CP, CPLANE1,*CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRLF1,*CRPPA, CRTAP, CRYL1, CSPP1, CSTB, CTC1, CTNS, CTPS1, CTSA, CTSC, CTSD, CTSF, CTSK, CUL4B, CUL7, CWC27,*CYB5R3, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DARS1, DARS2, DBT, DCAF17, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD2, DDR2, DDX11, DDX59, DENND5A, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHODH, DIS3L2, DKC1, DLAT, DLD, DLG3, DLL3, DMD, DNAAF1, DNAAF11, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC12, DNAJC19, DNAJC21, DNAJC6, DNAL1,*DNMT3B, DOCK2, DOCK6, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPH1, DPYD, DSP, DTNBP1, DUOX2, DUOX2A, DYM, DYNC2H1,*DYNC2I1, DYNC2I2, DYNC2LI1, DYSF, EARS2, ECEL1, ECHS1, EDA, EDAR, EFEMP2, EFN1 (analysed in males only), EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ELP1, ELP2, EMD, EML1, ENPP1,*EOGT, EPB42, EPCAM, EPG5, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2,*ERCC8, ESCO2, ETFA, ETFB, ETV2, ETV4, ETV5, EVC,*EVC2, EXOSC3, EXOSC8, EXTL3, EYS, F11, F2, F5, F7, F8, F9, FA2H, FAH, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FARS2, FAT4, FBLN5, FBP1, FBXL4, FBXO7, FERMT1, FERMT3, FGA, FGB, FGD1, FGD4, FGG, FH, FHL1, FIG4, FKBP10, FKBP14, FKBP, FKTN, FLAD1, FLNA, FLNB, FLVCR1, FLVCR2, FMO3, FOLR1, FOXE3, FOXN1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRRS1L, FTCD, FTO, FTSJ1, FUCA1, FXN, FYCO1, G6PC1, G6PC3, GAA, GALT, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GAN, GAS8, GATM, GBA,*GBA2, GBE1, GCDH, GCH1,*GDAP1, GDF1, GDF5, GDI1, GFM1, GFPT1, GH1, GHR, GHRHR, GJA1, GJB1, GJB2, GJB6,*

GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLIS3, GLYCTK, GM2A, GMPPA, GMPPB, GNAT2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNRHR, GNS, GORAB, GOSR2, GP1BA, GP9, GPAA1, GPC3, GPC6, GPHN, GPR143, GPR179, GPSM2, GPT2, GRHPR, GRIP1, GRM1, GSS, GTF2H5, GTPBP3, GUCY1A1, GUCY2C, GUCY2D, GUSB, GYS2, HACE1, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HCFC1, HELLS,*HEPACAM, HES7, HESX1, HEXA, HEXB,*HGD, HGSNAT,*HIBCH, HINT1, HJV, HK1, HLCS, HMGCL, HMGCS2, HMOX1, HOGA1, HOXA1, HPD, HPGD, HPRT1,*HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPD1, HSPG2, HTRA2, HUWE1, HYAL1, HYLS1, IARS1, IARS2, IBA57, ICOS, IDH3B, IDS, IDUA, IER3IP1, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT80, IGF1R, IGHMBP2, IGSF1, IKBKB, IL10RA, IL10RB, IL11RA, IL12RB1, IL17RA, IL1RAPL1, IL1RN, IL2RA, IL2RG, IL7R, INPP5E, INPP5K, INPPL1, INSR, INVS, IQCB1, IQSEC2, ISCA2, ITCH, ITGA2B, ITGA6, ITGB2, ITGB3, ITGB4, ITK, ITPA, ITPR1, IVD, IYD, JAGN1, JAK3, JAM3, JUP, KATNB1, KCNJ1, KCNJ10, KCNJ11, KCNQ1, KCNV2, KCTD7, KDM5C, KIAA0586,*KIF14, KIF1A, KIF1C, KIF7, KIFBP, KLHL40, KLHL41, KLHL7, KNL1,*KPTN, KRT10, KRT14, KRT5, KY, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LARGE1, LARP7, LARS1, LARS2, LAT, LBR, LCA5, LCAT, LCK, LDHA, LDLR, LDLRAP1, LEP, LGI4, LHCGR, LHX3, LIAS, LIFR, LIG4, LINS1, LIPA, LIPC, LIPN, LIPT1, LMAN1, LMBR1,*LMBRD1,*LMNA, LMOD3, LONP1, LOXHD1, LPAR6, LPIN1, LPIN2, LPL, LRAT, LRBA,*LRIG2, LRMDA, LRP2, LRP4, LRP5, LRPPRC, LRSAM1, LTBP3, LTBP4, LYRM7,*LYST, LZTFL1, MAK,*MALT1,*MAN1B1, MAN2B1, MANBA, MAOA, MAPKBP1, MARS1, MARS2, MASP1, MAT1A, MBOAT7, MBTPS2, MC2R, MCEE, MCFD2, MCOLN1, MCPH1, MEC2, MED12, MED17, MED23, MED25, MEFV, MEGF10, MEGF8, MERTK, MESP2, METTL23, MFN2, MFSD2A, MFSD8, MGAT2, MGME1, MGP,*MICU1,*MID1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP2, MMP21, MMUT, MOCS1, MOCS2, MPDZ, MPI, MPL, MPLKIP, MPV17, MPZ, MRAP, MRE11, MTFMT, MTHFD1, MTHFR, MTM1, MTMR2, MTO1, MTR, MTRR, MTPP, MUSK, MVK, MYD88, MYMK, MYO15A, MYO5B, MYO7A, NAA10, NAGA, NAGLU, NAGS, NALCN, NANS, NARS2,*NAXE, NBAS, NBEAL2, NBN, NCF2, NCF4, NDE1, NDP, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECTIN1, NEK1, NEK8, NEU1, NEXMIF, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHS, NIPAL4, NKX3-2, NKX6-2, NMNAT1, NNT, NONO, NPC1, NPC2, NPHP1,*NPHP3,*NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR2E3, NSDHL, NSUN2, NT5C2, NTRK1, NUBPL, NUP107, NUP93, NYX, OAT, OBSL1, OCRL, ODAD1, ODAD3, OFD1, OPA1, OPA3, OPHN1, ORAI1, ORC1, ORC6, OSGEP, OSTM1, OTC, OTOF, OTUD6B, P3H1, PAH, PAK3, PANK2, PAPSS2, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH15, PCDH19 (analysed in males only), PCNT, PCSK1, PCYT1A, PDE6A, PDE6C, PDHA1, PDHB, PDHX, PDP1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP1,*PGAP2, PGAP3, PGK1, PGM1, PGM3, PHF6, PHF8, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIBF1, PIEZO2, PIGA, PIGG, PIGL, PIGN, PIGO, PIGT, PIGV, PIP5K1C, PIVK, PKHD1, PKLR, PLA2G6, PLAA, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLOD2,*PLP1, PLPBP, PMM2, PMPCA, PNKP, PNP, PNPLA1, PNPLA6, PNPO, POC1A, POLG, POLH, POLR1C, POLR3A, POLR3B, POMC, POMGNT1, POMGNT2, POMK, POMP, POMT1, POMT2, POP1, POR, POU1F1, POU3F4, PPA2,*PPIB, PPT1, PQBP1, PRCD, PRDM12,*PRDM5, PREPL, PRF1, PRG4, PRICKLE1, PRKDC, PRKRA, PROC, PROP1, PROS1,*PRPS1, PRUNE1, PRX, PSAP, PSAT1, PSMB8, PSPH, PTH1R, PTPN23, PTPRC,*PTS, PUS1, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, RAB18, RAB23, RAB27A, RAB33B, RAB39B, RAB3GAP1, RAB3GAP2,*RAD50, RAG1, RAG2, RAPSN, RARB, RARS1, RARS2, RAX, RBBP8, RBCK1, RBM10, RCBTB1, RD3, RDH12, RDH5, RECQL4, REEP6, REN, RETREG1, RFT1, RFX5, RFX6, RFXANK, RFXAP, RHAG, RIN2, RIPK4, RLBP1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO3, ROGDI, ROR2, RORC, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L,*RPL10, RPS6KA3, RRM2B, RS1, RSPH1, RSPH4A, RSPH9, RTEL1, RTN4IP1, RTTN,*RXLYT1, RYR1,*SACS, SAG, SAMD9, SAMHD1, SAR1B, SARS2, SBDS, SBF2, SC5D, SCARB2, SCARF2, SCN9A, SCNN1A, SCNN1B, SCO1, SCO2, SCYL1, SDCCAG8, SDHAF1, SDR9C7, SEC23B, SELENON,*SEPSECS, SERAC1, SERPINF1, SERPINH1, SETX, SFTPB, SGCA, SGCB, SGCD, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SH3TC2, SIL1, SKIV2L, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC24A5, SLC25A1, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A38, SLC25A46, SLC26A2, SLC26A3, SLC26A4, SLC27A4, SLC29A3, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC34A3, SLC35A3, SLC35D1,*SLC37A4, SLC38A8, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC45A2, SLC46A1, SLC4A1,

SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A5, SLC5A7, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A3, SLC9A6, SMARCA1, SMPD1, SMS, SNAP29, SNX10, SNX14, SOST, SP110, SPAG1, SPART, SPATA5, SPATA7, SPEG, SPG11, SPG21, SPG7, SPINK5, SPINT2, SPR, SQSTM1, SRD5A2, SRD5A3, SSR4, ST3GAL5, STAMBP, STAR, STAT1, STIL, STIM1, STK4, STRA6, STRADA, STUB1, STX11, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNE4, SYP, TAFAZZIN, TALDO1, TANGO2, TAP1, TAT, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBX19, TCAP, TCIRG1, TCN2, TCTN1,* TCTN2, TCTN3, TDRD7, TECPR2, TELO2, TERT, TF, TFR2, TG, TGM1, TH, THOC2, TIMM8A, TJP2, TK2, TMC1, TMC01, TMEM107, TMEM126A, TMEM138, TMEM165, TMEM216, TMEM237, TMEM38B, TMEM67, TMEM70, TMPRSS3, TMTC3, TNFRSF11A,* TNFRSF11B, TNFSF11, TNNT1, TOE1, TPI1, TPK1, TPM3, TPO, TPP1, TRAPPC11, TRAPPC9, TRDN,* TREX1, TRHR, TRIM32, TRIM37, TRIP11, TRIT1, TRMT10A, TRMU, TRNT1,* TRPM6, TSEN2, TSEN34, TSEN54, TSFM,* TSHB, TSHR, TTC19, TTC21B, TTC37, TTC7A, TTC8, TTI2, TTN,* TTPA, TUBGCP4, TUBGCP6, TUFM, TULP1, TUSC3, TWNK, TXNL4A, TYK2, TYMP, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3B, UBR1,* UFM1,* UGT1A1, UMPS, UNC13D, UNC80, UPF3B, UROS, USB1, USH1C, USH1G, USH2A, USP9X, VAC14, VARS1, VARS2, VDR, VIPAS39, VKORC1, VLDLR, VMA21, VPS11, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WARS2, WAS, WDR19, WDR35,* WDR45B, WDR62, WDR73, WDR81, WFS1, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT7A, WRAP53, WRN, WWOX, XIAP, XPA, XPC, XRCC4, XYLT1,* XYLT2, YARS2, ZAP70, ZBTB24, ZC4H2, ZDHHC9, ZFYVE26, ZIC3, ZMPSTE24, ZMYND10, ZNF335, ZNF469, ZNF711, ZNHIT3

* The ability to detect mutations in these genes may be reduced compared to other genes

Personal Genetic Screen (v1.2)

Genes (84):

ABCD1, ACTA2, ACTC1, ACVRL1, APC, APOB, ATP7B, BAG3, BMPR1A, BRCA1, BRCA2, BTBD, CACNA1S, CALM1, CALM2, CALM3, CASQ2, COL3A1, CYP27A1, DES, DSC2, DSG2, DSP, ENG, FBN1, FLNC, GAA, GLA, HFE (C282Y homozygotes only), HNF1A, KCNH2, KCNQ1, LDLR, LMNA, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PALB2, PCSK9, PKP2,* PLN, PMS2,* PRKAG2,* PTEN, RB1,* RBM20, RET, RPE65, RYR1,* RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFB1,* TGFB2, TMEM127, TMEM43, TNNC1, TNNI3, TNNT2, TP53, TPM1, TRDN, TSC1, TSC2, TTN (truncating variants only),* TTR, VHL, WT1

* The ability to detect mutations in these genes may be reduced compared to other genes

Chromosome Analysis

Targeted susceptibility copy number variants (v1.0):

1q21.1 deletion (ISCA-37421), 2p16.3 deletion (includes NRXN1), 3q29 deletion (ISCA-37443), 6q24 duplication (ISCA-37442), 15q11.2q13 duplication (BP1-BP3, BP2-BP3; ISCA-37404, ISCA-37478), 15q13.3 deletion (ISCA-37411), 16p11.2 deletion (proximal, distal; ISCA-37400, ISCA-37486), 17p12 deletion/duplication (ISCA-37436), 17q12 duplication (ISCA-37432), 22q11.2 duplication (proximal A-B, proximal A-D; ISCA-37433, ISCA-37446).

Copy-number variant analysis is also performed across the genome for genes included in the Basic Carrier Screen, Expanded Carrier Screen, and Personal Genetic Screen (when these tests are requested), as well as for any other copy number variants that are 1 Mb or larger and assessed to be pathogenic or likely pathogenic.