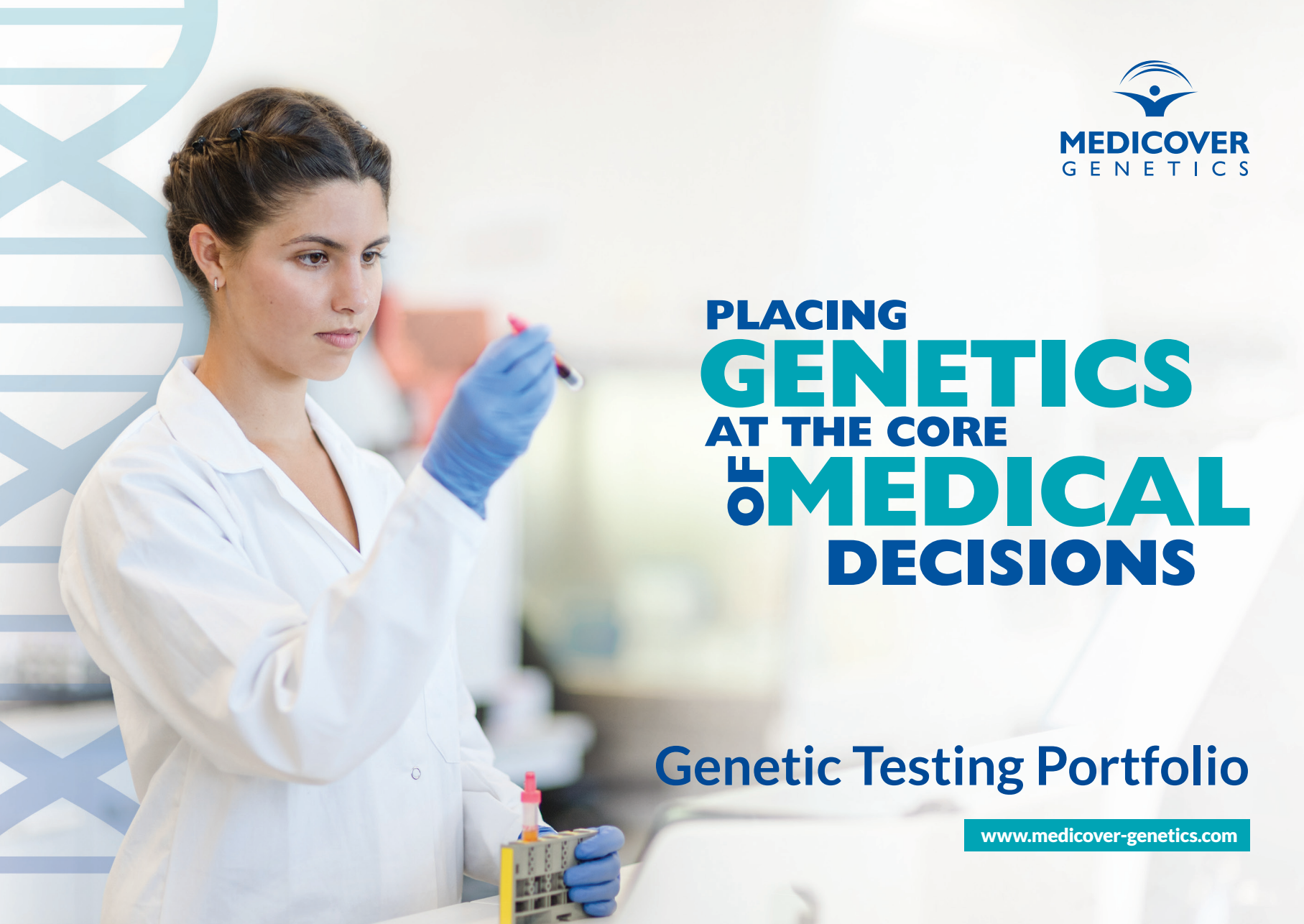

MEDICOVER GENETICS

PLACING GENETICS AT THE CORE OF MEDICAL DECISIONS

Genetic Testing Portfolio

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BLOOD DISORDERS

Disease/Indication	No. of genes	Gene/s
Alpha thalassemia	2 genes	*HBA1*, *HBA2*
Antithrombin III deficiency	1 gene	*SERPINC1*
Beta thalassemia	1 gene	*HBB*
Bleeding disorders	8 genes	*F2*, *F5*, *F7*, *F8*, *F9*, *F13A1*, *F13B*, *VWF*
Factor V deficiency	1 gene	*F5*
Factor V Leiden	1 gene	*F5*
Factor VII deficiency	1 gene	*F7*
Factor XIII deficiency	2 genes	*F13A1*, *F13B*
Hemophilia A	1 gene	*F8*
Hemophilia B	1 gene	*F9*
Protein C deficiency	1 gene	*PROC*
Protein S deficiency	1 gene	*PROS1*
Prothrombin deficiency	1 gene	*F2*
Prothrombin G20210A	1 gene	*F2*
Sickle cell disease	1 gene	*HBB*
Sideroblastic Anemia	1 gene	*ALAS2*
Spherocytosis, hereditary	5 genes	*ANK1*, *EPB42*, *SLC4A1*, *SPTA1*, *SPTB*
Telangiectasia, hereditary hemorrhagic	4 genes	*ACVRL1*, *ENG*, *GDF2*, *SMAD4*
Thrombophilia	5 genes	*F2*, *F5*, *PROC*, *PROS1*, *SERPINC1*
von Willebrand disease	1 gene	*VWF*

CARDIOVASCULAR DISORDERS

Disease/Indication	No. of genes	Gene/s
Alagille syndrome panel	2 genes	*JAG1*, *NOTCH2*
Aortic disorders	79 genes	*ACTA2*, *ADAMTS10*, *ADAMTS17*, *ADAMTS2*, *ADAMTSL2*, *ADAMTSL4*, *AEBP1*, *ALDH18A1*, *ATP6V0A2*, *ATP6V1A*, *ATP6V1E1*, *B3GALT6*, *B4GALT7*, *BGN*, *C1R*, *C1S*, *CHD2*, *CHST14*, *CNOT3*, *COL12A1*, *COL1A1*, *COL1A2*, *COL3A1*, *COL4A1*, *COL4A2*, *COL4A5*, *COL5A1*, *COL5A2*, *COL6A1*, *COL6A2*, *COL6A3*, *DSE*, *EFEMP1*, *EFEMP2*, *ELN*, *EMILIN1*, *FBLN5*, *FBN1*, *FBN2*, *FKBP14*, *FLNA*, *FOXE3*, *GATA5*, *GUCY1A1*, *LOX*, *LTBP2*, *LTBP3*, *LTBP4*, *MAT2A*, *MED12*, *MFAP5*, *MYH11*, *MYLK*, *NOTCH1*, *PHYKPL*, *PIEZO2*, *PLOD1*, *PLOD3*, *PRDM5*, *PRKG1*, *PYCR1*, *RNF213*, *ROBO4*, *SETD5*, *SKI*, *SLC2A10*, *SLC39A13*, *SMAD2*, *SMAD3*, *SMAD4*, *SMAD6*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*, *TNXB*, *UPF3B*, *ZDHHC9*, *ZNF469*
Arrhythmia	65 genes	*ABCC9*, *AKAP9*, *ANK2*, *CACNA1C*, *CACNA1D*, *CACNA2D1*, *CACNB2*, *CALM1*, *CALM2*, *CALM3*, *CASQ2*, *CAV3*, *DES*, *DMD*, *DMPK*, *DSP*, *EMD*, *GATA4*, *GJA5*, *GJC1*, *GLA*, *GNB2*, *GNB5*, *GPD1L*, *HCN1*, *HCN2*, *HCN4*, *KCNA5*, *KCND3*, *KCNE1*, *KCNE2*, *KCNE3*, *KCNE5*, *KCNH2*, *KCNJ2*, *KCNJ5*, *KCNJ8*, *KCNQ1*, *LAMP2*, *LMNA*, *MYH6*, *MYL4*, *NKX2-5*, *NPPA*, *PKP2*, *PRKAG2*, *RANGRF*, *RYR2*, *SCN10A*, *SCN1B*, *SCN2B*, *SCN3B*, *SCN4B*, *SCN5A*, *SGO1*, *SLC22A5*, *SLC4A3*, *SLC8A1*, *SNTA1*, *TBX5*, *TECRL*, *TNNI3K*, *TRDN*, *TRPM4*, *TTN*
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	10 genes	*DES*, *DSC2*, *DSG2*, *DSP*, *JUP*, *LMNA*, *PKP2*, *PLN*, *TGFB3*, *TMEM43*
Arrhytmia and cardiomyopathy	138 genes	*ABCC9*, *ACADVL*, *ACTC1*, *ACTN2*, *AGL*, *AKAP9*, *ALMS1*, *ALPK3*, *ANK2*, *ANKRD1*, *BAG3*, *BRAF*, *CACNA1C*, *CACNA1D*, *CACNA2D1*, *CACNB2*, *CALM1*, *CALM2*, *CALM3*, *CALR3*, *CASQ2*, *CAV3*, *CBL*, *COX15*, *CPT2*, *CRYAB*, *CSRP3*, *CTF1*, *DES*, *DMD*, *DMPK*, *DOLK*, *DSC2*, *DSG2*, *DSP*, *ELAC2*, *EMD*, *FHL1*, *FHL2*, *FHOD3*, *FKTN*, *FLNC*, *FXN*, *GAA*, *GATA4*, *GATAD1*, *GJA5*, *GJC1*, *GLA*, *GNB2*, *GNB5*, *GPD1L*, *HCN1*, *HCN2*, *HCN4*, *HRAS*, *ILK*, *JPH2*, *JUP*, *KCNA5*, *KCND3*, *KCNE1*, *KCNE2*, *KCNE3*, *KCNE5*, *KCNH2*, *KCNJ2*, *KCNJ5*, *KCNJ8*, *KCNQ1*, *KRAS*, *LAMA4*, *LAMP2*, *LDB3*, *LMNA*, *MAP2K1*, *MAP2K2*, *MIB1*, *MTO1*, *MYBPC3*, *MYH6*, *MYH7*, *MYL2*, *MYL3*, *MYL4*, *MYLK2*, *MYO6*, *MYOM1*, *MYOZ2*, *MYPN*, *NEBL*, *NEXN*, *NF1*, *NKX2-5*, *NPPA*, *PKP2*, *PLN*, *PRDM16*, *PRKAG2*, *PTPN11*, *RAF1*, *RANGRF*, *RASA1*, *RBM20*, *RIT1*, *RYR2*, *SCN10A*, *SCN1B*, *SCN2B*, *SCN3B*, *SCN4B*, *SCN5A*, *SGCD*, *SGO1*, *SHOC2*, *SLC22A5*, *SLC25A4*, *SLC4A3*, *SLC8A1*, *SNTA1*, *SOS1*, *SPRED1*, *TAZ*, *TBX5*, *TCAP*, *TECRL*, *TGFB3*, *TMEM43*, *TNNC1*, *TNNI3*, *TNNI3K*, *TNNT2*, *TPM1*, *TRDN*, *TRPM4*, *TTN*, *TTR*, *VCL*

Disease/Indication	No. of genes	Gene/s
Atrial fibrillation recommended*	11 genes	*GJA5, GJC1, KCNA5, KCNH2, KCNQ1, LMNA, MYL4, NPPA, SCN5A, TBX5, TTN*
Bicuspid aortic valve with risk of aortic valve stenosis and dilatation	4 genes	*GATA5, NOTCH1, ROBO4, SMAD6*
Brugada syndrome (BrS)	19 genes	*ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, TRPM4*
Brugada syndrome (BrS) recommended*	1 gene	*SCN5A*
Cardiac conduction disease (CCD) recommended*	18 genes	*DES, DMD, DMPK, DSP, EMD, GATA4, GJA5, GJC1, GLA, LAMP2, LMNA, MYL4, NKX2-5, PRKAG2, SCN5A, TBX5, TNNI3K, TRPM4*
Cardiomyopathy	92 genes	*ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COX15, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, FHL1, FHL2, FHOD3, FKTN, FLNC, FXN, GAA, GATAD1, GLA, HCN4, HRAS, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MIB1, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SLC25A4, SOS1, SPRED1, TAZ, TCAP, TGFB3, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL*
Catecholaminergic polymorphic ventricular tachycardia (CPVT) recommended*	8 genes	*CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN*
Congenital heart defect	95 genes	*ACTC1, ACVR2B, ADAMTS10, ARHGAP31, BMPR2, BRAF, CBL, CFAP53, CHD7, CITED2, CREBBP, CRELD1, DNAH11, DNAH5, DNAI1, DOCK6, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HRAS, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, LZTR1, MAP2K1, MAP2K2, MED12, MED13L, MGP, MMP21, MRAS, MYH11, MYH6, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NPHP4, NR2F2, NRAS, NSD1, PITX2, PKD1L1, PPP1CB, PTPN11, RAF1, RASA2, RBM10, RBPJ, RIT1, RRAS, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMAD4, SMAD6, SOS1, SOS2, SPRED1, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGFBR1, TGFBR2, TLL1, ZEB2, ZFPM2, ZIC3*

Disease/Indication	No. of genes	Gene/s
Dilated cardiomyopathy (DCM)	59 genes	*ABCC9, ACADVL, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, CALR3, CASQ2, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FKTN, FLNC, GATAD1, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, PKP2, PLN, PRDM16, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SGCD, TAZ, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL*
Dilated cardiomyopathy (DCM) recommended*	20 genes	*ACTC1, ACTN2, BAG3, DES, DSP, FLNC, JPH2, LMNA, MYBPC3, MYH7, NEXN, PLN, RBM20, SCN5A, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL*
Early repolarization syndrome recommended*	8 genes	*ABCC9, CACNA1C, CACNA2D1, CACNB2, KCND3, KCNJ8, SCN10A, SCN5A*
Ehlers-Danlos syndrome	29 genes	*ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, EMILIN1, FKBP14, FLNA, PHYKPL, PIEZO2, PLOD1, PLOD3, PRDM5, SLC2A10, SLC39A13, TNXB, ZNF469*
Heterotaxy (see Cilioptahies tab)	13 genes	*ACVR2B, CFAP53, CRELD1, DNAH11, DNAH5, DNAI1, GDF1, LEFTY2, MMP21, NODAL, NPHP4, PKD1L1, ZIC3*
Hypertrophic cardiomyopathy (HCM)	66 genes	*ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COX15, CPT2, CRYAB, CSRP3, CTF1, DES, ELAC2, FHL1, FHL2, FHOD3, FLNC, FXN, GAA, GLA, HRAS, JPH2, KRAS, LAMP2, LDB3, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEXN, NF1, PLN, PRKAG2, PTPN11, RAF1, RASA1, RIT1, SHOC2, SLC25A4, SOS1, SPRED1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL*
Hypertrophic cardiomyopathy (HCM) recommended*	34 genes	*ABCC9, ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CAV3, CRYAB, CSRP3, DES, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH7, MYL2, MYL3, MYO6, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, TNNC1, TNNI3, TNNT2, TPM1, TTR*
Isolated and syndromic heart defect panel	61 genes	*ACTC1, ADAMTS10, ARHGAP31, BMPR2, CHD7, CITED2, CREBBP, DOCK6, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GJA1, GPC3, JAG1, KDM6A, KMT2D, MED12, MED13L, MGP, MYH11, MYH6, NIPBL, NKX2-5, NKX2-6, NOTCH1, NOTCH2, NR2F2, NSD1, PITX2, RBM10, RBPJ, SALL1, SALL4, SEMA3E, SMAD4, SMAD6, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGFBR1, TGFBR2, TLL1, ZEB2, ZFPM2*
Loeys-Dietz syndrome	6 genes	*SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*

Disease/Indication	No. of genes	Gene/s
Long QT syndrome (LQTS)	19 genes	*AKAP9*, *ANK2*, *CACNA1C*, *CALM1*, *CALM2*, *CALM3*, *CAV3*, *KCNE1*, *KCNE2*, *KCNE3*, *KCNH2*, *KCNJ2*, *KCNJ5*, *KCNQ1*, *SCN4B*, *SCN5A*, *SNTA1*, *TECRL*, *TRDN*
Long QT syndrome (LQTS) recommended*	11 genes	*CACNA1C*, *CALM1*, *CALM2*, *CALM3*, *KCNE1*, *KCNE2*, *KCNH2*, *KCNJ2*, *KCNQ1*, *SCN5A*, *TRDN*
Marfan syndrome	3 genes	*FBN1*, *TGFBR1*, *TGFBR2*
Non-compaction cardiomyopathy (NCCM)	16 genes	*ACTC1*, *HCN4*, *LDB3*, *MIB1*, *MYBPC3*, *MYH7*, *MYL2*, *PRDM16*, *RBM20*, *RYR2*, *TAZ*, *TNNC1*, *TNNI3*, *TNNT2*, *TPM1*, *TTN*
RASopathies (see corresponding tab)	21 genes	*BRAF*, *CBL*, *HRAS*, *KRAS*, *LZTR1*, *MAP2K1*, *MAP2K2*, *MRAS*, *NF1*, *NRAS*, *PPP1CB*, *PTPN11*, *RAF1*, *RASA2*, *RIT1*, *RRAS*, *RRAS2*, *SHOC2*, *SOS1*, *SOS2*, *SPRED1*
Restrictive cardiomyopathy (RCM)	10 genes	*DES*, *FLNC*, *LMNA*, *MYBPC3*, *MYH7*, *MYL2*, *TNNI3*, *TNNT2*, *TPM1*, *TTN*
Short-QT syndrome (SQTS)	10 genes	*CACNA1C*, *CACNA2D1*, *CACNB2*, *GJA5*, *KCNH2*, *KCNJ2*, *KCNQ1*, *SCN5A*, *SLC22A5*, *SLC4A3*
Short-QT syndrome (SQTS) recommended*	4 genes	*KCNH2*, *KCNJ2*, *KCNQ1*, *SLC4A3*
Sinus node disease recommended*	16 genes	*CACNA1D*, *CASQ2*, *EMD*, *GNB2*, *GNB5*, *HCN1*, *HCN2*, *HCN4*, *KCNJ5*, *KCNQ1*, *LMNA*, *MYH6*, *RYR2*, *SCN5A*, *SGO1*, *SLC8A1*
Thoracic aortic aneurysm and dissection (TAAD)	36 genes	*ACTA2*, *BGN*, *COL1A1*, *COL3A1*, *COL4A5*, *COL5A1*, *COL5A2*, *EFEMP2*, *ELN*, *EMILIN1*, *FBLN5*, *FBN1*, *FBN2*, *FLNA*, *FOXE3*, *GATA5*, *LOX*, *LTBP3*, *MAT2A*, *MFAP5*, *MYH11*, *MYLK*, *NOTCH1*, *PLOD1*, *PRKG1*, *ROBO4*, *SKI*, *SLC2A10*, *SMAD2*, *SMAD3*, *SMAD4*, *SMAD6*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*

*recommended genes according to European Heart Rhythm Association (EHRA)/ Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases, Wilde et al. 2022, Europace. 24:1307

CILIOPATHIES

Disease/Indication	No. of genes	Gene/s
Bardet-Biedl syndrome	26 genes	*ALMS1*, *ARL6*, *BBIP1*, *BBS1*, *BBS10*, *BBS12*, *BBS2*, *BBS4*, *BBS5*, *BBS7*, *BBS9*, *C8orf37*, *CCDC28B*, *CEP290*, *IFT172*, *IFT27*, *IFT74*, *LZTFL1*, *MKKS*, *MKS1*, *NPHP1*, *SDCCAG8*, *TMEM67*, *TRIM32*, *TTC8*, *WDPCP*
Ciliary dyskinesia, primary	36 genes	*ARMC4*, *CCDC103*, *CCDC114*, *CCDC151*, *CCDC39*, *CCDC40*, *CCDC65*, *CCNO*, *CENPF*, *CFAP298*, *DNAAF1*, *DNAAF2*, *DNAAF3*, *DNAAF4*, *DNAAF5*, *DNAH1*, *DNAH11*, *DNAH5*, *DNAH8*, *DNAI1*, *DNAI2*, *DNAJB13*, *DNAL1*, *DRC1*, *GAS8*, *LRRC6*, *MCIDAS*, *NME8*, *PIH1D3*, *RSPH1*, *RSPH3*, *RSPH4A*, *RSPH9*, *SPAG1*, *TTC25*, *ZMYND10*
Ciliopathies	128 genes	*ACVR2B*, *AHI1*, *ALMS1*, *ARL13B*, *ARL6*, *ARMC4*, *ATXN10*, *B9D1*, *B9D2*, *BBIP1*, *BBS1*, *BBS10*, *BBS12*, *BBS2*, *BBS4*, *BBS5*, *BBS7*, *BBS9*, *C2CD3*, *C8orf37*, *CC2D2A*, *CCDC103*, *CCDC114*, *CCDC151*, *CCDC28B*, *CCDC39*, *CCDC40*, *CCDC65*, *CCNO*, *CENPF*, *CEP104*, *CEP120*, *CEP290*, *CEP41*, *CFAP298*, *CFAP53*, *CPLANE1*, *CRELD1*, *CSPP1*, *DDX59*, *DNAAF1*, *DNAAF2*, *DNAAF3*, *DNAAF4*, *DNAAF5*, *DNAH1*, *DNAH11*, *DNAH5*, *DNAH8*, *DNAI1*, *DNAI2*, *DNAJB13*, *DNAL1*, *DRC1*, *DYNC2H1*, *DYNC2LI1*, *EVC*, *EVC2*, *GAS8*, *GDF1*, *HYLS1*, *IFT122*, *IFT140*, *IFT172*, *IFT27*, *IFT43*, *IFT52*, *IFT57*, *IFT74*, *IFT80*, *INPP5E*, *INTU*, *INVS*, *IQCB1*, *KIAA0556*, *KIAA0586*, *KIAA0753*, *KIF14*, *KIF7*, *LEFTY2*, *LRRC6*, *LZTFL1*, *MCIDAS*, *MKKS*, *MKS1*, *MMP21*, *NEK1*, *NME8*, *NODAL*, *NPHP1*, *NPHP3*, *NPHP4*, *OFD1*, *PDE6D*, *PIH1D3*, *PKD1L1*, *POC1B*, *RPGRIP1L*, *RSPH1*, *RSPH3*, *RSPH4A*, *RSPH9*, *SCLT1*, *SDCCAG8*, *SPAG1*, *TBC1D32*, *TCTN1*, *TCTN2*, *TCTN3*, *TMEM107*, *TMEM138*, *TMEM216*, *TMEM231*, *TMEM237*, *TMEM67*, *TRAF3IP1*, *TRIM32*, *TTC21B*, *TTC25*, *TTC8*, *WDPCP*, *WDR19*, *WDR34*, *WDR35*, *WDR60*, *ZIC3*, *ZMYND10*, *ZNF423*
Heterotaxy	13 genes	*ACVR2B*, *CFAP53*, *CRELD1*, *DNAH11*, *DNAH5*, *DNAI1*, *GDF1*, *LEFTY2*, *MMP21*, *NODAL*, *NPHP4*, *PKD1L1*, *ZIC3*
Joubert syndrome	31 genes	*AHI1*, *ARL13B*, *ATXN10*, *B9D1*, *CC2D2A*, *CEP104*, *CEP290*, *CEP41*, *CPLANE1*, *CSPP1*, *HYLS1*, *INPP5E*, *KIAA0556*, *KIAA0586*, *KIF7*, *MKS1*, *NPHP1*, *OFD1*, *PDE6D*, *POC1B*, *RPGRIP1L*, *TCTN1*, *TCTN2*, *TCTN3*, *TMEM138*, *TMEM216*, *TMEM231*, *TMEM237*, *TMEM67*, *TTC21B*, *ZNF423*
Meckel-Gruber syndrome	19 genes	*B9D1*, *B9D2*, *CC2D2A*, *CEP290*, *CEP41*, *CPLANE1*, *CSPP1*, *KIF14*, *MKS1*, *NPHP3*, *RPGRIP1L*, *TCTN2*, *TCTN3*, *TMEM138*, *TMEM216*, *TMEM231*, *TMEM237*, *TMEM67*, *TTC21B*

Disease/Indication	No. of genes	Gene/s
Orofaciodigital syndrome	16 genes	*C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, SCLT1, TBC1D32, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, WDPCP*
Senior-Loken syndrome	9 genes	*CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19*
Short-rib thoracic dysplasia with or without polydactyly	20 genes	*CEP120, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60*

DISEASES OF THE CONNECTIVE TISSUE

Disease/Indication	No. of genes	Gene/s
Achondrogenesis type 2	1 gene	*COL2A1*
Achondroplasia	1 gene	*FGFR3*
Aortic disorders	79 genes	*ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALT6, B4GALT7, BGN, C1R, C1S, CHD2, CHST14, CNOT3, COL12A1, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL4A5, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, EFEMP1, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GATA5, GUCY1A1, LOX, LTBP2, LTBP3, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PHYKPL, PIEZO2, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RNF213, ROBO4, SETD5, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, UPF3B, ZDHHC9, ZNF469*
Apert syndrome	1 gene	*FGFR2*
Bicuspid aortic valve with risk of aortic valve stenosis and dilatation	4 genes	*GATA5, NOTCH1, ROBO4, SMAD6*
Contractural arachnodactyly, congenital	1 gene	*FBN2*
Craniosynostosis	28 genes	*ALPL, ALX4, CDC45, COLEC11, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT140, IFT43, IHH, IL11RA, IMPAD1, MYH3, P4HB, POR, RAB23, RECQL4, RUNX2, SCARF2, SEC24D, SMAD6, TCF12, TWIST1*

Disease/Indication	No. of genes	Gene/s
Crouzon syndrome	2 genes	*FGFR2*, *FGFR3*
Cutis laxa	9 genes	*ALDH18A1*, *ATP6V0A2*, *ATP6V1A*, *ATP6V1E1*, *EFEMP2*, *ELN*, *FBLN5*, *LTBP4*, *PYCR1*
Ectopia lentis	3 genes	*ADAMTSL4*, *FBN1*, *LTBP2*
Ehlers-Danlos syndrome	29 genes	*ADAMTS2*, *AEBP1*, *B3GALT6*, *B4GALT7*, *C1R*, *C1S*, *CHST14*, *COL12A1*, *COL1A1*, *COL1A2*, *COL3A1*, *COL5A1*, *COL5A2*, *COL6A1*, *COL6A2*, *COL6A3*, *DSE*, *EMILIN1*, *FKBP14*, *FLNA*, *PHYKPL*, *PIEZO2*, *PLOD1*, *PLOD3*, *PRDM5*, *SLC2A10*, *SLC39A13*, *TNXB*, *ZNF469*
Geleophysic dysplasia	3 genes	*ADAMTSL2*, *FBN1*, *LTBP3*
Hemorrhage, intracerebral	2 genes	*COL4A1*, *COL4A2*
Hypochondrogenesis	1 gene	*COL2A1*
Hypochondroplasia	1 gene	*FGFR3*
Kniest syndrome	1 gene	*COL2A1*
Langer mesomelic dysplasia/Léri-Weill dyschondrosteosis	1 gene	*SHOX*
Loeys-Dietz syndrome	6 genes	*SMAD2*, *SMAD3*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*
Lujan-Fryns syndrome	3 genes	*MED12*, *UPF3B*, *ZDHHC9*
Marfan syndrome	3 genes	*FBN1*, *TGFBR1*, *TGFBR2*
Marfan like disorders	13 genes	*ADAMTS10*, *ADAMTS17*, *ADAMTSL2*, *ADAMTSL4*, *EFEMP1*, *FBN1*, *FBN2*, *LTBP2*, *LTBP3*, *MED12*, *SKI*, *UPF3B*, *ZDHHC9*
Marshall syndrome	1 gene	*COL11A1*
Meester-Loeys syndrome	1 gene	*BGN*
Metaphyseal chondrodysplasia, Schmid type	1 gene	*COL10A1*
Moyamoya disease	6 genes	*ACTA2*, *CHD2*, *CNOT3*, *GUCY1A1*, *RNF213*, *SETD5*
Muenke syndrome	1 gene	*FGFR3*
Osteogenesis imperfecta	20 genes	*ALPL*, *BMP1*, *COL1A1*, *COL1A2*, *CREB3L1*, *CRTAP*, *FKBP10*, *IFITM5*, *MESD*, *P3H1*, *P4HB*, *PLOD2*, *PLS3*, *PPIB*, *SEC24D*, *SERPINF1*, *SERPINH1*, *SP7*, *TMEM38B*, *WNT1*

Disease/Indication	No. of genes	Gene/s
Otospondylomegaepiphyseal dysplasia (OSMED)	1 gene	*COL11A2*
Pfeiffer syndrome	2 genes	*FGFR1*, *FGFR2*
Pseudoxanthoma elasticum	1 gene	*ABCC6*
Robinow syndrome	5 genes	*DVL1*, *DVL3*, *NXN*, *ROR2*, *WNT5A*
Saethre-Chotzen syndrome	1 gene	*TWIST1*
Sceletal Disorders	77 genes	*ABCC6*, *ALPL*, *ALX4*, *BMP1*, *CDC45*, *CEP120*, *COL10A1*, *COL11A1*, *COL11A2*, *COL1A1*, *COL1A2*, *COL2A1*, *COL9A1*, *COL9A2*, *COLEC11*, *CREB3L1*, *CRTAP*, *CSPP1*, *DVL1*, *DVL3*, *DYNC2H1*, *DYNC2LI1*, *EFNB1*, *ERF*, *ESCO2*, *EVC*, *EVC2*, *FBN2*, *FGFR1*, *FGFR2*, *FGFR3*, *FKBP10*, *GLI3*, *IFITM5*, *IFT122*, *IFT140*, *IFT172*, *IFT43*, *IFT52*, *IFT80*, *IHH*, *IL11RA*, *IMPAD1*, *KIAA0586*, *MESD*, *MYH3*, *NEK1*, *NXN*, *P3H1*, *P4HB*, *PLOD2*, *PLS3*, *POR*, *PPIB*, *RAB23*, *RECQL4*, *ROR2*, *RUNX2*, *SCARF2*, *SEC24D*, *SERPINF1*, *SERPINH1*, *SHOX*, *SLC26A2*, *SMAD6*, *SP7*, *TCF12*, *TCTN3*, *TMEM38B*, *TTC21B*, *TWIST1*, *WDR19*, *WDR34*, *WDR35*, *WDR60*, *WNT1*, *WNT5A*
Short-rib thoracic dysplasia with or without polydactyly	20 genes	*CEP120*, *CSPP1*, *DYNC2H1*, *DYNC2LI1*, *EVC*, *EVC2*, *IFT122*, *IFT140*, *IFT172*, *IFT43*, *IFT52*, *IFT80*, *KIAA0586*, *NEK1*, *TCTN3*, *TTC21B*, *WDR19*, *WDR34*, *WDR35*, *WDR60*
Shprintzen-Goldberg syndrome	1 gene	*SKI*
Spondyloepiphyseal dysplasia	1 gene	*COL2A1*
Stickler syndrome	5 genes	*COL11A1*, *COL11A2*, *COL2A1*, *COL9A1*, *COL9A2*
Thanatophoric dysplasia	1 gene	*FGFR3*
Thoracic aortic aneurysm and dissection (TAAD)	36 genes	*ACTA2*, *BGN*, *COL1A1*, *COL3A1*, *COL4A5*, *COL5A1*, *COL5A2*, *EFEMP2*, *ELN*, *EMILIN1*, *FBLN5*, *FBN1*, *FBN2*, *FLNA*, *FOXE3*, *GATA5*, *LOX*, *LTBP3*, *MAT2A*, *MFAP5*, *MYH11*, *MYLK*, *NOTCH1*, *PLOD1*, *PRKG1*, *ROBO4*, *SKI*, *SLC2A10*, *SMAD2*, *SMAD3*, *SMAD4*, *SMAD6*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*
Type 1 fibrillinopathies	1 gene	*FBN1*
Weill-Marchesani syndrome	4 genes	*ADAMTS10*, *ADAMTS17*, *FBN1*, *LTBP2*

ENDOCRINE DISORDERS

Disease/Indication	No. of genes	Gene/s
AIP-associated hereditary isolated pituitary adenoma	1 gene	*AIP*
Alström syndrome	1 gene	*ALMS1*
Androgen Insensitivity Syndrome	1 gene	*AR*
Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED)	1 gene	*AIRE*
Azoospermia - step 1	-	AZF region
Azoospermia - step 2	6 genes	*AR, DMRT1, M1AP, NR5A1, TEX11, TEX14*
Bardet-Biedl syndrome	26 genes	*ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CCDC28B, CEP290, IFT172, IFT27, IFT74, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP*
Chylomicronemia syndrome / Primary hypertriglyceridaemia	5 genes	*APOA5, APOC2, GPIHBP1, LMF1, LPL*
Congenital adrenal hyperplasia (CAH)	5 genes	*CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2*
Congenital bilateral absence of the vas deferens (CBAVD)	1 gene	*CFTR*
Disorders of sex development	16 genes	*AR, DHH, DMRT1, HSD17B3, HSD3B2, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX9, SRD5A2, SRY, TSPYL1, WNT4, WT1, WWOX*
Familial hypocalciuric hypercalcemia (FHH)	3 genes	*AP2S1, CASR, GNA11*
Familial isolated hypoparathyroidism	4 genes	*CASR, GCM2, GNA11, PTH*
Gitelman Syndrome	1 gene	*SLC12A3*
Hypercholesterolemia, familial	4 genes	*APOB, LDLR, LDLRAP1, PCSK9*
Hyperlipidaemia / hyperlipoproteinaemia	13 genes	*APOA1, APOA5, APOB, APOC2, APOE, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LMF1, LPL, PCSK9*

Disease/Indication	No. of genes	Gene/s
Hyperlipoproteinemia, mixed	3 genes	*APOA1*, *APOE*, *LIPC*
Hyperparathyroidism-Pineal Tumor Syndrome	1 gene	*CDC73*
Hypoalphalipoproteinemia	3 genes	*ABCA1*, *APOA1*, *LCAT*
Hypobetalipoproteinemia	4 genes	*ANGPTL3*, *APOB*, *MTTP*, *PCSK9*
Hypochondroplasia	1 gene	*FGFR3*
Hypogonadotropic hypogonadism / Kallmann syndrome	25 genes	*ANOS1*, *CHD7*, *DUSP6*, *FEZF1*, *FGF17*, *FGF8*, *FGFR1*, *FLRT3*, *FSHB*, *GNRH1*, *GNRHR*, *HS6ST1*, *IL17RD*, *KISS1*, *KISS1R*, *LHB*, *NSMF*, *PROK2*, *PROKR2*, *SEMA3A*, *SOX10*, *SPRY4*, *TAC3*, *TACR3*, *WDR11*
Hypolipidaemia / hypolipoproteinaemia	8 genes	*ABCA1*, *ANGPTL3*, *APOA1*, *APOB*, *LCAT*, *MTTP*, *PCSK9*, *SAR1B*
Hypophosphataemia	9 genes	*CLCN5*, *DMP1*, *ENPP1*, *FAM20C*, *FGF23*, *PHEX*, *SLC34A1*, *SLC34A3*, *SLC9A3R1*
Hypophosphatasia	1 gene	*ALPL*
Klinefelter syndrome	-	Karyotyping (see also Cytogenomics)
Langer mesomelic dysplasia / Léri-Weill dyschondrosteosis	1 gene	*SHOX*
MODY (Maturity-onset diabetes of the young)	14 genes	*ABCC8*, *APPL1*, *BLK*, *CEL*, *GCK*, *HNF1A*, *HNF1B*, *HNF4A*, *INS*, *KCNJ11*, *KLF11*, *NEUROD1*, *PAX4*, *PDX1*
Multiple endocrine neoplasia type 1 (MEN1)	1 gene	*MEN1*
Multiple endocrine neoplasia type 2A and B (MEN2)	1 gene	*RET*
Multiple endocrine neoplasia type 4 (MEN4)	1 gene	*CDKN1B*
Neonatal severe primary hyperparathyroidism (NSPHPT)	1 gene	*CASR*
Obesity, monogenic	11 genes	*KSR2*, *LEP*, *LEPR*, *MC3R*, *MC4R*, *MRAP2*, *NTRK2*, *PCSK1*, *POMC*, *SH2B1*, *SIM1*
Osteoporosis, early onset	8 genes	*ALPL*, *BMP1*, *COL1A1*, *COL1A2*, *IFITM5*, *LRP5*, *PLS3*, *WNT1*
Ovarian dysgenesis	5 genes	*BMP15*, *FSHR*, *MCM9*, *NR5A1*, *PSMC3IP*

Disease/Indication	No. of genes	Gene/s
Overgrowth syndromes	13 genes	*CDKN1C, CHD8, DIS3L2, DNMT3A, EED, EZH2, GPC3, HERC1, HIST1H1E, NFIX, NSD1, OFD1, RNF135*
Pancreatitis	6 genes	*CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*
Paraganglioma pheochromocytoma syndrome	10 genes	*MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
Pendred syndrome	1 gene	*SLC26A4*
Premature ovarian failure (POF)	19 genes	*BMP15, DIAPH2, ERCC6, ESR1, FIGLA, FMR1, FOXL2, FSHR, GDF9, INHA, LHCGR, MCM9, NOBOX, NR5A1, PSMC3IP, SOHLH1, SOHLH2, STAG3, SYCE1*
Pseudohypoparathyroidism	1 gene	*GNAS*
Ullrich-Turner syndrome	-	Karyotyping & Fluorescence in situ hybridisation (FISH) (see also Cytogenomics)
von Hippel–Lindau syndrome	1 gene	*VHL*
Wolfram syndrome	1 gene	*WFS1*

EYE DISODERS

Disease/Indication	No. of genes	Gene/s
Alström Syndrome	1 gene	*ALMS1*
Bardet-Biedl syndrome	26 genes	*ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CCDC28B, CEP290, IFT172, IFT27, IFT74, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP*
Eye Disorders	136 genes	*ABCA4, ABHD12, ADAM9, ADGRV1, AGBL5, AIPL1, ALMS1, ARL2BP, ARL6, ARSG, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C8orf37, CA4, CCDC28B, CDH23, CDHR1, CEP250, CEP290, CEP78, CERKL, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, COL11A1, COL2A1, COL9A1, COL9A2, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FSCN2, GUCA1A, GUCA1B, GUCY2D, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IMPDH1, IMPG1, IMPG2, INVS, IQCB1, KCNV2, KERA, KIAA1549, KIZ, KLHL7, LCA5, LRAT, LRP5, LZTFL1, MAK, MERTK, MKKS, MKS1, MYO7A, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OFD1, OPA1, PAX6, PCARE, PCDH15, PDE6A, PDE6B, PDE6G, PDZD7, PEX1, PEX6, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RS1, SAG, SDCCAG8, SEMA4A, SLC7A14, SNRNP200, SPATA7, TMEM67, TOPORS, TRAF3IP1, TRIM32, TRPM1, TTC8, TUBB4B, TULP1, USH1C, USH1G, USH2A, VCAN, WDPCP, WDR19, WHRN, ZNF408, ZNF513*

Disease/Indication	No. of genes	Gene/s
Retinitis pigmentosa	96 genes	*ABCA4, ADAM9, AGBL5, AIPL1, ALMS1, ARL2BP, ARL6, BEST1, C8orf37, CA4, CDHR1, CERKL, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FSCN2, GUCA1A, GUCA1B, GUCY2D, IDH3A, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, IQCB1, KCNV2, KIAA1549, KIZ, KLHL7, LCA5, LRAT, LRP5, MAK, MERTK, MKKS, MKS1, MYO7A, NMNAT1, NR2E3, NRL, NYX, OFD1, OPA1, PAX6, PCARE, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RS1, SAG, SDCCAG8, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TRPM1, TTC8, TULP1, USH1C, USH1G, USH2A, WDR19, WHRN, ZNF408, ZNF513*
Senior-Loken syndrome	9 genes	*CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19*
Stickler syndrome	4 genes	*COL11A1, COL2A1, COL9A1, COL9A2*
Usher syndrome	18 genes	*ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, MYO7A, PCDH15, PDZD7, PEX1, PEX6, TUBB4B, USH1C, USH1G, USH2A, WHRN*
Wagner syndrome 1	1 gene	*VCAN*

FEVER SYNDROMES

Disease/Indication	No. of genes	Gene/s
Cryopyrin associated periodic syndrome (CAPS)	1 gene	*NLRP3*
Familial Mediterranean fever	1 gene	*MEFV*
Hyper-IgD syndrome	1 gene	*MVK*
Periodic fever syndromes	25 genes	*ADA2, CARD14, ELANE, IL1RN, IL36RN, LACC1, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RBCK1, RIPK1, TMEM173, TNFAIP3, TNFRSF1A, UBA1, WDR1*
Tumor necrosis factor receptor-associated periodic syndrome (TRAPS)	1 gene	*TNFRSF1A*

GASTROINTESTINAL DISORDERS

Disease/Indication	No. of genes	Gene/s
Celiac disease	2 genes	*HLA-DQA1, HLA-DQB1*
Crohn's disease	1 gene	*NOD2*
Lactose intolerance	1 gene	*LCT*

GLOBAL DEVELOPMENTAL DELAY

Disease/Indication	No. of genes	Gene/s
Autism	186 genes	*ADNP, AFF2, AHDC1, ALDH5A1, ANK3, ARID1B, ARX, ASH1L, ASPM, ATRX, AUTS2, BCAP31, BRAF, BRWD3, C12orf4, C12orf57, CACNA1C, CAMK2B, CAMK2G, CASK, CDK13, CDKL5, CDKN1C, CHAMP1, CHD7, CHD8, CIC, CIT, CLCN4, CNKSR2, CNOT3, CNTNAP2, CREBBP, CSNK2A1, CTCF, CTNNB1, DDX3X, DEAF1, DHCR7, DLG4, DNAJC12, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EIF2S3, EP300, EPB41L1, FGD1, FLNA, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, FTSJ1, GATAD2B, GCDH, GNAI1, GNB1, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, HCFC1, HCN1, HEPACAM, HIVEP2, HNRNPH2, HPRT1, HRAS, HUWE1, IL1RAPL1, IQSEC2, KANSL1, KAT6A, KDM5C, KIF14, KIF1A, KMT2A, KMT2E, KMT5B, KPTN, L1CAM, MAOA, MBD5, MBOAT7, MECP2, MED12, MED13, MED13L, MEF2C, METTL23, METTL5, MID1, MLC1, MTOR, MYT1L, NAA10, NAA15, NCAPD2, NEXMIF, NFIB, NFIX, NHS, NIPBL, NLGN3, NLGN4X, NRXN1, NSD1, NSUN2, NUS1, OPHN1, PACS1, PAFAH1B1, PCDH19, PHF6, PHF8, PIGG, PIGH, PIGV, PIGW, PNKP, POGZ, PPP2CA, PQBP1, PTCHD1, PTEN, PTPN11, RAB39B, RAD21, RLIM, RPL10, RPL5, RUSC2, SCN2A, SCN8A, SETBP1, SETD2, SETD5, SHANK2, SHANK3, SIK1, SLC35A1, SLC35C1, SLC6A8, SLC9A6, SMARCA2, SMARCB1, SMC1A, SMC3, SNX14, SON, SRCAP, STAG1, SYN1, SYNGAP1, SZT2, TAF1, TBC1D23, TBL1XR1, TBR1, TCF4, TLK2, TMLHE, TMTC3, TRAPPC9, TRIO, TRIP12, TRRAP, TSC1, TSC2, TUBA1A, TUBB2A, TUSC3, UBE2A, UBE3A, UPF3B, USP9X, VPS13B, WAC, ZDHHC9, ZEB2, ZMYND11, ZNF711*
Coffin-Siris syndrome	12 genes	*ARID1A, ARID1B, ARID2, DPF2, PHF6, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4*
Congenital disorders of glycosylation (CDGs)	43 genes	*ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, CAD, CCDC115, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3*
Cornelia de Lange syndrome	7 genes	*ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3*

Disease/Indication	No. of genes	Gene/s
Developmental disorders	546 genes	*AARS, ABCC9, ABCD1, ACSL4, ACTB, ACTG1, ADAT3, ADNP, AFF2, AHDC1, AIFM1, AKT3, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMER1, AMPD2, ANK3, ANKLE2, ANKRD11, AP1S2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASPA, ASPM, ATP6AP2, ATP7A, ATRX, AUTS2, B4GALT1, BCAP31, BCOR, BCS1L, BDNF, BRAF, BRD4, BRWD3, C12orf4, C12orf57, CA8, CACNA1C, CACNG2, CAD, CAMK2A, CAMK2B, CAMK2G, CASK, CBL, CC2D1A, CCDC115, CCDC22, CCND2, CDK13, CDK5, CDK5RAP2, CDK6, CDKL5, CDKN1C, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP85L, CHAMP1, CHD4, CHD7, CHD8, CHMP1A, CIC, CIT, CLCN4, CLIC2, CLN8, CLP1, CLTC, CNKSR2, CNOT3, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COL4A3BP, COLGALT1, COPB2, CRADD, CRBN, CREBBP, CSNK2A1, CTCF, CTNNB1, CUL4B, DBH, DCX, DDC, DDOST, DDX3X, DEAF1, DHCR24, DHCR7, DIS3L2, DKC1, DLG3, DLG4, DNAJC12, DNM1, DNMT3A, DOCK7, DOLK, DONSON, DPAGT1, DPF2, DPM1, DPM2, DPM3, DVL1, DVL3, DYNC1H1, DYRK1A, EBP, EDC3, EED, EEF1A2, EHMT1, EIF2B5, EIF2S3, EIF3F, ELP2, EP300, EPB41L1, EXOSC3, EXOSC8, EXOSC9, EZH2, FANCB, FBXO31, FGD1, FLNA, FMN2, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, FTSJ1, GABRA1, GALT, GATAD2B, GCDH, GCH1, GDI1, GFAP, GK, GLI3, GNAI1, GNAO1, GNB1, GPAA1, GPC3, GPSM2, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, HCCS, HCFC1, HCN1, HDAC4, HDAC6, HDAC8, HEPACAM, HERC1, HIST1H1E, HIVEP2, HMGB3, HNRNPH2, HPRT1, HRAS, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IMPA1, IQSEC2, ITPA, KANSL1, KAT6A, KAT6B, KATNB1, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KDM5C, KDM6A, KIF11, KIF14, KIF1A, KIF4A, KIF7, KIRREL3, KLHL15, KLHL7, KMT2A, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, L1CAM, LAMB1, LAMP2, LAS1L, LINGO1, LINS1, LMNB1, LMNB2, LZTR1, MACF1, MAGT1, MAN1B1, MAOA, MAP11, MAP2K2, MBD5, MBOAT7, MBTPS2, MCPH1, MECP2, MED12, MED13, MED13L, MED23, MEF2C, METTL23, METTL5, MFSD2A, MGAT2, MICU1, MID1, MID2, MLC1, MOGS, MPDU1, MPI, MRAS, MSL3, MTM1, MTOR, MYT1L, NAA10, NAA15, NALCN, NCAPD2, NCAPD3, NCAPH, NDE1, NDP, NDST1, NDUFA1, NEXMIF, NFIB, NFIX, NGLY1, NHS, NIPBL, NLGN3, NLGN4X, NONO, NRAS, NRXN1, NSD1, NSDHL, NSUN2, NTNG1, NUP37, NUS1, NXF5, NXN, OCRL, OFD1, OGT, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PCBD1, PCDH11X, PCDH19, PCNT, PDHA1, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PHC1, PHF6, PHF8, PIGA, PIGB, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3R2, PLP1, PMM2, PNKP, POGZ, PORCN, PPP1CB, PPP2CA, PPP2R1A, PPP2R5D, PPT1, PQBP1, PRPS1, PRSS12, PTCH1, PTCH2, PTCHD1, PTEN, PTPN11, PTS, PURA, PUS3, QDPR, RAB39B, RAB40AL, RAC1, RAD21, RAF1, RAI1, RARS2, RASA2, RBM10, RBMX, RELN, RFT1, RHEB, RIT1, RLIM, RNF113A, RNF135, ROR2, RPL10, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS6KA3, RPS7, RRAS, RUSC2, SASS6, SATB2, SCN1A, SCN2A, SCN8A, SCN9A, SEPSECS, SET, SETBP1, SETD2, SETD5, SHANK2, SHANK3, SHOC2, SIK1, SLC12A5, SLC13A5, SLC16A2, SLC18A2, SLC25A22, SLC25A46, SLC25A5, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SLC6A17, SLC6A3, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMPD4, SMS, SNX14, SON, SOS1, SOS2, SOX11, SOX3, SOX4, SOX5, SPR, SPTAN1, SRCAP, SRD5A3, SSR4, ST3GAL3, STAG1, STAG2, STIL, STT3A, STT3B, STXBP1, SUFU, SYN1, SYNGAP1, SYP, SZT2, TAF1, TAF13, TAF2, TBC1D23, TBC1D24, TBC1D7, TBCD, TBCK, TBL1XR1, TBR1, TCF4, TECR, TH, THOC2, TIMM8A, TLK2, TMCO1, TMEM165, TMEM199, TMLHE, TMTC3, TNIK, TOE1, TPH2, TRAPPC9, TRIO, TRIP12, TRMT1, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TSR2, TTI2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUSC3, UBE2A, UBE3A, UPF3B, USP27X, USP9X, VPS13B, VPS51, VPS53, VRK1, WAC, WASHC4, WASHC5, WDFY3, WDR45, WDR62, WNT5A, WWOX, ZBTB11, ZBTB18, ZC3H14, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZMYM3, ZMYND11, ZNF335, ZNF41, ZNF674, ZNF711, ZNF81*
CHARGE syndrome	1 gene	*CHD7*
Coffin-Lowry syndrome	1 gene	*RPS6KA3*
Fragile X syndrome	1 gene	*FMR1*
Glycosylphosphatidylinositol (GPI) biosynthesis defect	21 genes	*GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY*

Disease/Indication	No. of genes	Gene/s
Hydrops fetalis	98 genes	*ACAD9, ALG1, ALG12, ALG8, ARSB, ASAH1, BRAF, CBL, CCBE1, CEP55, CFH, CHD7, CTSA, DHCR24, DHCR7, DMPK, EBP, FAT4, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GBA, GBE1, GLA, GLB1, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KLF1, KLHL40, KRAS, LBR, LIPA, LZTR1, MAP2K1, MAP2K2, MKKS, MRAS, MVK, MYH3, MYRF, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PIEZO1, PKLR, PMM2, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RPL11, RRAS, SF3B4, SHOC2, SLC17A5, SLC22A5, SMPD1, SOS1, SOS2, SPRED1, STAT3, SUMF1, SUZ12, TALDO1, TAZ, UROS, WAC, ZEB2*
Kabuki syndrome	2 genes	*KDM6A, KMT2D*
Macrocephaly	64 genes	*ABCC9, AKT3, AMER1, ASPA, BRWD3, CCDC22, CCND2, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, DVL1, DVL3, EED, EZH2, FOXP1, GFAP, GLI3, GPC3, GRIA3, HERC1, HRAS, HUWE1, KIF7, KPTN, KRAS, LZTR1, MED12, MLC1, MTOR, NDUFA1, NFIB, NFIX, NONO, NRAS, NSD1, OFD1, PIGA, PIGN, PIGT, PIGV, PIK3R2, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, RAB39B, RAF1, RHEB, RIT1, RNF135, ROR2, SETD2, SHOC2, SOS1, SUFU, TBC1D7, TMCO1, UPF3B, WASHC5, WNT5A, ZDHHC9*
MECP2 duplication syndrome	1 gene	*MECP2*
Microcephalic osteodysplastic primordial dwarfism	1 gene	*PCNT*
Microcephalies, primary, autosomal recessive	31 genes	*ANKLE2, ASPM, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CIT, COPB2, DONSON, KIF11, KIF14, KNL1, LMNB1, LMNB2, MAP11, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PCNT, PHC1, SASS6, STIL, WDFY3, WDR62, ZNF335*
Mowat-Wilson syndrome	1 gene	*ZEB2*
Neurotransmitter disorders, pediatric	12 genes	*DBH, DDC, GCH1, MAOA, PCBD1, PTS, QDPR, SLC18A2, SLC6A3, SPR, TH, TPH2*
Overgrowth syndromes	13 genes	*CDKN1C, CHD8, DIS3L2, DNMT3A, EED, EZH2, GPC3, HERC1, HIST1H1E, NFIX, NSD1, OFD1, RNF135*
Pitt-Hopkins syndrome	1 gene	*TCF4*
Rett syndrome	3 genes	*CDKL5, FOXG1, MECP2*
Rett syndrome and Rett syndrome-like disorders	22 genes	*ALDH5A1, ARX, BDNF, CDKL5, CNTNAP2, FOXG1, FOXP2, IQSEC2, KCNA2, KCNQ2, KIF1A, MECP2, MEF2C, NRXN1, NTNG1, PLP1, SCN2A, SCN8A, STXBP1, TCF4, UBE3A, ZEB2*

Disease/Indication	No. of genes	Gene/s
Robinow syndrome	5 genes	*DVL1*, *DVL3*, *NXN*, *ROR2*, *WNT5A*
Rubinstein-Taybi syndrome	2 genes	*CREBBP*, *EP300*
Sotos syndrome	1 gene	*NSD1*
Weaver syndrome	1 gene	*EZH2*

HEARING LOSS

Disease/Indication	No. of genes	Gene/s
Deafness / hearing loss	148 genes	*ABCC1*, *ABHD12*, *ACTG1*, *ADCY1*, *ADGRV1*, *AIFM1*, *ATP6V1B1*, *BDP1*, *BSND*, *CABP2*, *CACNA1D*, *CCDC50*, *CD164*, *CDC14A*, *CDH23*, *CEACAM16*, *CEP250*, *CIB2*, *CISD2*, *CLDN14*, *CLDN9*, *CLIC5*, *CLPP*, *CLRN1*, *CLRN2*, *COCH*, *COL11A1*, *COL11A2*, *COL4A6*, *COX1*, *CRYM*, *DCDC2*, *DIABLO*, *DIAPH1*, *DIAPH3*, *DMXL2*, *EDN3*, *EDNRB*, *ELMOD3*, *EPS8*, *EPS8L2*, *ERAL1*, *ESPN*, *ESRP1*, *ESRRB*, *EYA1*, *EYA4*, *FAM189A2*, *GAB1*, *GATA3*, *GIPC3*, *GJB2*, *GJB6*, *GPRASP2*, *GPSM2*, *GRAP*, *GRHL2*, *GRXCR1*, *GRXCR2*, *GSDME*, *HARS2*, *HGF*, *HOMER2*, *HSD17B4*, *ILDR1*, *KARS*, *KCNE1*, *KCNJ10*, *KCNQ1*, *KCNQ4*, *KITLG*, *LARS2*, *LHFPL5*, *LMX1A*, *LOXHD1*, *LRTOMT*, *MARVELD2*, *MET*, *MIR182*, *MIR183*, *MIR96*, *MITF*, *MPZL2*, *MSRB3*, *MYH14*, *MYH9*, *MYO15A*, *MYO3A*, *MYO6*, *MYO7A*, *NARS2*, *NLRP3*, *OSBPL2*, *OTOA*, *OTOF*, *OTOG*, *OTOGL*, *P2RX2*, *PAX3*, *PCDH15*, *PDE1C*, *PDZD7*, *PJVK*, *PLS1*, *PNPT1*, *POU3F4*, *POU4F3*, *PPIP5K2*, *PRPS1*, *PTPRQ*, *RDX*, *REST*, *RIPOR2*, *ROR1*, *S1PR2*, *SCD5*, *SERPINB6*, *SIX1*, *SLC12A2*, *SLC17A8*, *SLC26A4*, *SLC26A5*, *SLC44A4*, *SLITRK6*, *SMPX*, *SNAI2*, *SOX10*, *SPNS2*, *STRC*, *SYNE4*, *TBC1D24*, *TECTA*, *TJP2*, *TMC1*, *TMEM132E*, *TMIE*, *TMPRSS3*, *TNC*, *TPRN*, *TRIOBP*, *TRRAP*, *TWNK*, *USH1C*, *USH1G*, *USH2A*, *WBP2*, *WFS1*, *WHRN*
Deafness / hearing loss, nonsyndromic	2 genes	*GJB2*, *GJB6*
Deafness / hearing loss, syndromic	38 genes	*ABHD12*, *ADGRV1*, *AIFM1*, *ATP6V1B1*, *BSND*, *CACNA1D*, *CDH23*, *CLPP*, *CLRN1*, *DIAPH3*, *EDN3*, *EDNRB*, *ERAL1*, *EYA1*, *GATA3*, *GPSM2*, *HSD17B4*, *KCNE1*, *KCNJ10*, *KCNQ1*, *LARS2*, *MITF*, *MYH9*, *MYO7A*, *PAX3*, *PCDH15*, *PDZD7*, *SLC12A2*, *SLC26A4*, *SLITRK6*, *SNAI2*, *SOX10*, *TWNK*, *USH1C*, *USH1G*, *USH2A*, *WFS1*, *WHRN*
Pendred syndrome	1 gene	*SLC26A4*

Disease/Indication	No. of genes	Gene/s
Perrault syndrome	6 genes	*CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK*
Usher syndrome	18 genes	*ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, MYO7A, PCDH15, PDZD7, PEX1, PEX6, TUBB4B, USH1C, USH1G, USH2A, WHRN*
Waardenburg syndrome	6 genes	*EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10*
Wolfram syndrome	1 gene	*WFS1*

HEMATOLOGY

Disease/Indication	No. of genes	Gene/s
Alpha thalassemia	2 genes	*HBA1, HBA2*
Antithrombin III deficiency	1 gene	*SERPINC1*
Beta thalassemia	1 gene	*HBB*
Bleeding disorders	8 genes	*F2, F5, F7, F8, F9, F13A1, F13B, VWF*
Bone Marrow Failure Syndromes	143 genes	*ACD, AK2, ALAS2, ANKRD26, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CDAN1, CDKN2A, CEBPA, CLPB, CSF3R, CTC1, CTSC, CXCR4, DDX41, DKC1, DNAJC21, DNASE2, DTNBP1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, GLRX5, GP1BA, HAX1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2B, ITK, JAGN1, KCNN4, LAMTOR2, LYST, MAGT1, MAP2K1, MAP2K2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MYH9, MYO5A, NAF1, NBN, NF1, NHP2, NOP10, NRAS, OBFC1, PALB2, PAX5, PGM3, PMS2, POT1, PRF1, RAB27A, RAC2, RAD51C, RECQL4, RIT1, RPL11, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SEC23B, SH2D1A, SLC19A2, SLC25A38, SLC37A4, SLX4, SMARCD2, SOS1, SRP54, STX11, STXBP2, TBXAS1, TERC, TERT, THPO, TINF2, TP53, TUBB1, UBE2T, UNC13D, USB1, VPS13B, WAS, WDR1, WIPF1, WRAP53, XRCC2, ZCCHC8*
Chronic granulomatous disease	6 genes	*CYBA, CYBB, G6PD, NCF1, NCF2, NCF4*

Disease/Indication	No. of genes	Gene/s
Comprehensive Hematology Panel	246 genes	*ABCA3, ABCB7, ABCG5, ABCG8, ACD, ACTN1, ADAMTS13, AK1, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, AP3D1, ARPC1B, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CD59, CDAN1, CDKN2A, CEBPA CECR1, CLCN7, CLPB, CSF3R, CTC1, CTLA4, CTSC, CXCR4, CYB5R3, DDX41, DKC1, DNAJC21, DNASE2, DTNBP1, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPOR, ERCC4, ERCC6L2, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FLI1, FLNA, FYB, G6PC3, G6PD, GATA1, GATA2, GBA, GCLC, GFI1, GFI1B, GGCX, GINS1, GLRX5, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAVCR2, HAX1, HFE, HMOX1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KCNN4, KIF23, KLF1, LAMTOR2, LMAN1, LPIN2, LYST, MAGT1, MAP2K1, MAP2K2, MASTL, MCFD2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MTHFD1, MTR, MYH9, MYO5A, NAF1, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, NT5C3A, OBFC1, OCA2, P2RY12, PALB2, PARN, PAX5, PC, PDHA1, PDHX, PGK1, PGM3, PIEZO1, PKLR, PMS2, POT1, PRF1, PRKACG, PROC, PROS1, PTPN11, PUS1, RAB27A, RAC2, RAD51C, RASGRP2, RECQL4, REN, RHAG, RIT1, RNF168, RPL11, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCD2, SOS1, SPTA1, SPTB, SRC, SRP54, STAT3, STX11, STXBP2, TBXA2R, TBXAS1, TCN2, TERC, TERT, TF, THBD, THPO, TINF2, TMPRSS6, TP53, TPI1, TRNT1, TUBB1, UBE2T, UNC13D, USB1, VPS13B, WAS, WDR1, WIPF1, WRAP53, XRCC2, YARS2, ZCCHC8*
Factor V deficiency	1 gene	*F5*
Factor VII deficiency	1 gene	*F7*
Factor XIII deficiency	2 genes	*F13A1, F13B*
Fanconi anemia	22 genes	*BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2*
Hemophilia A	1 gene	*F8*
Hemophilia B	1 gene	*F9*
Li Fraumeni syndrome	1 gene	*TP53*
Neurofibromatosis type 1	1 gene	*NF1*

Disease/Indication	No. of genes	Gene/s
Neutropenia, congenital	26 genes	*AP3B1, CLPB, CSF3R, CXCR4, DNAJC21, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, LYST, MRTFA, RAB27A, SBDS, SLC37A4, SMARCD2, TAZ, USB1, VPS13B, VPS45, WAS, WDR1*
Neutropenia, cyclic / Neutropenia, severe congenital 1	1 gene	*ELANE*
Protein C deficiency	1 gene	*PROC*
Protein S deficiency	1 gene	*PROS1*
Prothrombin deficiency	1 gene	*F2*
Sickle cell disease	1 gene	*HBB*
Sideroblastic Anemia	1 gene	*ALAS2*
Spherocytosis, hereditary	5 genes	*ANK1, EPB42, SLC4A1, SPTA1, SPTB*
Telangiectasia, hereditary hemorrhagic	4 genes	*ACVRL1, ENG, GDF2, SMAD4*
Thrombophilia	3 genes	*F2, F5, PROC, PROS1, SERPINC1*
von Willebrand disease	1 gene	*VWF*
Wiskott-Aldrich syndrome	1 gene	*WAS*

IMMUNODEFICIENCIES

Disease/Indication	No. of genes	Gene/s
Agammaglobulinemia	8 genes	*BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A, PIK3R1*
Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED)	1 gene	*AIRE*
Chronic granulomatous disease	6 genes	*CYBA, CYBB, G6PD, NCF1, NCF2, NCF4*

Disease/Indication	No. of genes	Gene/s
Combined T- and B-cell immunodeficiencies	42 genes	*ADA, AK2, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DOCK8, FOXN1, IKZF1, IL2RG, IL7R, ITK, JAK3, LCK, LIG4, MAGT1, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, UNC119, ZAP70*
Disorders of leukocyte motility	12 genes	*ACTB, CEBPE, CTSC, DNAJC21, FERMT3, FPR1, ITGB2, MRTFA, RAC2, SBDS, SLC35C1, WDR1*
Hyper-IgM syndrome	4 genes	*AICDA, CD40, CD40LG, UNG*
Immunodeficiencies	86 genes	*ACTB, ADA, AICDA, AIRE, AK2, AP3B1, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CEBPE, CIITA, CLPB, CORO1A, CSF3R, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DNAJC21, DOCK8, ELANE, FERMT3, FOXN1, FPR1, G6PC3, G6PD, GATA1, GATA2, GFI1, HAX1, HYOU1, IKZF1, IL2RG, IL7R, ITGB2, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LYST, MAGT1, MRTFA, NCF1, NCF2, NCF4, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAB27A, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, SBDS, SH2D1A, SLC35C1, SLC37A4, SMARCD2, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TAZ, UNC119, UNG, USB1, VPS13B, VPS45, WAS, WDR1, ZAP70*
Lymphoproliferative syndrome, X-linked, 1	1 gene	*SH2D1A*
Neutropenia, congenital	26 genes	*AP3B1, CLPB, CSF3R, CXCR4, DNAJC21, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, LYST, MRTFA, RAB27A, SBDS, SLC37A4, SMARCD2, TAZ, USB1, VPS13B, VPS45, WAS, WDR1*
Neutropenia, cyclic / Neutropenia, severe congenital 1	1 gene	*ELANE*
Severe combined immunodeficiency, X-linked (X-SCID)	1 gene	*IL2RG*
Shwachman-Diamond syndrome	1 gene	*SBDS*
Wiskott-Aldrich syndrome	1 gene	*WAS*
X-linked agammaglobulinemia (XLA)	1 gene	*BTK*

LUNG DISEASES

Disease/Indication	No. of genes	Gene/s
Alpha-1 antitrypsin deficiency	1 gene	*SERPINA1*
Cystic fibrosis (CF)	1 gene	*CFTR*
Interstitial lung disease / surfactant metabolism dysfunction, pulmonary	8 genes	*ABCA3*, *CSF2RA*, *CSF2RB*, *FLNA*, *FOXF1*, *NKX2-1*, *SFTPB*, *SFTPC*
Pulmonary alveolar microlithiasis	1 gene	*SLC34A2*
Pulmonary arterial hypertension	13 genes	*ACVRL1*, *BMPR1B*, *BMPR2*, *CAV1*, *EIF2AK4*, *ENG*, *GDF2*, *KCNA5*, *KCNK3*, *SMAD1*, *SMAD4*, *SMAD9*, *TBX4*

METABOLIC DISORDERS

Disease/Indication	No. of genes	Gene/s
Abetalipoproteinemia	1 gene	*MTTP*
Alcohol intolerance	2 genes	*ADH1B*2*, *ALDH2*2 alleles*
ApoA-I deficiency	1 gene	*APOA1*
Apolipoprotein B deficiency (basic)	1 gene	*APOB p.Arg3527Gln*
Biotinidase deficiency	1 gene	*BTD*
Chylomicronemia syndrome	5 genes	*APOA5*, *APOC2*, *GPIHBP1*, *LMF1*, *LPL*
Cobalamin metabolism disorders	5 genes	*AMN*, *CUBN*, *MMAA*, *MMAB*, *MMUT*
Congenital adrenal hyperplasia	5 genes	*CYP11B1*, *CYP11B2*, *CYP17A1*, *CYP21A2*, *HSD3B2*
Congenital disorders of glycosylation (CDGs)	43 genes	*ALG1*, *ALG11*, *ALG12*, *ALG13*, *ALG2*, *ALG3*, *ALG6*, *ALG8*, *ALG9*, *B4GALT1*, *CAD*, *CCDC115*, *COG1*, *COG4*, *COG5*, *COG6*, *COG7*, *COG8*, *DDOST*, *DOLK*, *DPAGT1*, *DPM1*, *DPM2*, *DPM3*, *MGAT2*, *MOGS*, *MPDU1*, *MPI*, *NGLY1*, *PGM1*, *PMM2*, *RFT1*, *SLC35A1*, *SLC35A2*, *SLC35C1*, *SLC39A8*, *SRD5A3*, *SSR4*, *STT3A*, *STT3B*, *TMEM165*, *TMEM199*, *TUSC3*

Disease/Indication	No. of genes	Gene/s
Crigler-Najjar syndrome	1 gene	*UGT1A1*
Diarrhea 1, secretory chloride, congenital	1 gene	*SLC26A3*
Dihyropyrimidine dehydrogenase deficiency	1 gene	*DPYD*
Fabry disease	1 gene	*GLA*
Fatty acid oxidation disorders	7 genes	*ACADM, ACADVL, ETFA, ETFB, ETFDH, HADHA, HADHB*
Fish-eye disease	1 gene	*LCAT*
Fructose intolerance	1 gene	*ALDOB*
Fructose-1,6-bisphosphatase deficiency	1 gene	*FBP1*
Galactosemia	1 gene	*GALT*
Gaucher disease	1 gene	*GBA*
Gilbert syndrome	1 gene	*UGT1A1 *28-Allel*
Glucose-6-phosphate dehydrogenase deficiency (Favism)	1 gene	*G6PD*
Glutaric acidemia I (GA1)	1 gene	*GCDH*
Glycine encephalopathy	1 gene	*GLDC*
Glycogen storage disease II (Pompe disease)	1 gene	*GAA*
Hepatic lipase deficiency	1 gene	*LIPC*
Hereditary hemochromatosis - step 1	1 gene	*HFE C282Y & H63D*
Hereditary hemochromatosis - step 2	6 genes	*BMP6, HAMP, HFE, HJV, SLC40A1, TFR2*
Hypercholesterolemia, familial	4 genes	*APOB, LDLR, LDLRAP1, PCSK9*
Hyperhomocysteinemia	1 gene	*MTHFR*
Hyperlipidaemia / hyperlipoproteinaemia	13 genes	*APOA1, APOA5, APOB, APOC2, APOE, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LMF1, LPL, PCSK9*
Hyperlipoproteinemia, mixed	3 genes	*APOA1, APOE, LIPC*

Disease/Indication	No. of genes	Gene/s
Hyperlipoproteinemia, type Ib	1 gene	*APOC2*
Hyperlipoproteinemia, type III	1 gene	*APOE isoforms E2/E3/E4*
Hyperoxaluria	3 genes	*AGXT, GRHPR, HOGA1*
Hypertriglyceridemia	5 genes	*APOA5, APOC2, GPIHBP1, LMF1, LPL*
Hypoalphalipoproteinemia	3 genes	*ABCA1, APOA1, LCAT*
Hypobetalipoproteinemia	4 genes	*ANGPTL3, APOB, MTTP, PCSK9*
Hypolipidaemia / hypolipoproteinaemia	8 genes	*ABCA1, ANGPTL3, APOA1, APOB, LCAT, MTTP, PCSK9, SAR1B*
Hypophosphataemia	9 genes	*CLCN5, DMP1, ENPP1, FAM20C, FGF23, PHEX, SLC34A1, SLC34A3, SLC9A3R1*
Hypophosphatasia	1 gene	*ALPL*
Isovaleric acidemia	1 gene	*IVD*
Krabbe disease	2 genes	*GALC, PSAP*
Lactase deficiency, congenital	1 gene	*LCT*
Lactose intolerance, adult onset	1 gene	*LCT c.-13910C/T*
LCAT deficiency	1 gene	*LCAT*
Lipoprotein lipase deficiency	1 gene	*LPL*
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	2 genes	*HADHA, HADHB*
Lysinuric protein intolerance	1 gene	*SLC7A7*
Lysosomal storage disorders	22 genes	*ARSB, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, NPC1, NPC2, PSAP, SGSH, SMPD1*
Malignant hyperthermia	2 genes	*CACNA1S, RYR1*
Maple syrup urine disease (MSUD)	3 genes	*BCKDHA, BCKDHB, DBT*
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	1 gene	*ACADM*

Disease/Indication	No. of genes	Gene/s
Methylmalonic aciduria	1 gene	*MMUT*
Mevalonic aciduria	1 gene	*MVK*
Mitochondrial carnitine-acylcarnitine cycle disorders	3 genes	*CPT1A, CPT2, SLC25A20*
MODY (Maturity-onset diabetes of the young)	14 genes	*ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1*
Mucopolysaccharidosis	11 genes	*ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH*
Multiple acyl-CoA dehydrogenase deficiency (MADD)	3 genes	*ETFA, ETFB, ETFDH*
Niemann-Pick disease	3 genes	*NPC1, NPC2, SMPD1*
Obesity	11 genes	*KSR2, LEP, LEPR, MC3R, MC4R, MRAP2, NTRK2, PCSK1, POMC, SH2B1, SIM1*
Phenylketonuria	1 gene	*PAH*
Phosphoenolpyruvate carboxykinase deficiency, cytosolic	1 gene	*PCK1*
Porphyrias	8 genes	*ALAD, ALAS2, CPOX, FECH, HMBS, PPOX, UROD, UROS*
Propionicacidemia	2 genes	*PCCA, PCCB*
Smith-Lemli-Opitz syndrome	1 gene	*DHCR7*
Sphingolipidoses	10 genes	*GALC, GBA, GLA, GM2A, HEXA, HEXB, NPC1, NPC2, PSAP, SMPD1*
Tangier disease	1 gene	*ABCA1*
Tay-Sachs disease	3 genes	*GM2A, HEXA, HEXB*
Tyrosinemia, type I	1 gene	*FAH*
Urea cycle disorders	8 genes	*ARG1, ASL, ASS1, CPS1, NAGS, OTC, SLC25A13, SLC25A15*
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	1 gene	*ACADVL*
Wilson disease	1 gene	*ATP7B*

MITOCHONDRIAL DISEASES

Disease/Indication	No. of genes	Gene/s
Leber hereditary optic neuropathy (LHON)	all mt genes	mt genome
Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)	all mt genes	mt genome
Myoclonus Epilepsy with Ragged-Red Fibers (MERRF)	all mt genes	mt genome

MUSCULAR DISODERS

Disease/Indication	No. of genes	Gene/s
Bulbospinal muscular atrophy (Kennedy's disease)	1 gene	*AR*
Core myopathies	9 genes	*ACTA1, BIN1, DNM2, MTM1, RYR1, SELENON, TPM2, TPM3, TTN*
Duchenne/Becker Muscular dystrophy	1 gene	*DMD*
Dystrogylcanopathy	14 genes	*B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*
Emery-Dreifuss muscular dystrophy	4 genes	*EMD, FHL1, LMNA, SYNE2*
Hypokalemic periodic paralysis	3 genes	*CACNA1S, KCNJ2, SCN4A*
Hypokalemic periodic paralysis & Myotonias, non-dystrophic	5 genes	*CACNA1S, CLCN1, HSPG2, KCNJ2, SCN4A*
limb-girdle muscular dystrophy	14 genes	*ANO5, CAPN3, CAV3, DYSF, FKRP, FKTN, LMNA, MYOT, SGCA, SGCB, SGCD, SGCG, TCAP, TRIM32*
Metabolic myopathies	33 genes	*ACADVL, AGK, ALDOA, CPT2, DGUOK, ETFA, ETFB, ETFDH, FBXL4, GAA, HADHA, HADHB, INIP, ISCU, LAMA2, LDHA, LPIN1, MGME1, MPV17, PFKM, PGAM2, PHKA1, PHKB, POLG, PYGM, RRM2B, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP*

Disease/Indication	No. of genes	Gene/s
Muscle Disorders	147 genes	*ACADVL, ACTA1, AGK, ALDOA, ANO5, ASAH1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, BVES, CACNA1S, CAPN3, CAV3, CCDC78, CFL2, CHCHD10, CHKB, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNAJB2, DNAJB6, DNM2, DPM3, DYNC1H1, DYSF, EMD, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBXL4, FBXO38, FHL1, FKRP, FKTN, FLNC, GAA, GARS, GMPPB, GNE, HADHA, HADHB, HNRNPDL, HSPB8, HSPG2, IGHMBP2, INIP, ISCU, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, KY, LAMA2, LARGE1, LDB3, LDHA, LIMS2, LMNA, LMOD3, LPIN1, MATR3, MEGF10, MGME1, MICU1, MPV17, MTM1, MTMR14, MYF6, MYH2, MYH7, MYL1, MYO18B, MYOT, MYPN, NEB, ORAI1, PFKM, PGAM2, PHKA1, PHKB, PLEC, PLEKHG5, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYGM, PYROXD1, REEP1, RRM2B, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC25A20, SLC25A4, SLC5A7, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, SUCLG1, SYNE2, TCAP, TFG, TK2, TMEM43, TNNT1, TNPO3, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRPV4, TTN, TWNK, TYMP, UBA1, VAPB, VCP, VRK1*
Muscular dystrophies	43 genes	*ANO5, B3GALNT2, B4GAT1, BVES, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DMD, DNAJB6, DNM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, HNRNPDL, ITGA7, LAMA2, LARGE1, LIMS2, LMNA, MATR3, MYOT, PLEC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SELENON, SGCA, SGCB*
Muscular dystrophy, collagen-associated and others	7 genes	*CHKB, COL6A1, COL6A2, COL6A3, FHL1, ITGA7, SELENON*
Muscular dystrophy, congenital	21 genes	*CHKB, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DNM2, DPM3, FHL1, FKRP, FKTN, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMT1, POMT2, SELENON, TCAP*
Myopathies	78 genes	*ACADVL, ACTA1, AGK, ALDOA, BAG3, BIN1, CCDC78, CFL2, CNTN1, CPT2, CRYAB, DES, DGUOK, DNAJB6, DNM2, ETFA, ETFB, ETFDH, FBXL4, FHL1, FLNC, GAA, GNE, HADHA, HADHB, INIP,ISCU, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LDB3, LDHA, LMOD3, LPIN1, MEGF10, MGME1, MICU1, MPV17, MTM1, MTMR14, MYF6, MYH2, MYH7, MYL1, MYO18B, MYOT, MYPN, NEB, ORAI1, PFKM, PGAM2, PHKA1, PHKB, PLEC, POLG, PYGM, PYROXD1, RRM2B, RYR1, SELENON, SLC25A20, SLC25A4, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, SUCLG1, TK2, TNNT1, TPM2, TPM3, TTN, TWNK, TYMP, VCP*
Myopathy, congenital	34 genes	*ACTA1, BIN1, CCDC78, CFL2, CNTN1, DNM2, GNE, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MICU1, MTM1, MTMR14, MYF6, MYH2, MYH7, MYL1, MYO18B, MYPN, NEB, ORAI1, RYR1, SELENON, SPEG, SPTBN4, STAC3, STIM1, TNNT1, TPM2, TPM3, TTN, VCP*

Disease/Indication	No. of genes	Gene/s
Myopathy, myofibrillar	12 genes	*BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PLEC, PYROXD1, TTN*
Nemaline myopathy	11 genes	*ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3*
Spinal muscular atrophy (SMA1, 2, 3, 4)	1 gene	*SMN1*
Spinal muscular atrophy	22 genes	*ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HSPB8, IGHMBP2, PLEKHG5, REEP1, SLC5A7, TFG, TRIP4, TRPV4, UBA1, VAPB, VRK1*
Spinal muscular atrophy (late-onset)	13 genes	*ATP7A, BICD2, BSCL2, CHCHD10, DNAJB2, FBXO38, GARS, HSPB8, REEP1, SLC5A7, TFG, TRPV4, VAPB*
Spinal muscular atrophy (neonatal / early-onset) and pontocerebellar hypoplasia	9 genes	*ASAH1, ATP7A, EXOSC3, EXOSC8, IGHMBP2, PLEKHG5, TRPV4, UBA1, VRK1*
Steinert myotonic dystrophy	1 gene	*DMPK*

NEUROLOGICAL DISODERS

Disease/Indication	No. of genes	Gene/s
Absence seizures	7 genes	*CLCN2, GABRA1, GABRA5, GABRB3, GABRG2, SLC2A1, SLC6A1*
Alzheimer's disease	4 genes	*APOE, APP, PSEN1, PSEN2*
Alzheimer's disease, early-onset	3 genes	*APP, PSEN1, PSEN2*
Alzheimer's disease, late-onset	1 gene	*APOE*
Ataxia with oculomotor apraxia	4 genes	*APTX, PIK3R5, PNKP, SETX*

Disease/Indication	No. of genes	Gene/s
Ataxias	154 genes	*ABCB7, ABHD12, ADGRG1, AFG3L2, AHI1, AMACR, ANO10, APTX, ARL13B, ARSA, ATCAY, ATM, ATP13A2, ATP1A3, ATP8A2, ATXN10, B4GALNT1, BTD, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CC2D2A, CCDC88C, CEP290, CEP41, CHP1, CLCN2, CLN5, CLN6, COA7, COQ8A, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, GALC, GBA, GBA2, GCLC, GDAP2, GJB1, GJC2, GOSR2, GRID2, GRM1, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0586, KIF1C, KIF26B, KIF7, MARS2, MICU1, MME, MRE11, MTPAP, NEU1, NKX6-2, NPC1, NPC2, NPHP1, OFD1, OPA1, OPA3, PANK2, PDE10A, PDE6D, PDHX, PDYN, PEX10, PEX2, PIK3R5, PLA2G6, PLD3, PLP1, PMPCA, PNKP, PNPLA6, POC1B, POLG, POLR3A, PPT1, PRKCG, PRNP, PUM1, RNF216, RPGRIP1L, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC17A5, SLC1A3, SLC9A1, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, THG1L, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRPC3, TTBK2, TTC21B, TTPA, TUBB4A, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR81, WFS1, WWOX, XRCC1, ZNF423*
Ataxias, episodic	5 genes	*CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3*
Ataxias, syndromic	80 genes	*ABCB7, ABHD12, ADGRG1, AFG3L2, AHI1, AMACR, ARL13B, ARSA, ATCAY, ATM, ATP1A3, ATP8A2, ATXN10, BTD, CA8, CAPN1, CC2D2A, CEP290, CEP41, CLCN2, CLN5, CLN6, COQ8A, CP, CPLANE1, CSPP1, CYP27A1, DARS2, DLAT, DNAJC19, DNMT1, FLVCR1, GALC, GBA, GBA2, GJB1, GOSR2, INPP5E, KCNJ10, KIAA0586, KIF1C, KIF7, MARS2, MRE11, MTPAP, NPC1, NPC2, NPHP1, OFD1, OPA1, OPA3, PANK2, PDE10A, PDE6D, PEX10, PEX2, PLA2G6, PNPLA6, POC1B, POLG, POLR3A, RPGRIP1L, SACS, SIL1, SLC17A5, SPG7, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TUBB4A, VAMP1, VLDLR, WFS1, ZNF423*
ATTR amyloidosis, hereditary	1 gene	*TTR*
CADASIL (autosomal dominant cerebral arteriopathy with subcortical infarcts and leukoencephalopathy)	2 genes	*HTRA1, NOTCH3*
Charcot-Marie-Tooth disease type 1A	1 gene	*PMP22*
Choreatic movement disorders	23 genes	*ADCY5, ARSA, ATM, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, FRRS1L, FTL, GM2A, GNAO1, KCNA1, NKX2-1, OPA3, PANK2, PDE10A, PRNP, RNF216, SETX, TBP, VPS13A, XK*

Disease/Indication	No. of genes	Gene/s
Creutzfeldt–Jakob disease (CJD)	1 gene	*PRNP*
Dentatorubral pallidoluysian atrophy	1 gene	*ATN1*
Dravet syndrome	6 genes	*GABRD*, *GABRG2*, *PCDH19*, *SCN1A*, *SCN1B*, *SCN2A*
Epilepsy	147 genes	*AARS*, *ACTL6B*, *ADAM22*, *ADGRV1*, *ADRA2B*, *ALDH7A1*, *ALG13*, *AP3B2*, *ARHGEF15*, *ARHGEF9*, *ARV1*, *ARX*, *ATP1A2*, *BRAT1*, *CACNA1A*, *CACNA1E*, *CAD*, *CDK19*, *CDKL5*, *CERS1*, *CHD2*, *CHRNA2*, *CHRNA4*, *CHRNB2*, *CLCN2*, *CLCN4*, *CLN8*, *CNPY3*, *CNTN2*, *CPLX1*, *CSTB*, *CUX2*, *CYFIP2*, *DCX*, *DENND5A*, *DEPDC5*, *DMXL2*, *DNM1*, *DOCK7*, *DYRK1A*, *EEF1A2*, *EPM2A*, *FGF12*, *FOXG1*, *FRRS1L*, *GABBR2*, *GABRA1*, *GABRA2*, *GABRA5*, *GABRB1*, *GABRB2*, *GABRB3*, *GABRD*, *GABRG2*, *GAD1*, *GAL*, *GLDC*, *GLS*, *GLUL*, *GNAO1*, *GOSR2*, *GOT2*, *GPHN*, *GRIN2A*, *GRIN2B*, *GRIN2D*, *GUF1*, *HCN1*, *HDAC4*, *HNRNPU*, *IQSEC2*, *ITPA*, *KCNA1*, *KCNA2*, *KCNB1*, *KCNC1*, *KCNH5*, *KCNMA1*, *KCNQ2*, *KCNQ3*, *KCNT1*, *KCNT2*, *KCTD7*, *LGI1*, *LMNB2*, *MBD5*, *MDH2*, *MECP2*, *MEF2C*, *NECAP1*, *NEUROD2*, *NHLRC1*, *NPRL2*, *NPRL3*, *NTRK2*, *PACS2*, *PARS2*, *PCDH19*, *PHACTR1*, *PIGA*, *PIGB*, *PIGP*, *PIGQ*, *PLCB1*, *PLPBP*, *PNKP*, *PNPO*, *POLG*, *PPP3CA*, *PRDM8*, *PRICKLE2*, *PRRT2*, *RANBP2*, *RANGAP1*, *RELN*, *RHOBTB2*, *RNF13*, *ROGDI*, *SCARB2*, *SCN1A*, *SCN1B*, *SCN2A*, *SCN3A*, *SCN8A*, *SLC12A5*, *SLC13A5*, *SLC1A2*, *SLC25A12*, *SLC25A22*, *SLC2A1*, *SLC35A2*, *SLC6A1*, *SMC1A*, *SPTAN1*, *ST3GAL3*, *STX1B*, *STXBP1*, *SYNGAP1*, *SYNJ1*, *SZT2*, *TBC1D24*, *TRAK1*, *UBA5*, *UGDH*, *UGP2*, *WWOX*, *YWHAG*
Epilepsy, familial focal	13 genes	*CHRNA2*, *CHRNA4*, *CHRNB2*, *DEPDC5*, *GAL*, *GRIN2A*, *KCNA1*, *KCNT1*, *LGI1*, *NPRL2*, *NPRL3*, *RELN*, *SCN3A*
Epilepsy, generalized myoclonic	19 genes	*ADRA2B*, *CERS1*, *CLCN2*, *CNTN2*, *CSTB*, *EPM2A*, *GABRA1*, *GABRD*, *GOSR2*, *KCNC1*, *KCNMA1*, *KCTD7*, *LMNB2*, *NHLRC1*, *PRDM8*, *PRICKLE2*, *SCARB2*, *SLC6A1*, *TBC1D24*
Epilepsy, therapy relevant	7 genes	*ALDH7A1*, *KCNQ2*, *PLPBP*, *PNPO*, *PRRT2*, *SCN1A*, *SLC2A1*
Epileptic encephalopathy, early infantile (EIEE) (extended)	96 genes	*AARS*, *ACTL6B*, *ADAM22*, *ALG13*, *AP3B2*, *ARHGEF9*, *ARV1*, *ARX*, *CACNA1A*, *CACNA1E*, *CAD*, *CDK19*, *CDKL5*, *CHD2*, *CNPY3*, *CPLX1*, *CUX2*, *CYFIP2*, *DCX*, *DENND5A*, *DMXL2*, *DNM1*, *DOCK7*, *EEF1A2*, *FGF12*, *FOXG1*, *FRRS1L*, *GABBR2*, *GABRA1*, *GABRA2*, *GABRA5*, *GABRB1*, *GABRB2*, *GABRB3*, *GABRG2*, *GAD1*, *GLS*, *GNAO1*, *GOT2*, *GPHN*, *GRIN2B*, *GRIN2D*, *GUF1*, *HCN1*, *HNRNPU*, *ITPA*, *KCNA2*, *KCNB1*, *KCNQ2*, *KCNT1*, *KCNT2*, *MDH2*, *MECP2*, *NECAP1*, *NEUROD2*, *NTRK2*, *PACS2*, *PARS2*, *PCDH19*, *PHACTR1*, *PIGA*, *PIGB*, *PIGP*, *PIGQ*, *PLCB1*, *PNKP*, *POLG*, *PPP3CA*, *RANGAP1*, *RHOBTB2*, *RNF13*, *SCN1A*, *SCN1B*, *SCN2A*, *SCN3A*, *SCN8A*, *SLC12A5*, *SLC13A5*, *SLC1A2*, *SLC25A12*, *SLC25A22*, *SLC35A2*, *SMC1A*, *SPTAN1*, *ST3GAL3*, *STXBP1*, *SYNGAP1*, *SYNJ1*, *SZT2*, *TBC1D24*, *TRAK1*, *UBA5*, *UGDH*, *UGP2*, *WWOX*, *YWHAG*

Disease/Indication	No. of genes	Gene/s
Febrile seizures, familial / Epilepsy, generalized, with febrile seizures plus	10 genes	*ADGRV1, GABRD, GABRG2, HCN1, PCDH19, PRRT2, SCN1A, SCN1B, SCN2A, STX1B*
Female restricted epilepsy with intellectual disability	1 gene	*PCDH19*
Friedreich ataxia	1 gene	*FXN*
Gerstmann-Straussler disease	1 gene	*PRNP*
GLUT1 deficiency syndrome	1 gene	*SLC2A1*
Hereditary neuropathy with liability to pressure palsies	1 gene	*PMP22*
Hereditary spastic paraplegia (HSP)	65 genes	*AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, C19orf12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENOI, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TUBB4A, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27*
Holoproencephaly	15 genes	*CDON, CNOT1, DHCR7, DISP1, DLL1, FGF8, FGFR1, GLI2, PTCH1, SHH, SIX3, STAG2, TDGF1, TGIF1, ZIC2*
Huntington disease	1 gene	*HTT*
Hyperekplexia	5 genes	*ARHGEF9, ATAD1, GLRA1, GLRB, SLC6A5*
Leukoencephalopathy and Leukodystrophy	86 genes	*AARS, AARS2, ABCD1, ACBD5, ACOX1, ADAR, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, CLDN11, CNP, CSF1R, CTC1, CYP27A1, DARS, DARS2, DEGS1, EARS2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS, FAM126A, GALC, GAN, GBE1, GFAP, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, HTRA1, IFIH1, KARS, L2HGDH, LMNB1, LSM11, MLC1, NAXD, NAXE, NKX6-2, NOTCH3, OCLN, PLAA, PLEKHG2, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RAB11B, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU7-1, SAMHD1, SCP2, SLC16A2, SLC17A5, SLC25A12, SNORD118, SOX10, STAT2, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYROBP, UFM1, VPS11, ZNHIT3*

Disease/Indication	No. of genes	Gene/s
Lissencephaly	17 genes	*ACTB*, *ACTG1*, *ARX*, *CDK5*, *CEP85L*, *CRADD*, *DCX*, *KATNB1*, *LAMB1*, *MACF1*, *NDE1*, *PAFAH1B1*, *RELN*, *TMTC3*, *TUBA1A*, *TUBB2B*, *TUBG1*
Migraine, familial hemiplegic	3 genes	*ATP1A2*, *CACNA1A*, *SCN1A*
Neuropathies, hereditary	105 genes	*AARS*, *ABCD1*, *ABHD12*, *AIFM1*, *ALDH18A1*, *ALS2*, *AP5Z1*, *ARL6IP1*, *ATL1*, *ATL3*, *ATP13A2*, *ATP1A1*, *ATP7A*, *B4GALNT1*, *BAG3*, *BICD2*, *BSCL2*, *C12orf65*, *C19orf12*, *CAPN1*, *CTDP1*, *CYP2U1*, *CYP7B1*, *DCTN1*, *DDHD1*, *DNAJB2*, *DNM2*, *DNMT1*, *DST*, *DYNC1H1*, *EGR2*, *ELP1*, *FA2H*, *FBLN5*, *FBXO38*, *FGD4*, *FIG4*, *GAN*, *GARS*, *GDAP1*, *GJB1*, *GNB4*, *HARS*, *HINT1*, *HK1*, *HSPB1*, *HSPB8*, *IGHMBP2*, *INF2*, *KIF1A*, *KIF5A*, *LITAF*, *LRSAM1*, *MARS*, *MATR3*, *MCM3AP*, *MFN2*, *MME*, *MORC2*, *MPV17*, *MPZ*, *MTMR2*, *NDRG1*, *NEFH*, *NEFL*, *NGF*, *NTRK1*, *PDK3*, *PLEKHG5*, *PMP22*, *PNKP*, *PNPLA6*, *POLG*, *PRDM12*, *PRPS1*, *PRX*, *RAB7A*, *REEP1*, *RETREG1*, *SACS*, *SBF1*, *SBF2*, *SCN10A*, *SCN11A*, *SCN9A*, *SEPT9*, *SETX*, *SH3TC2*, *SIGMAR1*, *SLC12A6*, *SLC25A46*, *SLC5A7*, *SOX10*, *SPAST*, *SPG11*, *SPTLC1*, *SPTLC2*, *SYT2*, *TFG*, *TRIM2*, *TRPV4*, *TTR*, *VCP*, *WNK1*, *YARS*
Polymicrogyria	10 genes	*ADGRG1*, *AKT3*, *CCND2*, *GRIN2B*, *KIF1BP*, *PI4KA*, *PIK3CA*, *PIK3R2*, *TUBA8*, *TUBB2B*
Pontocerebellar hypoplasia	21 genes	*AMPD2*, *CASK*, *CHMP1A*, *CLP1*, *COASY*, *EXOSC3*, *EXOSC8*, *EXOSC9*, *PPIL1*, *RARS2*, *SEPSECS*, *SLC25A46*, *TBC1D23*, *TOE1*, *TSEN15*, *TSEN2*, *TSEN34*, *TSEN54*, *VPS51*, *VPS53*, *VRK1*
Pyridoxine-dependent epilepsy	1 gene	*ALDH7A1*
Seizures, benign familial neonatal	6 genes	*CHRNA2*, *KCNQ2*, *KCNQ3*, *PRRT2*, *SCN2A*, *SCN8A*
Small fiber neuropathy	3 genes	*SCN10A*, *SCN11A*, *SCN9A*
Spastic ataxia	15 genes	*ABHD12*, *AFG3L2*, *B4GALNT1*, *CAPN1*, *CHP1*, *GBA2*, *GJC2*, *KIF1C*, *MARS2*, *MTPAP*, *NKX6-2*, *POLR3A*, *SACS*, *SPG7*, *VAMP1*
Spinocerebellar ataxias, autosomal dominant (SCA)	6 genes	*ATXN1*, *ATXN2*, *ATXN3*, *ATXN7*, *CACNA1A*, *TBP*

Disease/Indication	No. of genes	Gene/s
Spinocerebellar and other hereditary ataxias	144 genes	*ABCB7, ADGRG1, AFG3L2, AHI1, AMACR, ANO10, APTX, ARL13B, ARSA, ATCAY, ATP13A2, ATP1A3, ATP8A2, ATXN10, BTD, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CC2D2A, CCDC88C, CEP290, CEP41, CHP1, CLCN2, CLN5, COA7, COQ8A, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, GALC, GBA, GBA2, GCLC, GDAP2, GJB1, GOSR2, GRID2, GRM1, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0586, KIF1C, KIF26B, KIF7, MARS2, MICU1, MME, MRE11, MTPAP, NEU1, NPC1, NPC2, NPHP1, OPA1, OPA3, PANK2, PDE10A, PDE6D, PDHX, PDYN, PEX10, PEX2, PIK3R5, PLD3, PLP1, PMPCA, PNKP, PNPLA6, POC1B, POLG, PRKCG, PRNP, PUM1, RNF216, RPGRIP1L, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC17A5, SLC1A3, SLC9A1, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, THG1L, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRPC3, TTBK2, TTC21B, TTPA, TUBB4A, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR81, WFS1, WWOX, XRCC1, ZNF423*
Tubulinopathies	8 genes	*TBCD, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1*

PANCREATIC DISEASES

Disease/Indication	No. of genes	Gene/s
Pancreatitis	6 genes	*CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*
Shwachman-Diamond syndrome	1 gene	*SBDS*

RASOPATHIES

Disease/Indication	No. of genes	Gene/s
Cardiofaciocutaneous syndrome	4 genes	*BRAF, KRAS, MAP2K1, MAP2K2*
Costello syndrome	1 gene	*HRAS*

Disease/Indication	No. of genes	Gene/s
Legius syndrome	1 gene	*SPRED1*
LEOPARD syndrome	3 genes	*BRAF, PTPN11, RAF1*
Neurofibromatosis type 1 (NF1)	1 gene	*NF1*
Neurofibromatosis-Noonan syndrome	2 genes	*NF1, PTPN11*
Noonan syndrome	19 genes	*BRAF, CBL, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2*
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	1 gene	*CBL*
Noonan syndrome-like disorder with loose anagen hair	2 genes	*PPP1CB, SHOC2*
RASopathies	21 genes	*BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1*

RENAL DISEASES

Disease/Indication	No. of genes	Gene/s
Alport syndrome, Thin-basement-membrane nephropathy	4 genes	*COL4A3, COL4A4, COL4A5, MYH9*
CAKUT (congenital anomalies of the kidney and urinary tract)	50 genes	*ACE, ACTA2, ACTG2, AGT, AGTR1, ANOS1, BICC1, BMP4, BMP7, CDC5L, CHD1L, CHRM3, DACH1, DSTYK, ETV4, ETV5, EYA1, FGF20, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GATA3, GDNF, GREM1, GRIP1, HNF1B, HPSE2, ITGA3, ITGA8, LRIG2, MUC1, PAX2, PAX8, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SOX17, TRAP1, UMOD, UPK2, UPK3A, WNT4, WT1, ZIC3*
Gitelman syndrome	1 gene	*SLC12A3*
Hyperoxaluria	3 genes	*AGXT, GRHPR, HOGA1*
Kidney agenesis/dysgenesis	18 genes	*ANOS1, BMP4, CDC5L, DSTYK, FGF20, FRAS1, FREM1, FREM2, GREM1, HNF1B, ITGA8, PAX2, RET, SALL1, SIX2, TRAP1, UPK3A, WNT4*

Disease/Indication	No. of genes	Gene/s
Lower urinary tract obstruction (LUTO), congenital	10 genes	*ACTA2, ACTG2, BMP4, CHD1L, CHRM3, DSTYK, GATA3, HNF1B, PAX2, ROBO2*
Nephrogenic diabetes insipidus (NDI)	2 genes	*AQP2, AVPR2*
Nephronophthisis	27 genes	*AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, PAX2, RPGRIP1L, SDCCAG8, SLC41A1, TMEM216, TMEM237, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423*
Nephrotic syndrome / Focal segmental glomerulosclerosis	39 genes	*ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, GLA, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, MYH9, MYO1E, NEIL1, NPHS1, NPHS2, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, WT1*
Polycystic kidney disease	7 genes	*DNAJB11, DZIP1L, GANAB, HNF1B, PKD1, PKD2, PKHD1*
Renal cysts and diabetes syndrome	1 gene	*HNF1B*
Renal Disease	125 genes	*ACE, ACTA2, ACTG2, ACTN4, AGT, AGTR1, AGXT, AHI1, ANKS6, ANLN, ANOS1, APOL1, AQP2, ARHGAP24, ARHGDIA, AVPR2, BICC1, BMP4, BMP7, CC2D2A, CD2AP, CDC5L, CEP164, CEP290, CEP83, CFH, CHD1L, CHRM3, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DACH1, DCDC2, DGKE, DNAJB11, DSTYK, DZIP1L, EMP2, ETV4, ETV5, EYA1, FGF20, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GANAB, GATA3, GDNF, GLA, GLIS2, GREM1, GRHPR, GRIP1, HNF1B, HOGA1, HPSE2, IFT172, INF2, INVS, IQCB1, ITGA3, ITGA8, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, LRIG2, MAPKBP1, MUC1, MYH9, MYO1E, NEIL1, NEK8, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, PAX2, PAX8, PDSS2, PKD1, PKD2, PKHD1, PLCE1, PTPRO, REN, RET, ROBO2, RPGRIP1L, SALL1, SCARB2, SDCCAG8, SIX1, SIX2, SIX5, SLC12A3, SLC41A1, SLC7A7, SMARCAL1, SOX17, TMEM216, TMEM237, TMEM67, TRAP1, TRPC6, TTC21B, UMOD, UPK2, UPK3A, WDR19, WNT4, WT1, XPNPEP3, ZIC3, ZNF423*
Renal tubular dysgenesis	4 genes	*ACE, AGT, AGTR1, REN*
Urinary tract malformation	23 genes	*ACTA2, ACTG2, BMP4, BMP7, CHD1L, CHRM3, DACH1, DSTYK, EYA1, FRAS1, FREM1, FREM2, GATA3, HNF1B, HPSE2, ITGA8, LRIG2, PAX2, RET, ROBO2, SALL1, SOX17, TRAP1*

WHOLE EXOME SEQUENCING

Whole exome sequencing (WES) is a comprehensive genetic test which analyzes the protein-coding regions (exons) of all human genes (~20,000 genes). WES substantially increases the chances of finding the genetic cause of diseases with complex and non-specific symptoms. It reduces the time and cost from symptom presentation to diagnosis, and has an increased diagnostic yield. Following is a table outlining some recommended indications for WES.

WES ANALYSIS

Type of Exome Analysis	Individuals Tested
Quad exome analysis	Patient and both biological parents and one additional relative (e.g., sibling)
Trio exome analysis	Patient and both biological parents (most common for rare disease diagnosis) or relatives are tested
Duo exome analysis*	Patient and one biological parent or relative are tested
Single exome analysis*	Only the patient is tested

* (Please ONLY request if parent(s) sample(s) are absolutely not available)

Disease/Indication	Type of Exome Analysis
Autism spectrum disorder with unspecific phenotype	TRIO exome analyses
Deafness in combination with developmental delay	TRIO exome analyses
Global developmental delay with unspecific phenotype	TRIO exome analyses
Intellectual disability with unspecific phenotype	TRIO exome analyses
Neonatal cardiomyopathy	TRIO exome analyses with mtDNA
Neurological phenotype in combination with developmental delay	TRIO exome analyses
Syndromic congenital heart defect	TRIO exome analyses

FLEXOME

In traditional exome sequencing, all the protein-coding regions in the genome (exons) are sequenced. This provides an extensive overview of an individual's genetic makeup. However, it can be challenging to interpret this vast amount of data, especially when searching for the genetic basis of complex diseases that may be caused by pathogenic variants in any of numerous different genes.

FlexOme sequencing is focusing on the genes associated with a particular disorder or group of disorders. This allows for efficient and precise genetic analysis in situations where the number of potentially causal genes is high and no specific genetic panel has been definitively established. This approach provides a more targeted yet adaptable tool for diagnosing genetic disorders. Following is a table outlining some possible indications for FlexOme sequencing.

Indications for FlexOme Sequencing	No. of genes	Genes
Ataxia flexome panel	257 genes	*AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ACO2, ADCK3, ADPRHL2, AFG3L2, AGTPBP1, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP13A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, AUH, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C12ORF4, C5ORF42, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHCHD10, CLCN2, CLN5, CLN6, CLN8, CLPB, CLPP, COA7, COASY, COQ2, COQ4, COX20, CP, CSTB, CTBP1, CTDP1, CTSA, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DHPS, DNAJC19, DNAJC5, DNMT1, DOCK3, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HEXB, HIBCH, INPP5E, IRF2BPL, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF5A, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, LRSAM1, MARS2, MECR, MGME1, MKKS, MKS1, MME, MRE11A, MSTO1, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX2-1, NKX6-2, NOL3, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PANK2, PAX6, PDYN, PEX10, PEX16, PEX2, PEX3, PEX6, PEX7, PHYH, PMM2, PNKD, PNKP, PNPLA6, POLG, PRKCG, PRRT2, PUM1, RNF216, RORA, RPGRIP1L, RUBCN, SACS, SAMD9L, SCYL1, SERAC1, SETX, SH3TC2, SIL1, SLC17A5, SLC1A3, SLC20A2, SLC25A15, SLC25A46, SLC2A1, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG11, SPG20, SPG7, SPTBN2, SQSTM1, STUB1, STXBP1, SUOX, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRAPPC11, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, UBA5, UBTF, UCHL1, VAMP1, VLDLR, VPS13D, VWA3B, WDPCP, WDR81, WFS1, WWOX, XRCC1, ZFYVE26, ZNF423*

Indications for FlexOme Sequencing	No. of genes	Genes
Cholestasis flexome panel	52 genes	*ABCB11, ABCB4, ABCC2, AKR1D1, ATP8B1, BAAT, CFTR, CLDN1, CREB3L3, CYP7B1, DCDC2, DGUOK, EPCAM, FAH, HSD3B7, JAG1, LCT, LIPA, LMF1, MKS1, MPV17, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, SCYL1, SERPINA1, SLC25A13, SLC26A3, SLCO1B1, SLCO1B3, SMPD1, SPINT2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, VIPAS39, VPS33B*
CMT, Charcot-Marie-Tooth neuropathy flexome panel	153 genes	*AARS, AGTPBP1, AIFM1, AMACR, ARHGEF10, ATAD3A, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12ORF65, CCT5, CHCHD10, COA7, COX10, COX6A1, CTDP1, CYP27A1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FBXO38, FGD4, FIG4, FXN, GAN, GARS, GDAP1, GJB1, GNB4, GNE, GSN, HADHB, HARS, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTMR2, MYOT, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, PNKP, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A, REEP1, SACS, SBF1, SBF2, SCN11A, SCN9A, SCYL1, SEPT9, SETX, SH3TC2, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SMAD3, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, TYMP, UBA1, VCP, WNK1, YARS, ZFYVE26*
Comprehensive cardiology flexome panel	260 genes	*AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ALPK3, ANK2, ANO5, APOA1, ATPAF2, BAG3, BRAF, CACNA1C, CACNA1D, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CASZ1, CAV3, CBL, CDH2, CHKB, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, ENPP1, EPG5, ETFA, ETFB, ETFDH, FAH, FBXL4, FBXO32, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA5, GATA6, GATAD1, GATC, GBE1, GFM1, GLA, GLB1, GMPPB, GNB5, GSK3B, GTPBP3, GUSB, HADHA, HAND1, HAND2, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, KLHL24, KRAS, LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPEP, MLYCD, MRPL3, MRPL44, MRPS22, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYO18B, MYOT, MYPN, MYRF, NDUFAF2, NDUFB11, NEXN, NF1, NKX2-5, NONO, NOS1AP, NRAP, NRAS, NUP155, PARS2, PCCA, PCCB, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMT1, PPA2, PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC12A3, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, STAG2, TAB2, TANGO2, TAZ, TBX20, TBX5, TCAP, TECRL, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM32, TRPM4, TSFM, TTN, TTR, VARS2, VCL, VCP, VPS13A, XK*

Indications for FlexOme Sequencing	No. of genes	Genes
Comprehensive monogenic diabetes flexome panel	67 genes	*ABCC8, APPL1, BLK, CEL, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC16A1, SLC2A2, UCP2, WFS1, ZFP57*
Cystic kidney disease flexome panel	43 genes	*ANKS6, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, EYA1, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, SIX5, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423*
Dilated cardiomyopathy flexome panel	130 genes	*ABCC6, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, APOA1, BAG3, CASZ1, CHKB, CHRM2, CPT2, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DYSF, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FHOD3, FKRP, FKTN, FLNC, FOXD4, GATA4, GATA6, GATC, GBE1, GLB1, GSK3B, HAND1, HCN4, ILK, JPH2, JUP, KLHL24, LAMP2, LDB3, LEMD2, LMNA, LMOD2, LRRC10, MLYCD, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYBPC3, MYBPHL, MYH6, MYH7, MYL4, NEXN, NKX2-5, NRAP, PCCA, PCCB, PKP2, PLEKHM2, PLN, PPCS, PRDM16, QRSL1, RAF1, RBCK1, RBM20, RMND1, SCN5A, SLC22A5, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TTN, TTR, VCL, VPS13A*
Dystonia flexome panel	68 genes	*ADCY5, ANO3, ATP1A3, BCAP31, CACNA1B, CACNA1G, DCAF17, DNAJC12, FA2H, FITM2, GCH1, GNAL, KCNMA1, KMT2B, MECR, MIPEP, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, PDE10A, PDGFB, PDGFRB, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC39A14, SPR, TH, THAP1, TOR1A, UBTF, VAC14*
Ehlers-Danlos flexome panel	41 genes	*ABCC6, ADAMTS2, ADAMTSL2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, CHST3, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, GORAB, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFBR1, TGFBR2, ZNF469*
Glycogen storage disorder flexome panel	29 genes	*AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4*

Indications for FlexOme Sequencing	No. of genes	Genes
Hemochromatosis flexome panel	5 genes	*HAMP, HFE, HFE2, SLC40A1, TFR2*
Hereditary angioedema (HAE) flexome panel	9 genes	*SERPING1, PLG, F12, KNG1, MYOF, ANGPT1, HS3ST6, CPN1, DAB2IP*
Hereditary endocrine cancer flexome panel	22 genes	*AIP, APC, CDC73, CDKN1B, DICER1, FH, MAX, MEN1, MET, NF1, PRKAR1A, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, VHL, WRN*
Hereditary hemorrhagic telangiectasia (HHT) flexome panel	6 genes	*ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*
Hereditary paraganglioma-pheochromocytoma flexome panel	24 genes	*ATRX, CSDE1, DLST, EGLN1, EGLN2, FH, H3F3A, HIF2A, IDH1, IRP1, KIF1B, MAX, MDH2, NF1, PHD1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL*
Hyperlipidemia flexome panel	20 genes	*ABCA1, ABCG5, ABCG8, ALMS1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9*
Hyperparathyroidism flexome panel	14 genes	*AIRE, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GCM2, GNA11, MEN1, PTH, RET, TRPV6*
Hypertrophic cardiomyopathy (HCM) flexome panel	92 genes	*ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALPK3, APOA1, BAG3, BRAF, CACNA1C, CBL, COX15, CPT2, CSRP3, DES, ELAC2, EPG5, FBXL4, FHL1, FHOD3, FLNC, FXN, GAA, GLA, GSK3B, HRAS, JPH2, KLHL24, KRAS, LAMP2, MAP2K1, MAP2K2, MIPEP, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTO1, MYBPC3, MYH7, MYL2, MYL3, NDUFAF2, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, SOS1, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL*
Hypoglykemia flexome panel	50 genes	*ABCC8, ACAT1, ACSF3, AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, MPV17, NHLRC1, OXCT1, PC, PCK1, PDX1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, UCP2*
Marfan flexome panel	36 genes	*ABL1, ADAMTS10, ADAMTS17, ADAMTSL4, AEBP1, B3GAT3, BGN, CBS, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DLG4, EFEMP2, FBN1, FBN2, FLCN, LOX, MAT2A, MED12, PLOD1, SKI, SLC2A10, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, VCAN, ZDHHC9*
Migraine flexome panel	47 genes	*ATP1A2, ATP1A3, CACNA1A, KCNK18, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1*

Indications for FlexOme Sequencing	No. of genes	Genes
MODY flexome panel	54 genes	*ABCC8, APPL1, BLK, CEL, GATA6, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NEUROD1, PAX4, PDX1, RFX6, WFS1*
Monogenic obesity flexome panel	41 genes	*ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CPE, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP*
Neurofibromatosis flexome panel	9 genes	*KIT, KITLG, LZTR1, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1*
Noonan flexome panel	36 genes	*ACTB, ACTG1, BMP2, BRAF, CBL, CCNK, CDC42, EPHB4, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NF1, NF2, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SASH1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, STAMBP, SYNGAP1*
Organic acidemia/aciduria and cobalamin defiency flexome panel	54 genes	*ABCD4, ACADSB, ACAT1, ACSF3, ADK, AHCY, AMN, BCKDHA, BCKDHB, BCS1L, CBS, CD320, CLPB, CTH, CUBN, D2HGDH, DBT, DLD, ETFA, ETFB, ETFDH, FLAD1, GCDH, GIF, GNMT, HCFC1, HIBCH, HMGCL, IDH2, IVD, L2HGDH, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, PCCA, PCCB, PEPD, SERAC1, SLC25A1, SUCLA2, SUCLG1, SUGCT, TCN2, UMPS*
Osteogenesis flexome panel	33 genes	*ALPL, ARCN1, B3GAT3, B4GALT7, BMP1, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, FAM46A, FGF23, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC29A3, SLC34A3, SP7, SPARC, TAPT1, TMEM38B, WNT1*
Overgrowth syndrome flexome panel	48 genes	*AKT1, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF135, SETD2, SYN1, TMEM94, TSC1, TSC2, UPF3B, ZBTB20*
Palmoplantar flexome panel	26 genes	*AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJB2, GJB4, GJB6, JUP, KRT1, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, PKP1, SERPINB7, SLURP1, SMARCAD1, TRPV3, WNT10A*
Pancreatitis flexome panel	9 genes	*APOA5, APOC2, CFTR, CPA1, CTRC, GPIHBP1, PRSS1, SPINK1, UBR1*

Indications for FlexOme Sequencing	No. of genes	Genes
Parkinson	82 genes	*ATP13A2, ATP1A3, ATP7B, C19ORF12, CHCHD10, CHCHD2, CP, CSF1R, DCTN1, DNAJC12, DNAJC5, DNAJC6, FBXO7, FTL, GBA, GCH1, GRN, LRRK2, MAPT, 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-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NUS1, PANK2, PARK2, PARK7, PDE10A, PDE8B, PDGFB, PDGFRB, PINK1, PLA2G6, POLG, PRKRA, PSEN1, RAB39B, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SPR, SYNJ1, TH, VPS13A, VPS13C, VPS35, XPR1*
Polycystic kidney disease	13 genes	*DNAJB11, DZIP1L, GANAB, HNF1B, JAG1, LRP5, NOTCH2, PKD1, PKD2, PKHD1, PRKCSH, SEC61A1, SEC63*
Polyneuropathy (CMT)	236 genes	*AARS, DHTKD1, HSPB3, PDK3, SLC12A6, ABCA1, DHX9, HSPB8, PDXK, SLC25A19, ABCD1, DNAJB2, IARS2, PDYN, SLC25A46, ABHD12, DNAJB6, IGHMBP2, PEX10, SLC52A2, ACOX1, DNAJC3, INF2, PEX7, SLC52A3, AGTPBP1, DNM2, ITPR3, PHYH, SLC5A6, AGXT, DNMT1, JAG1, PIGB, SLC5A7, AIFM1, DRP2, KCNA2, PLEKHG5, SMN1, AMACR, DST, KIF1A, PLP1, SORD, ANO5, DYNC1H1, KIF1B, PMM2, SOX10, AP1S1, DYSF, KIF5A, PMP2, SPART, APOA1, EGR2, LDB3, PMP22, SPAST, APTX, ELP1, LITAF, PNKP, SPG11, ARHGEF10, EMILIN1, LMNA, PNPLA6, SPG7, ARSA, ERCC6, LRSAM1, POLG, SPTAN1, ATL1, ERCC8, LYST, POLG2, SPTBN4, ATL3, ETFDH, MAG, POLR3A, SPTLC1, ATM, FAH, MARS, POLR3B, SPTLC2, ATP1A1, FAM126A, MATR3, PPOX, SUCLA2, ATP7A, FBLN5, MCM3AP, PRDM12, SURF1, B4GALNT1, FBXO38, MFN2, PRKCG, SYT2, BAG3, FGD4, MMACHC, PRNP, TCAP, BCKDHB, FIG4, MME, PRPS1, TDP1, BICD2, FLNC, MORC2, PRX, TECPR2, BSCL2, FLVCR1, MPV17, PSMC3, TFG, C12orf65, FXN, MPZ, PTEN, TIA1, C1orf194, GALC, MT-ATP6, PTPN11, TRIM2, CADM3, GAN, MTMR2, PTRH2, TRPA1, CAV3, GARS, MT-RNR1, TRPV4, CD59, GBA2, MT-TL1, REEP1, TTN, CHCHD10, GBF1, MTTP, RETREG1, TTPA, CNTNAP1, GDAP1, MYH14, RTN2, TTR, COA7, GJB1, MYH7, SACS, TUBB3, COQ7, GJC2, MYOT, SARS, TWNK, COX20, GLA, NAGA, SBF1, TYMP, COX6A1, GNB4, NAGLU, SBF2, UBA1, CPOX, GNE, NDRG1, SCARB2, VAPB, CRYAB, GSN, NEB, SCN10A, VCP, CTDP1, HADHA, NEFH, SCN11A, VPS13A, CYP27A1, HADHB, NEFL, SCN9A, VRK1, DARS2, HARS, NEMF, SCO2, VWA1, DCTN1, HEXA, NGF, SCYL1, WARS, DCTN2, HEXB, NTRK1, SEPT9, WNK1, DEGS1, HINT1, NUDT2, SETX, XK, DES, HK1, OPA1, SGPL1, XPA, DGAT2, HMBS, OPA3, SH3TC2, XRCC1, DGUOK, HSPB1, PDHA1, SIGMAR1, YARS, RAB7A, ZFYVE26*

Indications for FlexOme Sequencing	No. of genes	Genes
Primary immunodeficiency flexome panel	336 genes	*ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP3B1, AP3D1, ARHGEF1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CDK9, CEBPE, CECR1, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COG6, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DGAT1, DGKE, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, EFL1, ELANE, EPG5, ERCC6L2, EXTL3, FADD, FANCA, FAS, FASLG, FAT4, FCGR3A, FCHO1, FERMT3, FOXN1, FOXP3, G6PC, G6PC3, G6PD, GATA2, GFI1, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MASP1, MEFV, MKL1, MOGS, MRE11A, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, OBFC1, OFD1, ORAI1, OTULIN, PARN, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLA1, POLD1, POLE, POLE2, POMP, PRF1, PRG4, PRKCD, PRKDC, PSENEN, PSMB4, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RECQL4, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RLTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TINF2, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UBA1, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341*
Pulmonary artery hypertension (PAH) flexome panel	23 genes	*ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, KLF2, NFU1, NOTCH3, RASA1, SARS2, SMAD4, SMAD9, SOX17, STRA6, TBX4*
Thrombocytopenia flexome panel	37 genes	*ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CYCS, EFL1, ETV6, FLI1, FLNA, FYB, GATA1, GBA, GFI1B, GP1BA, GP1BB, GP9, HOXA11, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, SLFN14, SRC, SRP54, THBD, TUBB1, WAS, WIPF1*

ONCOLOGY

HEREDITARY CANCER PANELS

Disease/Indication	No. of genes	Gene/s
BAP1 tumor predisposition syndrome	1 gene	*BAP1*
Birt Hogg Dubé syndrome	1 gene	*FLCN*
Breast and ovarian cancer	28 genes	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, SMARCA4, SMARCB1, STK11, TP53, XRCC2*
Breast and ovarian cancer – PARP inhibitor treatment	2 genes	*BRCA1, BRCA2*
Breast and ovarian cancer – recommended*	18 genes	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
Colon cancer	23 genes	*APC, AXIN2, BMPR1A, CHEK2, EPCAM, GALNT12, GREM1*[1)]*, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11, TP53*
Colon cancer – recommended**	17 genes	*APC, BMPR1A, EPCAM, GREM1*[1)]*, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
DICER1 syndrome	1 gene	*DICER1*
Endocrine tumors	14 genes	*AIP, CDC73, CDKN1B, MAX, MEN1, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
Familial adenomatous polyposis coli (FAP), MUTYH associcated polyposis	2 genes	*APC, MUTYH*
Familial atypical multiple mole melanoma (FAMMM) and pancreatic cancer syndrome	1 gene	*CDKN2A*
Familial isolated pituitary adenoma (FIPA)	1 gene	*AIP*

Disease/Indication	No. of genes	Gene/s
Fanconi anemia	22 genes	*BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2*
Gastrointestinal tumors	21 genes	*APC, BMPR1A, CDH1, CTNNA1, EPCAM, GREM1*[1)], *KIT, MLH1, MSH2, MSH6, MUTYH, PDGFRA, PMS2, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, STK11*
Hereditary cancer panel	137 genes	*AIP, AKT1, ANKRD26, AP2S1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1, CASR, CDC73, CDH1, CDH23, CDK12, CDK4, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CEBPA, CHEK1, CHEK2, CTNNA1, DDB2, DDX41, DICER1, DLST, EGLN1, EPAS1, EPCAM, EPOR, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GCM2, GNA11, GNAS, GPR101, GREM1*[1)], *HOXB13, KIF1B, KIT, KITLG, LZTR1, MAD2L2, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTCH2, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54L, RB1, RECQL, RECQL4, RET, RFWD3, RNF43, RPS20, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPRED1, SRP72, STK11, SUFU, TERT, TMEM127, TP53, TSC1, TSC2, UBE2T, VHL, WT1 XPA, XPC, XRCC2, XRCC3*
Hereditary diffuse gastric cancer	2 genes	*CDH1, CTNNA1*
Hereditary leimyomatosis and renal cell carcinoma (HLRCC)	1 gene	*FH*
Hereditary papillary renal cell carcinoma (HPRCC)	1 gene	*MET*
Hyperparathyroidism jaw tumor syndrome	1 gene	*CDC73*
Juvenile polyposis syndrome	2 genes	*BMPR1A, SMAD4*
Kidney cancers	11 genes	*BAP1, FH, FLCN, MET, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, VHL*
Li Fraumeni syndrome	1 gene	*TP53*
Multiple endocrine neoplasia type 1	1 gene	*MEN1*
Multiple endocrine neoplasia type 2	1 gene	*RET*

Disease/Indication	No. of genes	Gene/s
Multiple endocrine neoplasia type 4	1 gene	*CDKN1B*
Nervous system/brain tumors	14 genes	*AIP, LZTR1, NF1, NF2, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SMARCE1, TP53, VHL*
Neurofibromatosis type 1	1 gene	*NF1*
Nevoid basal cell carcinoma syndrome	3 genes	*PTCH1, PTCH2, SUFU*
Pancreatic tumors	15 genes	*ATM, BRCA1, BRCA2, CDKN1B, CDKN2A, EPCAM, MEN1, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53, VHL*
Peutz-Jeghers syndrome	1 gene	*STK11*
Pheochromocytoma paraganglioma syndrome	7 genes	*MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127*
Prostate cancer	11 genes	*ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2*
PTEN hamartoma tumor syndrome	1 gene	*PTEN*
Retinoblastoma	1 gene	*RB1*
Schwannomatosis	3 genes	*LZTR1, NF2, SMARCB1*
Skin tumors	15 genes	*BAP1, CDK4, CDKN2A, EPCAM, MITF, MLH1, MSH2, MSH6, NF1, PMS2, POT1, PTCH1, PTCH2, PTEN, SUFU*
Tuberous sclerosis complex	2 genes	*TSC1, TSC2*
Unspecific tumor syndromes	8 genes	*BAP1, CDKN1B, DICER1, NF1, PTEN, RB1, STK11, TP53*
von Hippel Lindau syndrome	1 gene	*VHL*

[1)] CNV analysis only for 5'-UTR region chr15:32,699,799-32,702,799 (hg38)

REPRODUCTIVE HEALTH

INFERTILITY TESTING

Disease/Indication	No. of genes	Gene/s
Androgen insensitivity	1 gene	*AR*
Azoospermia - step 1	-	AZF region
Azoospermia - step 2	6 genes	*AR, DMRT1, M1AP, NR5A1, TEX11, TEX14*
Congenital adrenal hyperplasia (CAH)	4 genes	*CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2*
Congenital bilateral absence of the vas deferens (CBAVD)	1 gene	*CFTR*
Disorders of sex development (DSDs)	16 genes	*AR, DHH, DMRT1, HSD17B3, HSD3B2, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX9, SRD5A2, SRY, TSPYL1, WNT4, WT1, WWOX*
Factor V Leiden	1 gene	*F5*
Fragile X-associated primary ovarian insufficiency (FXPOI)	1 gene	*FMR1*
FSH receptor deficiency	1 gene	*FSHR*
Hypogonadotropic hypogonadism / Kallmann syndrome	25 genes	*ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SOX10, SPRY4, TAC3, TACR3, WDR11*
Ovarian dysgenesis	5 genes	*BMP15, FSHR, MCM9, NR5A1, PSMC3IP*
Premature ovarian failure (POF) - step 1	1 gene	*FMR1*
Premature ovarian failure (POF) - step 2	18 genes	*BMP15, DIAPH2, ERCC6, ESR1, FIGLA, FOXL2, FSHR, GDF9, INHA, LHCGR, MCM9, NOBOX, NR5A1, PSMC3IP, SOHLH1, SOHLH2, STAG3, SYCE1*
Prothrombin G20210A	1 gene	*F2*
Recurrent pregnancy loss (M2/ANXA5 haplotype)	1 gene	*ANXA5*

About us

Medicover Genetics is a leading healthcare company specialising in genetic medicine, with more than 25 years of experience in genetics diagnostics. Medicover Genetics offers genetic testing services and genetic counselling, proprietary CE-IVD marked solutions and a unique Technology Transfer Platform which enables partners to perform high fidelity genetic tests in-house. With our services in over 50 countries across Europe, Asia, and Africa, we empower laboratories, healthcare professionals and patients to place genetics at the core of medical decisions. Committed to enhancing health and well-being, we provide meaningful, actionable diagnostic solutions, improving disease prognosis, clinical management, and therapy selection for genetic disorders. Our CAP-accredited, CLIA-, GMP- and ISO9001, 15189, and 13485 certified laboratories ensure the highest quality standards.

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