

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Attachment I – NGS PANELS

Panel name	Gene number	Gene list
CARDIOLOGY		
Cardiac arrhythmias	202	A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CDH2, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNT1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYOM1, MYOT, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLN, PMM2, PPA2, PRDM16, PRKG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK
Aortic / connective tissue diseases (basic panel)	64	ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTS4, ATP7A, B3GALT6, B3GLCT, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, GAA, HRAS, IPO8, KCNJ8, LOX, LTBP2, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1, TGFB2R, TNXB, ZNF469
Aortic / connective tissue diseases (extended panel)	121	ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTS4, AEBP1, ALDH18A1, ALPL, ARHGAP31, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BGN, BMP1, BMP4, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, EMILIN1, EOGT, FBLN5, FBNI, FBN2, FKBP10, FKBP14, FLCN, FLNA, FOXE3, GAA, GGCX, GZF1, IPO8, GORAB, GYPC, HRAS, IFITM5, KCNJ8, KIF22, LOX, LRP5, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, P3H1, PIEZ02, PLOD1, PLOD2, PLOD3, PLS3, PPIB, PRDM5, PRKG1, PTPN11, PYCR1, RBPJ, RET, RIN2, ROBO3, SERPINF1, SERPINH1, SGMS2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, SPARC, TAB2, TGFB2, TGFB3, TGFB1, TGFB2R, TMEM38B, TNXB, TPSAB1, VCAN, WNT1, ZNF469
Heritable pulmonar arterial hypertension	11	ACVRL1, BMPR1B, BMPR2, CAV1, CBLN2, EIF2AK4, ENG, FOXF1, KCNA5, KCNK3, SMAD9
Monogenic hypertension	27	AOC1, ASIC3, CLCNKB, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNH2, KCNJ1, KLHL3, NOS3, NR3C2, PDE3A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC12A1, SLC12A3, SCNN1B, SCNN1G, VHL, WNK1, WNK4
Left ventricular noncompaction	16	ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1
Arryhtmogenic cardiomyopathy	19	CASQ2, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TTN
Dilated miocardiopathy (basic panel)	35	ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DSG2, DMD, DSP, EMD, EYA4, FLNC, JPH2, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PSEN1, PSEN2, RBM20, RPL3L, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL
Dilated miocardiopathy (extended panel)	101	ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CAV3, CAVIN4, CHRM2, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, GATA6, GATAD1, GLA, HAMP, HCNA4, HFE, HFE2, IDH2, ILK, JPH2, JUP, KCND3, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MED12, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ1, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PKP2, PLEKHM2, PLN, PRDM16, PRKG2, PSEN1, PSEN2, RAB3GAP2, RAF1, RBM20, RPL3L, RYR2, SCN1B, SCN5A, SDHA, SGCA, SGCB, SGCD, SLC22A5, SLC40A1, SOS1, SYNE1, SYNE2, TAZ, TCAP, TFR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL, XK
Hypertrophic miocardiopathy (sarcomeric genes)	8	MYBPC3, MYH7, TNNT2, TNNI3, TPM1, ACTC1, MYL2, MYL3
Hypertrophic miocardiopathy (basic panel)	26	ACTC1, ACTN2, ALPK3, CSRP3, GLA, KRAS, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, NEXN, PLN, PRKG2, PTPN11, RAF1, RIT1, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR
Hypertrophic miocardiopathy (extended panel)	86	A2ML1, AARS2, ACADVL, ACTA1, ACTC1, ACTN2, AGL, ALPK3, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COA5, CPT2, CRYAB, CSRP3, DES, ELAC2, FHL1, FHOD3, FLNC, FOXRED1, FXN, GAA, GATA4, GLA, GLB1, GUSB, HRAS, JPH2, KCNQ1, KLF10, KRAS, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEXN, NF1, NRAS, OBSCN, PDLIM3, PLN, PRKG2, PTPN11, RAF1, RASA1, RIT1, RRAS, RYR2, SCN5A, SGCD, SLC22A5, SLC25A3, SOS1, SOS2, SPRED1, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, VCL

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		A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CPT2, CRYAB, CSR3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GDP1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNT1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LRRC10, LZTR1, MAP2K1, MAP2K2, MED12, MIB1, MYLCD, MRPL3, MRPL44, MRPS22, MT01, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAB2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK
Cardiomyopathy and arrhythmia	228	A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALM1, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, CPT2, CRYAB, CSR3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GLA, GLB1, GDP1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNT1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MED12, MIB1, MRPL3, MT01, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAB2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK
Cardiomyopathies	169	A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALM1, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, CPT2, CRYAB, CSR3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GLA, GLB1, GDP1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MED12, MIB1, MRPL3, MT01, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAB2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK
Sudden death	299	A2ML1, AARS2, ABCA1, ABCA12, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, AKT1, ALG10, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, ANO5, APOA2, APOA5, APOB, APOC2, APOC3, APOE, APTX, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CBL, CBS, CDH2, CHRM2, CLCF1, COA5, COA6, COL3A1, COL5A1, COL5A2, C002, COX15, COX6B1, CPT1A, CPT2, CREB3L3, CRLF1, CRYAB, CSR3, CTF1, CTNNA3, CYP27A1, DEPD5, DES, DLD, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, EFEMP2, ELAC2, ELN, EMD, ENPP1, EPG5, EPHX2, EYA4, FAH, FBN1, FBN2, FGF12, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNA, FLNC, FOXRED1, FXN, GAA, GATA4, GATA5, GATA6, GATAD1, GFM1, GHR, GJA1, GJA5, GLA, GLB1, GNPTAB, GDP1L, GPIHBP1, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, IKZF1, ILK, ITIH4, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNQ3, KCNT1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLRAP1, LIAS, LIPA, LIP1, LMF1, LMNA, LPL, LRP6, LRRC10, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MFAP5, MIB1, MYLCD, MRPL3, MRPL44, MRPS22, MT01, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NOTCH1, NPPA, NRAS, OBSCN, PCDH19, PCSK9, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PPA2, PPP1R17, PRDM16, PRKAG2, PRKG1, PRRT2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RRAS, RPL3L, RYR2, SCN10A, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN8A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SKI, SLC22A5, SLC25A3, SLC25A4, SLC2A1, SLC2A10, SLC40A1, SLMAP, SMAD3, SMAD4, SMAD6, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAB2, TBX20, TBX5, TCAP, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, USF1, VCL, XK
Aortic diseases	31	ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, GATA5, IPO8, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PL0D1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB1, TGFB1, TNXB
Brugada syndrome	42	ABCC9, AKAP9, ANK2, ANK3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, DPP6, FGF12, GPD1L, HCN4, IRX5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCNT3, KCNT4, KCNT5, KCNT6, KCNT7, KCNT8, KCNT9, KCNT10, KCNT11, KCNT12, KCNT13, KCNT14, KCNT15, KCNT16, KCNT17, KCNT18, KCNT19, KCNT20, KCNT21, KCNT22, KCNT23, KCNT24, KCNT25, KCNT26, KCNT27, KCNT28, KCNT29, KCNT30, KCNT31, KCNT32, KCNT33, KCNT34, KCNT35, KCNT36, KCNT37, KCNT38, KCNT39, KCNT40, KCNT41, KCNT42, KCNT43, KCNT44, KCNT45, KCNT46, KCNT47, KCNT48, KCNT49, KCNT50, KCNT51, KCNT52, KCNT53, KCNT54, KCNT55, KCNT56, KCNT57, KCNT58, KCNT59, KCNT60, KCNT61, KCNT62, KCNT63, KCNT64, KCNT65, KCNT66, KCNT67, KCNT68, KCNT69, KCNT70, KCNT71, KCNT72, KCNT73, KCNT74, KCNT75, KCNT76, KCNT77, KCNT78, KCNT79, KCNT80, KCNT81, KCNT82, KCNT83, KCNT84, KCNT85, KCNT86, KCNT87, KCNT88, KCNT89, KCNT90, KCNT91, KCNT92, KCNT93, KCNT94, KCNT95, KCNT96, KCNT97, KCNT98, KCNT99, KCNT100, KCNT101, KCNT102, KCNT103, KCNT104, KCNT105, KCNT106, KCNT107, KCNT108, KCNT109, KCNT110, KCNT111, KCNT112, KCNT113, KCNT114, KCNT115, KCNT116, KCNT117, KCNT118, KCNT119, KCNT120, KCNT121, KCNT122, KCNT123, KCNT124, KCNT125, KCNT126, KCNT127, KCNT128, KCNT129, KCNT130, KCNT131, KCNT132, KCNT133, KCNT134, KCNT135, KCNT136, KCNT137, KCNT138, KCNT139, KCNT140, KCNT141, KCNT142, KCNT143, KCNT144, KCNT145, KCNT146, KCNT147, KCNT148, KCNT149, KCNT150, KCNT151, KCNT152, KCNT153, KCNT154, KCNT155, KCNT156, KCNT157, KCNT158, KCNT159, KCNT160, KCNT161, KCNT162, KCNT163, KCNT164, KCNT165, KCNT166, KCNT167, KCNT168, KCNT169, KCNT170, KCNT171, KCNT172, KCNT173, KCNT174, KCNT175, KCNT176, KCNT177, KCNT178, KCNT179, KCNT180, KCNT181, KCNT182, KCNT183, KCNT184, KCNT185, KCNT186, KCNT187, KCNT188, KCNT189, KCNT190, KCNT191, KCNT192, KCNT193, KCNT194, KCNT195, KCNT196, KCNT197, KCNT198, KCNT199, KCNT200, KCNT201, KCNT202, KCNT203, KCNT204, KCNT205, KCNT206, KCNT207, KCNT208, KCNT209, KCNT210, 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KCNT947, KCNT948, KCNT949, KCNT949, KCNT950, KCNT951, KCNT952, KCNT953, KCNT954, KCNT955, KCNT956, KCNT957, KCNT958, KCNT959, KCNT959, KCNT9

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Galli-Galli disease	7	ADAM10, ADAR, KRT14, KRT5, POFUT1, POGLUT1, PSENEN
Epidermolysis bullosa	14	COL17A1, COL7A1, DSP, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5
Epidermolysis bullosa (extended panel)	31	ATP2C1, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSG4, DSP, DST, EXPH5, FERMT1, GRIP1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, SERPINB8, TGM5
Pachyonychia congenita	8	AAGAB, GJB6, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3
Palmoplantar keratoderma	25	AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJB2, GJB4, GJB6, JUP, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT14, KRT16, KRT17, LOR, MBTPS2, PKP1, SERPINB7, SLURP1, TRPV3, WNT10A
Rothmund-Thomson syndrome	46	ATM, BLM, BRCA2, BRIP1, CTC1, DDB2, DKC1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FAM111B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FERMT1, MAD2L2, NHP2, NOP10, PALB2, PARN, POLH, RAD51, RAD51C, RECQL4, RFWD3, RTEL1, SLX4, TERC, TERT, TINF2, UBE2T, USB1, WRAP53, WRN, XPA, XPC, XRCC2
IMMUNE SYSTEM DISEASES		
Autoinflammatory diseases	105	ADA, ADA2, ADAM17, ADAR, AICDA, AP1S3, AP3B1, ASA1, BLOC1S6, BTK, CARD14, CASP10, CASP8, CD27, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DDX58, DKC1, DOCK8, ELANE, EPCAM, FADD, FARSA, FAS, FASLG, FOXP3, G6PC3, HAX1, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL2RA, IL2RG, IL36RN, ITGB2, ITK, KRAS, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MEFV, MVK, MYO5B, NCF1, NCF2, NCF4, NEUROG3, NFAT5, NLRC4, NLRP1, NLRP2, NLRP3, NOD2, NRAS, OTULIN, PIK3CD, PIK3R1, PLCG2, PRF1, PRKCD, PSMB8, PSTPIP1, RAB27A, RAG1, RAG2, RASGRP1, RBC1, RNASEH2A, RNASEH2B, RNASEH2C, RTEL1, SAMHD1, SH2D1A, SI, SKIV2L, SLC29A3, SLC37A4, SLC7A7, STAT1, STAT3, STIM1, STX11, STXBP2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A1, TREX1, TTC37, TTC7A, UNC13D, WAS, XIAP, ZAP70
Inflammatory bowel disease	55	ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, EPCAM, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RG, ITGB2, LIG4, LRBA, MALT1, MEFV, MVK, MYO5B, NCF1, NCF2, NCF4, NEUROG3, NFAT5, NLRC4, NOD2, PIK3CD, PIK3R1, PLCG2, RAG1, RAG2, RTEL1, SH2D1A, SI, SKIV2L, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TTC37, TTC7A, WAS, XIAP, ZAP70
Recurrent fevers	35	ADAR, AP1S3, ASA1, CARD14, DDX58, ELANE, HAX1, IFIH1, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP2, NLRP3, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RBC1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A1, TNFRSF1A, TREX1
Primary immunodeficiencies	502	ABCD4, ACD, ACKR1, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGA, AICDA, AIRE, AK2, ALG1, ALG12, ALPI, AMT, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATG4A, ATM, ATP6AP1, AUH, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, BTK, C17orf62, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDC47, CEBPA, CEBPE, CFB, CFH, CFHR1 (exceto ex1), CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CFTR, CHD7, CHUK, CIB1, CIITA, CLCN7, CLPB, COG4, COPA, CORO1A, CR2, CREBBP, CSF2RA, CSF2RB, CSF3, CTC1, CTLA4, CTNNBL1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, EFL1, ELANE, EP300, EPG5, ERBIN, ERCC4, ETV6, EXT1, FAAP24, FADD, FAM111A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FBXL4, FCGR3A, FCCHO1, FCN3, FDX2, FERM1, FERM3, FH, FIBP, FM03, FNIP1, FOXN1, FOXP3, FPR1, FUT8, G6PC1, G6PC3, G6PD, GATA1, GATA2, GCSH, GF1, GINS1, GLDC, GSS, GTF2H5, HAVCR2, HAX1, HELLS, HMOX1, HPS5, HTRA2, HWE1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IRAK1, IRAK4, IREB2, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, IVD, JAGN1, JAK1, JAK3, KDM6A, KMT2A, KMT2D, KRAS, LAMTOR2, LAT, LCK, LCP2, LEF1, LIG1, LIG4, LMBRD1, LPIN2, LRBA, LSM11, LYST, MAD2L2, MAGT1, MALT1, MAP3K14, MAPK8, MASP2, MCM10, MCM4, MDM4, MECOM, MEFV, MKL1, MLH1, MMAA, MMAB, MMACHC, MOGS, MPL, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MTR, MUT, MVK, MYD88, MYSM1, NBAS, NBN, NCFC2, NCFC4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC3, NLRC4, NLRP1, NLRP2, NLRP3, NLRP7, NOD2, NOP10, NOS2, NRAS, NSMC3, OAS1, OPA3, ORAI1, OSTM1, OTULIN, PACS2, PALB2, PARN, PAX1, PCCA, PCCB, PEPD, PGM3, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLEKHM1, PMM2, PMS2, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3C, POLR3F, POMP, PRDM5, PRDX1, PRF1, PRKCD, PRKDC, PSEN1, PSENEN, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RANBP2, RASGRP1, RBC1, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOG, RHOC, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPL11, RPL18, RPL26, RPL5, RPS14, RPS19, RPS7, RPSA, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SEMA3E, SERPING1, SH2D1A, SH3BP2, SH3KBP1, SKIV2L, SLC19A2, SLC29A3, SLC33A1, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A5, SLC46A1, SLC7A7, SLX4, SMARCAL1, SMARCD2, SNX10, SOCS1, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK11, STK4, STN1, STX11, STXBP2, TAFAZZIN, TALDO1, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX21, TCF3, TCIRG1, TCN2, TDP2, TERT, TET2, TFR2, TGFB1, TGFB2, THBD, THG1L, TICAM1, TINF2, TIRAP, TLR3, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF9, TNFSF11, TNFSF12, TNFSF13, TONS1, TOP2B, TP53, TPP1, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBA1, UBE2T, UGT1A1, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS33A, VPS45, WAS, WDR1, WFS1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZNF341
Autoimmune lymphoproliferative syndrome (ALPS)	19	ADA2, AICDA, CASP10, CASP8, CD40LG, CTLA4, FADD, FAS, FASLG, ITK, KRAS, LRBA, MAGT1, NRAS, PRKCD, RASGRP1, SH2D1A, STAT3, WAS
Complement disorders and hemolytic uremic syndrome	12	C3, CD46, CFB, CFH, CFHR1, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD, VTN
METABOLIC DISEASES		
Familial dyslipidemia / Hypercholesterolemia	27	ABCA1, ABCG5, ABCG8, APOA2, APOA5, APOB, APOC2, APOC3, APOE, APTX, CREB3L3, CYP27A1, EPHX2, GHR, GPD1, GPIHBP1, ITIH4, LDLR, LDLRAP1, LIPA, LIPI, LMF1, LPL, LRP6, PCSK9, PPP1R17, USF1
Tangier disease	2	ABCA1, LCAT

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Mitochondrial diseases (nuclear genes)	373	AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, AC02, ACOX1, ACSF3, ACSL4, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH1A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMT, APOPT1, ATIC, ATP5A1, ATP5E, ATP7B, ATPAF2, ATXN2, AUH, BAX, BCKDHA, BCKDHB, BCKDK, BCL2, BCS1L, BOLA3, BRIP1, BTD, C12orf65, CA5A, CASP8, CAT, CAVIN1, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COASY, COMT, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRBN, CYB5A, CYB5R3, CYC1, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, EHHADH, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FECH, FH, FKBP10, FOXRED1, FTH1, FXN, GARS, GATM, GCDH, GCSE, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, ISCA2, ISCU, IVD, KARS, KIF1B, KRT5, L2HGDH, LARS2, LIAS, LIPT1, LONP1, LRPPRC, LYRM4, LYRM7, MAOA, MAOB, MARS2, MCC1, MCC2, MCEE, MFN2, MGME1, MICU1, MIP, MLH1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSRB3, MTFTM, MTO1, MTPAP, MTRR, MUT, MUTYH, NADK2, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NNT, NTHL1, NUBPL, OAT, OGDH, OGG1, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PNPLA8, PNPO, PNPT1, POLG, POLG2, PPM1K, PPOX, PRODH, PTGS1, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS, RARS2, RDH11, REQL4, RMND1, RNASEH1, RNASEL, RPIA, RPL35A, RPS14, RRM2B, SARDH, SARS2, SC01, SC02, SCP2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A44, SLC37A4, SLC9A6, SNAP29, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, STOM, SUCLA2, SUCLG1, SUGCT, SUO, SURF1, TACO1, TARS2, TCIRG1, TIMM44, TIMM8A, TK2, TMEM70, TMLHE, TP1, TPK1, TRMU, TRNT1, TSCF19, TUBB3, TUFM, TTC19, TXNRD2, TYMP, UNG, UQCQC2, UQCRCB, UQCRCQ, VARS2, WDR81, WFS1, XPNPEP3, YARS2
Glycogenoses	30	AGL, ALDOA, ALDOB, AMPD1, CPT2, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4
Hyperammonemia	4	CA5A, CPS1, NAGS, OTC
Nonketotic hyperglycinemia	2	AMT, GLDC
Leukodystrophies	92	ABCD1, ACADS, AC02, ACOX1, ADAR, ADGRG1, ADSL, ALDH3A2, AP4B1, ARSA, ASPA, B3GALNT2, COX7B, CSF1R, CYP27A1, DARS, DARS2, DDOST, DHFR, DNM1L, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ERCC6, FA2H, FAM126A, FKRP, FKTN, FOLR1, FOXG1, FUCA1, GALC, GBE1, GFAP, GFM1, GJA1, GJB1, GJC2, GLUL, HEPACAM, HSD17B4, HSPD1, HTRA1, IDUA, ISCA2, KCNT1, L2HGDH, LAMA2, LARGE1, LMNB1, MARS2, MCOLN1, MEF2C, MLC1, MLYCD, MTPP, MUT, NOTCH3, PAH, PEX7, PHGDH, PLP1, POLG, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PTEN, PSAP, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SLC16A2, SLC17A5, SLC25A12, SLC33A1, SOX10, STXBP1, SUMF1, TREX1, TUBB4A, TYMP, UBE2A

ENDOCRINOLOGY

Hypogonadotropic hypogonadism (Kallmann syndrome)	33	ANOS1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NR0B1, NSMF, POLR3B, PROK2, PROKR2, SEMA3A, SEMA3E, SEMA7A, SOX10, SPRY4, SRA1, TAC3, TACR3, WDR11
Hypomagnesemia	24	BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, EGFR, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, MNGT1, NIPA2, PCBD1, SARS2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7
MODY	15	ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, SLC16A1
Obesity	44	ADRB2, ADRB3, AGRP, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CARTPT, CEP290, DYRK1B, ENPP1, FFAR4, FTO, GHRL, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, MRAP2, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, SDCCAG8, SIM1, SLC6A14, TRIM32, TTC8, UCP1, UCP2, UCP3, VPS13B, WDPCP
Nonsyndromic obesity	25	ADRB2, ADRB3, AGRP, ALMS1, ARL6, CARTPT, DYRK1B, ENPP1, FFAR4, FTO, GHRL, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SLC6A14, UCP1, UCP2, UCP3
Syndromic obesity	20	BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CEP290, GNAS, MAGEL2, MKKS, MKS1, NTRK2, PHF6, SDCCAG8, SIM1, TRIM32, TTC8, VPS13B, WDPCP
Pseudohypoaldosteronism type 2	4	CUL3, KLHL3, WNK1, WNK4
Central precocious puberty	4	KISS1, KISS1R, MKRN3, DLK1
Insulin resistance	12	ABCC8, AGPAT2, BSCL2, CAVIN1, GCK, GLUD1, HADH, HNF4A, IGF1R, INSR, KCNJ11, UCP2

GASTROENTEROLOGY

Familial intrahepatic cholestasis	3	ABCB11, ABCB4, ATP8B1
Neonatal intrahepatic cholestasis	58	ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, ATP7B, ATP8B1, BAAT, CC2D2A, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, GALE, GALK1, GALM, GALT, GBA, GBE1, HADHA, HNF1B, HSD3B7, INV5, JAG1, LIPA, MPI, MPV17, MVK, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX6, PKHD1, POLG, SLC25A13, SMPD1, TALD01, TJP2, TRMU, UGT1A1, VIPAS39, VPS33B
Hemochromatosis	7	FTH1, FTL, HAMP, HFE, HFE2, SLC40A1, TFR2
Hereditary pancreatitis	8	CASR, CFTR, CLDN2, CPA1, CTRC, SPINK1, PRSS1, PRSS2

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Hereditary porphyria	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS
HEMATOLOGY		
Diamond-Blackfan anemia	20	GATA1, RPL5, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL36, RPS7, RPS10, RPS15, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, TSR2
Fanconi anemia	21	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2
Vascular and lymphatic malformations	63	ACVRL1, ADAMTS13, ADAMTS3, AKT1, AKT3, ANTXR1, ATM, BMPR2, BRAF, CAV1, CBL, CCBE1, CCM2, DOCK6, ELM02, ENG, EPHB4, FAT4, FLT4, FOXC2, GATA2, GDF2, GJC2, GLMN, GNAQ, KCNK3, KDR, KIF11, KRAS, KRIT1, LZTR1, MAP2K1, MAP3K3, MTOR, NF1, NF2, NRAS, PDCD10, PDGFRB, PIEZ01, PIK3CA, PTEN, PTPN11, RAF1, RASA1, RIT1, SHOC2, SMAD2, SMAD3, SMAD4, SMARCB1, SOS1, SOX18, SPRED1, STAMBMP, TEK, TGFB2, TGFB3, TGFB1R, TGFB2R, TSC1, TSC2, VEGFC
Hemophagocytic syndrome	7	DCLRE1C, PRF1, STX11, STXBP2, RAG1, RAG2, UNC13D
Hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease)	5	ACVRL1, ENG, GDF2, RASA1, SMAD4
Glanzmann thrombasthenia	2	ITGA2B, ITGB3
NEFROLOGY		
Renal tubular acidosis	3	ATP6V0A4, ATP6V1B1, SLC4A1
Cystinuria	2	SLC3A1, SLC7A9
Cholestasis	58	ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, GALE, GALM, GALT, GBA, HADHA, HNF1B, HSD3B7, JAG1, KIF12, LIPA, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX12, PEX26, PEX6, PKHD1, POLG, RINT1, SERPINA1, SLC25A13, SMPD1, TALDO1, TJP2, TRMU, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, YARS, ZFYVE19
Chronic renal disease in the young	85	ACE, ACTG2, ACTN4, AGT, AGTR1, AMN, ANKS6, ARHGDIA, C3, CD46, CEP164, CFB, CFH, CFHR5, CFI, CHD7, CHRM3, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CTNS, CUBN, DGKE, DSTYK, EYA1, FAN1, FRAS1, FREM1, FREM2, GATA3, GLA, GLI3, GRIP1, HNF1B, HPSE2, INF2, INV5, ITGA3, ITGA8, LAMB2, LMX1B, LRIG2, MUC1, MYH9, MYO1E, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NUP107, NUP93, OFD1, PAX2, PKD1, PKD2, PKHD1, PLCE1, REN, RPGRIP1L, RRM2B, SALL1, SARS2, SCARB2, SGPL1, SIX5, SMARCAL1, TBX18, TMEM67, TRAP1, TRIM8, TRPC6, TSC1, TSC2, TTC21B, UMOD, VHL, VIPAS39, VPS33B, WT1, XPNPEP3
Polycystic kidney disease	6	DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1
Polycystic kidney disease (extended panel)	32	ABCC8, ALG8, BICC1, BLK, COL4A1, DNAJB11, DZIP1L, GANAB, GCK, HNF1B, HNF4A, INS, INV5, KCNJ11, KLF11, LRP5, NEUROD1, NOTCH2, NPHP3, OFD1, PAX4, PDX1, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63, TSC1, TSC2, UMOD, VHL
Familial renal glucosuria	2	SLC2A2, SLC5A2
Hyperoxaluria, primary	3	AGXT, GRHPR, HOGA1
Renal lithiasis	30	ADCY10, AGXT, AP2S1, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, OCRL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1
Autosomal dominant interstitial nephritis	4	HNF1B, REN, SEC61A1, UMOD We also have available the analysis of the insC in the MUC1 gene and MLPA analysis in the HNF1B gene - see Attachment II.
Tubulointerstitial nephritis	23	ADAMTS9, ANKS6, CEP164, CEP83, DCDC2, DNAJB11, GATM, GLIS2, HNF1B, INV5, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, REN, SEC61A1, TMEM67, TTC21B, UMOD, WDR19, XPNPEP3, ZNF423
Nephrocalcinosis	45	ADCY10, AGK, AGXT, AP2S1, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, FGF23, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, OCRL, PHEX, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC9A3, SLC9A3R1, STRADA, TRPM6, VDR, XDH, ZNF365
Nephronophthisis	17	ANKS6, CEP164, CEP290, GLIS2, IFT172, INV5, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423
Alport syndrome	4	COL4A3, COL4A4, COL4A5, COL4A6
Bartter syndrome and Gitelman syndrome	6	BSND, CLCNKA, CLCNKB, KCNJ1, SLC12A1, SLC12A3
Liddle syndrome	2	SCNN1B, SCNN1G
Nephrotic syndrome	58	AACTN4, AMN, APOE, APOL1, ARHGDIA, CD151, CD2AP, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DAAM2, DGKE, DLC1, FAT1, FN1, GLA, GON7, INF2, ITGA3, ITSN1, LAGE3, LAMB2, LCAT, LMX1B, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP85, NUP93, OCRL, OSGE, PAX2, PDSS2, PLCE1, PODXL, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TRIM8, TRPC6, TTC21B, WDR73, WT1, YRDC
Tubulopathies	48	AP2S1, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, CTNS, CUL3, CYP24A1, FAH, GATM, GNA11, HNF1B, HNF4A, KCNJ1, KCNJ10, KCNJ16, KLHL3, MAGED2, NR3C2, REN, RRAGD, SARS2, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC4A1, SLC4A4, SLC5A2, TRPM6, UMOD, VIPAS39, VPS33B, WNK4
NEUROLOGY		

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Familial amyloid angiopathy	27	APOE (alelo e4), APP, CHCHD10, CHMP2B, COL4A1, COL4A2, CSF1R, CST3, CTC1, DCTN1, ITM2B, GLA, GRN, GSN, HTR1A, MAPT, NOTCH3, PRNP, PSEN1, PSEN2, SNCA, SNCB, SQSTM1, TARDBP, TBK1, TREX1, TTR
Recessive ataxias	17	AFG3L2, ANO10, ATM, COQ8A, MRE11, MTPAP, MTTP, PIK3R5, POLG, SACS, SETX, SIL1, SPTBN2, SYNE1, SYT14, TDP1, ZNF592
Ataxias (extended panel)	158	ABC7, ABHD12, ACO2, AFG3L2, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP1A3, ATP2B3, ATP8A2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C5orf42, CA8, CACNA1A, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COASY, COQ8A, COX20, CP, CSTB, CWF19L1, CYP27A1, CYP2U1, DNAJC19, DNMT1, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, MARS2, MKKS, MKS1, MME, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NOL3, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PAX6, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PPP2R2B, PRKCG, PRRT2, RNF216, RPGRIP1L, RUBCN, SACS, SERAC1, SETX, SIL1, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A2, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDR, WDPPC, WDR81, WFS1, WWOX, ZFYVE26, ZNF423
Spinal muscular atrophy	31	AR, ASA1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC8, FBXO38, GARS, HSPB1, HSPB8, HSPB3, IGHMBP2, PLEKHG5, RAX2, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, TBCE, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS
Cerebral cavernous malformations	3	CCM2, KRIT1, PDCD10
Dystonias	82	ADAR, ADCY5, ANO3, ATP13A2, ATP1A3, ATP7B, BTD, C19orf12, CACNA1B, COASY, COL4A1, CP, CTSF, DCAF17, DDC, DLAT, DRD2, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FBXO7, FTL, GALC, GAMT, GATM, GCDH, GCH1, GLB1, GNAL, HEXA, HPRT1, KMT2B, LIAS, LRRK2, NPC1, NPC2, PANK2, PARK7, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PINK1, PLA2G6, PLP1, PNKD, POLG, PRKN, PRKRA, PRRT2, PTS, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SGCE, SLC19A3, SLC20A2, SLC2A1, SLC30A10, SLC6A3, SLC6A8, SPR, SUCLA2, SYNJ1, TAF1, TH, THAP1, TIMM8A, TOR1A, TPK1, TREX1, TUBB4A, VPS13A, WDR45, XK
Limb-girdle muscular dystrophies	28	ANO5, BVES, CAPN3, CAV3, DES, DNAJB6, DYSF, FKRP, FKTN, GMPPB, ISPD, HNRNPD1, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TRAPP11, TRIM32, TTN
Alzheimer disease and frontotemporal dementia	15	APOE, APP, CHMP2B, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, TARDBP, TREM2, VCP
Charcot-Marie-Tooth disease	63	AARS, AIFM1, ATL1, ATP7A, BSCL2, C12orf65, COX6A1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, ELP1, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SH3TC2, SLC12A6, SPTLC1, SPTLC2, TRIM2, TRPV4, TTR, WNK1, YARS
Moyamoya disease	3	ACTA2, GUCY1A3, RNF213
Parkinson disease (basic panel)	25	ATP13A2, ATP6AP2, CHCHD2, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, HTRA2, LRRK2, PARK7, PINK1, PLA2G6, PODXL, PRKN, RIC3, SLC30A10, SLC6A3, SNCA, SYNJ1, TAF1, TMEM230, UCHL1, VPS13C, VPS35
Parkinson disease (extended panel)	47	ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, COQ2, DNAJC13, DNAJC6, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, NR4A2, PARK7, PDGFB, PDGFRB, PINK1, PLA2G6, PODXL, PRKN, PRKRA, RAB39B, RIC3, SLC18A2, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCAIP, SPR, SYNJ1, TAF1, TBP, TH, TMEM230, TRPM7, UCHL1, VPS13A, VPS35, XPR1
Connective tissue diseases	51	ABL1, ACTA2, ADAMTS2, ADAMTS21, ARHGAP36, ATP7A, B3GALT6, B4GALT7, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL5A3, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRLF1, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM2, DYSF, ECEL1, EMD, ENO3, EPG5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLNB, FLMN, GAA, GBA, GBE1, GFPT1, GLDN, GLE1, GMPPB, GNE, GYG1, GYS1, HADHA, HADHB, HSPB1, HSPB8, HSPG2, INPP5K, IRF6, ISC1, ISPD, ITGA7, KAT6B, KBTBD13, KLHL40, KLHL41, KLHL7, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, LRP4, MAP3K20, MATR3, MEGF10, MICU1, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYO18B, MYOT, NALCN, NEB, ORAI1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZ02, PLEC, PLOD1, PLOD2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRG4, PRKAG2, PYGM, RAPSN, RBCK1, RIPK4, RRM2B, RYR1, SCARF2, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SKI, SLC18A3, SLC22A5, SLC25A4, SLC5A7, SMAD3, SMAD4, SPEG, SQSTM1, STAC3, STIM1, SUCLA2, SYNE1, TCAP, TGFB2, TGFB3, TGFB1, TGFB2, TIA1, TK2, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, ZC4H2, ZMPSTE24
Neuromuscular diseases	207	

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS



Neuromuscular and muscular diseases	247	ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACTG2, ACVR1, ADAMTS10, ADGRG6, AGL, AGRN, ALDOA, AMPD1, ANO5, ANTXR2, ASXL1, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRN, CHRNE, CHRNG, CHST14, CLCN1, CNBP, CNTN1, CNTNAP1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT1B, CPT2, CRLF1, CRYAB, DAG1, DES, DMD, DNA2, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYSF, ECEL1, EMD, ENO3, EPG5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLNB, FLNC, GAA, GBA, GBE1, GFPT1, GLDN, GLE1, GMPPB, GNE, GOSR2, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HNRNPA1, HNRNPA2B1, HRAS, HSPB1, HSPB8, HSPG2, INPP5K, IRF6, ISCU, ISPD, ITGA7, KAT6B, KBTBD13, KLHL40, KLHL41, KLHL7, KLHL9, KY, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, LRP4, MAMLD1, MAP3K20, MATR3, MEGF10, MICU1, MSTN, MTM1, MTMR14, MUSK, MYBPC1, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYO18B, MYOT, NALCN, NEB, OPA1, OPA3, ORAI1, PABPN1, PDHA1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PLD1, PLD2, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRG4, PRKAG2, PUS1, PYGM, RAPSN, RBC1, RIPK4, RRM2B, RYR1, SCARF2, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SKI, SLC16A1, SLC18A3, SLC22A5, SLC25A20, SLC25A4, SLC5A7, SMAD3, SMAD4, SPEG, SQSTM1, STAC3, STIM1, SUCLA2, SYNE1, TAZ, TCAP, TGFB2, TGFB3, TGFB1, TGFB2, TIA1, TK2, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, YARS2, ZC4H2, ZMPSTE24
Epileptic encephalopathy	49	ACY1, ADSL, ALDH7A1, AMT, ARHGEF9, ARX, CDKL5, CNTNAP2, CPT2, FOLR1, FOXG1, GABRG2, GAMT, GCSH, GLDC, GRIN2A, GRIN2B, KCNA2, KCNA10, KCNQ2, MAPK10, MECP2, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC19A3, SLC25A22, SLC2A1, SLC9A6, SPTAN1, STXBP1, TBCE, TCF4, TREX1, UBE3A, ZEB2
Migraine	10	ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1
Nocturnal frontal lobe epilepsy	6	CHRNA2, CHRNA4, CHRN2, CRH, DEPDC5, KCNT1
Amyotrophic lateral sclerosis	5	FUS, SOD1, TARDBP, SETX, VCP
Juvenile amyotrophic lateral sclerosis	3	ALS2, ERLIN2, SETX
Tuberous sclerosis	2	TSC1, TSC2
Peripartum intraventricular hemorrhage	5	COL4A1, COL4A2, GLA, HTRA1, TREX1
Myopathies	156	ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACTG2, ACVR1, AGL, AGRN, AMPD1, ANO5, ATP2A1, B4GAT1, BAG3, BIN1, CAPN3, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRN, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT1B, CPT2, CRYAB, DAG1, DES, DNA2, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM3, DYSF, ENO3, ETFA, ETFB, ETFDH, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GBE1, GFPT1, GNE, GOSR2, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HNRNPA1, HNRNPA2B1, HRAS, ISCU, ISPD, ITGA7, KBTBD13, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, MAMLD1, MATR3, MEGF10, MICU1, MSTN, MTM1, MTMR14, MUSK, MYF6, MYH14, MYH2, MYH3, MYH7, MYOT, NEB, OPA1, OPA3, ORAI1, PABPN1, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PLEC, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMT1, POMT2, PRKAG2, PUS1, PYGM, RAPSN, RBC1, RRM2B, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC22A5, SLC25A20, SPEG, STAC3, STIM1, SUCLA2, TAZ, TCAP, TIA1, TK2, TNNT1, TPM2, TPM3, TRIM32, TTN, TWNK, TYMP, VCP, VMA21, YARS2
Metabolic myopathies and Rabdomyolysis	64	ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGL, AHCY, ALDOA, AMPD1, ANO5, CACNA1S, CAV3, CPT2, DMD, DYSF, ECHS1, ENO3, ETFA, ETFB, ETFDH, FKRP, FKTN, GAA, GBE1, GYG1, GYS1, GYS2, HADHA, HADHB, HSPB8, ISCA2, ISCU, KARS, LAMP2, LDHA, LPIN1, MPV17, MRM2, MTO1, NDUFB8, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, POLG, POLG2, PRKAG2, PYGM, RBC1, RRM2B, RYR1, SIL1, SLC22A5, SLC25A1, SLC25A20, SUCLA2, TANGO2, TAZ, TK2, TSFM, TYMP, VMA21
Myofibrillar myopathies	8	BAG3, CRYAB, DES, DNAJB6, FHL1, FLCN, LDB3, MYOT
Nondystrophic myopathies	11	ATP2A1, CACNA1A, CACNA1S, CAV3, CLCN1, HINT1, HSPG2, KCNA1, KCNE3, KCNJ18, SCN4A
Neurodegeneration with brain iron accumulation disorders (NBIA)	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
Neuropathies	69	AARS, ABHD12, AIFM1, ATL1, ATP7A, BSCL2, COX6A1, ELP1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, EXOSC8, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, RETREG1, SBF2, SCN10A, SCN1A, SCN9A, SH3TC2, SLC12A6, SLC52A3, SLC5A7, SMN1, SPTLC1, SPTLC2, SYT2, TFG, TRPV4, TTR, TWNK, VCP, WNK1, YARS
Progressive external ophthalmoplegia	16	DGUOK, DNA2, MGME1, MFN2, MPV17, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP
Periodic paralysis	11	ATP1A2, ATP1A3, CACNA1S, DNM1L, KCNJ12 (=KCNJ18), KCNJ2, KCNJ5, MCM3AP, RYR1, SCN4A, SLC12A3
Spastic paraparesis	78	ADAR, ALDH18A1, ALDH3A2, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATAD3A, ATL1, ATP13A2, ATP2B4, B4GALNT1, BICD2, BSCL2, C12orf65, C19orf12, CAPN1, CCT5, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DNM2, DSTYK, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR2, GAD1, GBA2, GJC2, GRID2, HSPD1, IBA57, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TTR, TUBB4A, UCHL1, USP8, VAMP1, VPS37A, WASHC5, WDR48, ZFR, ZFYVE26, ZFYVE27
Fahr syndrome	4	PDGFRB, PDGFB, SLC20A2, XPR1

OFTALMOLOGY

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Congenital cataracts	41	AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, EYA1, FAM126A, FOXE3, FYCO1, GALK1, GBA2, GCNT2, GJA3, GJA8, HSF4, LIM2, MAF, MIP, MIR184, NHS, P3H2, PAX6, PITX3, PXDN, SIL1, SLC16A12, SLC33A1, TDARD7, VIM
Macular dystrophies	28	ABC4, BEST1, C1QTNF5, CDH3, CERKL, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RP1L1, RPGR, RS1, TIMP3
Retinitis pigmentosa	160	ABC4, ABHD12, ADAM9, ADGRA3, AGBL5, AIPL1, ARL2BP, ARL3, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CC2D2A, CDH23, CDHR1, CEP290, CEP78, CERKL, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNM4, CRB1, CRX, CYP4V2, DHDDS, DHX38, DTHD1, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GDF6, GNAT1, GNPTG, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, INPP5E, INVS, IQCB1, KCNJ13, KCNV2, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, MKKS, MVK, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OFD1, OPN1LW, OTX2, PCDH15, PDE6A, PDE6B, PDE6C, PDE6H, PEX1, PEX2, PEX26, PEX7, PHYH, PITPNM3, PLA2G5, POC1B, POMGNT1, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RAB28, RAX2, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRIM32, TRNT1, TRPM1, TTC8, TTLL5, TTPA, TUB, TULP1, UNC119, USH1C, USH2A, WDR19, WFS1, WHRN, ZNF408, ZNF513

ONCOLOGY

BRCA1 and BRCA2 genes – Pack 1 [insAlu (blood) + NGS (blood or FFPE) + MLPA (blood)]	2	BRCA1, BRCA2* *Includes the research of the founder mutation c.156_157insAlu in the BRCA2 gene and MLPA in the blood sample. Includes the confirmation of the mutation's origin (germinal or somatic). Note: for somatic mutations, in the tumor sample, we request the tumor infiltration percentage
BRCA1 and BRCA2 genes – Pack 2 [insAlu (blood) + NGS (blood or FFPE)]	2	BRCA1, BRCA2* *Includes Portuguese founder mutation (c.156_157insAlu) research in the BRCA2 gene in the blood sample. Includes the confirmation of the mutation's origin (germinal or somatic). Note: for somatic mutations, in the tumor sample, we request the tumor infiltration percentage
Hereditary breast/ovarian cancer	30	ATM, BARD1, BLM, BRCA1, BRCA2*, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, FANCC, FANCM, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, SLX4, STK11, TP53, XRCC2 *Includes Portuguese founder mutation (c.156_157insAlu) research in the BRCA2 gene
Hereditary colorectal cancer	39	APC, ATM, AURKA, AXIN2, BLM, BMPR1A, BRCA1, BRCA2*, BRIP1, CCND1, CDH1, CHEK2, EPCAM, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH6, MSH3, MUTYH, NTHL1, ODC1, PIF1, PLA2G2A, POLD1, POLE, PMS2, PTEN, RBL1, RNF43, RPS20, SMAD4, SMAD7, STK11, TEL02, TGFB2R, TP53, XAF1 *Includes Portuguese founder mutation (c.156_157insAlu) research in the BRCA2 gene
Hereditary pancreatic cancer	18	APC, ATM, BMPR1A, BRCA1, BRCA2*, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53 *Includes Portuguese founder mutation (c.156_157insAlu) research in the BRCA2 gene
Hereditary prostate cancer	20	ATM, BRCA1, BRCA2*, HOXB13, CHEK2, RAD51C, RAD51D, PALB2, ATR, NBN, GEN1, MLH1, MSH2, MSH6, PMS2, MRE11, BRIP1, FAM175A, EPCAM, TP53 *Includes Portuguese founder mutation (c.156_157insAlu) research in the BRCA2 gene
Familial malignant melanoma	5	BAP1, CDKN2A, CDK4, MITF, POT1
Paraganglioma and pheochromocytoma	18	EGLN1, EPAS1, FH, IDH1, KIF1B, MAX, MDH2, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL
Familial paraganglioma	3	SDHD, SDHB, SDHC
Lynch syndrome	4	MLH1, MSH2, MSH6, PMS2
Hereditary renal tumors (basic panel)	10	AKT1, BAP1, FLNC, FH, MET, PTEN, PIK3CA, STK11, SDHB, VHL
Hereditary renal tumors (extended panel)	25	BAP1, CDC73, CHEK2, DICER1, DIS3L2, EPCAM, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1

OTORRINOLARINGOLOGY

Syndromic and nonsyndromic hearing loss	241	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BMP5, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CDC14A, CDC6, CDH23, CDKN1C, CDT1, CEACAM16, CEMIP, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DFNA5, DFNB59, DHODH, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, ECE1, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, ELMOD3, EPS8, EPS8L2, ERCC2, ERCC3, ESPN, ESRRB, EVC, EYA1, EYA4, FAS, FGF10, FGF3, FGFR3, FOXI1, FOXI3, FRAS1, FREM2, GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB4, GJB6, GNA13, GPSM2, GRHL2, GRIP1, GRXCR1, GRXCR2, GSC, GSTP1, HAAO, HAL, HARS2, HGF, HMX1, HOMER2, HOXA2, HOXB1, HSD17B4, HSPA9, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KDM6A, KIT, KITLG, KMT2D, LARS2, LEMD3, LHFLP5, LHX1, LOXHD1, LRP2, LRTOMT, MANBA, MARVELD2, MASP1, MCM2, MET, MIR96, MITF, MSRB3, MTAP, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO1F, MYO3A, MYO6, MYO7A, NARS2, NDP, NF2, NKX3-2, NLRP3, NR2F1, OFD1, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTOR, P2RX2, PAX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PEX7, PHYH, PLCB4, PMP22, PNPT1, POLR1A, POLR1C, POLR1D, PORCN, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, RPS28, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC4A11, SLTRK6, SMPX, SNAI2, SOX10, SOX2, SPATA5, SPINK5, STRC, SYNE4, TBC1D24, TBL1X, TBX1, TCF21, TCOF1, TECTA, TFAP2A, TFCP2, TIMM8A, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TMPRSS5, TNC, TPRN, TRIOPB, TSPEAR, TWNK, TWSG1, TYR, USH1C, USH2A, USH2A, WFS1, WHRN
Usher and Alström syndrome	14	ADGRV1, ALMS1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN

REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

Usher syndrome	13	<i>ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN</i>
Waardenburg syndrome	7	<i>EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR</i>
PEDIATRICS		
Distal arthrogryposis	9	<i>ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2, TPM2, TNNT2, TNNT3</i>
Craniosynostosis	42	<i>ALPL, ALX3, ALX4, BMP4, EDN3, EDNRB, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, IL11RA, IMPAD1, IRX5, MASP1, MEGF8, MITF, MSX2, NOG, PAX3, POR, RAB23, RECQL4, RET, SCARF2, SKI, SOX10, TCF12, TGFB1, TGFB2, TMCO1, TTR, TWIST1, WDR19, WDR35</i>
Multiple epiphyseal dysplasias	8	<i>COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, UFSP2</i>
Skeletal dysplasias	106	<i>ACP5, ADAMTS10, ADAMTSL2, AGPS, ALPL, ANKH, ARSE, B3GALT6, BMP1, BMPR1B, CA2, CANT1, CDC6, CDKN1C, CDT1, CHST3, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CRTAP, CSPP1, CTSK, CUL7, CYP27B1, DHCR24, DLL3, DVL1, DYM, DYNC2H1, EBP, EIF2AK3, EMILIN1, ENPP1, ESCO2, EVC, EVC2, FAM20C, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GDF5, GNPA1, HSPG2, IFT140, IFT172, IFT80, IHH, IKBKG, KAT6B, LBR, LIFR, LMX1B, LRP5, LTBP2, MATN3, MMP9, NEK1, NPR2, OBSL1, ORC1, ORC4, ORC6, P3H1, PAPSS2, PCNT, PEX7, PHEX, PLOD2, PPIB, PTH1R, ROR2, RUNX2, SBDS, SERPIN1F, SERPINH1, SHOX, SLC26A2, SLC34A3, SLC39A13, SMAD4, SMARCAL1, SOX9, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TRAPP2, TRPV4, TTC21B, VDR, WDR19, WDR35, WISP3, WNT5A</i>
Hemifacial microsomia	13	<i>CHD7, DHODH, EFTUD2, EYA1, GNAI3, PLCB4, POLR1C, POLR1D, SALL1, SF3B4, SIX1, SIX5, TCOF1</i>
Osteogenesis Imperfecta	2	<i>COL1A1, COL1A2</i>
Hypophosphatemic rickets	11	<i>ALPL, CLCN5, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR</i>
Bardet-Biedl syndrome	20	<i>ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP</i>
Cornelia de Lange syndrome	5	<i>HDAC8, NIPBL, RAD21, SMC1A, SMC3</i>
Klippel-Feil syndrome	3	<i>GDF3, GDF6, MEOX1</i>
Noonan syndrome / RASopathies	25	<i>A2ML1, BRAF, CBL, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1</i>
Treacher Collins syndrome	3	<i>TCOF1, POLR1C, POLR1D</i>
PNEUMOLOGY		
Surfactant dysfunction	7	<i>ABCA3, CSF2RA, CSF2RB, SFTP1A, SFTP1B, SFTP1C, SFTP1D</i>
Pulmonary disease (extended panel)	67	<i>ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRN1B, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DCK1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI2, DNAL1, EDN3, EFEMP2, ELMOD2, ELN, FBLN5, FLCN, FOXF1, GAS8, GLRA1, HPS1, HPS4, ITGA3, LTBP4, MECP2, NAF1, NF1, NKX2-1, NME8, PARN, PHOX2B, PIH1D3, RAPSN, RET, RSPH3, RSPH4A, RSPH9, RTEL1, SCN4A, SCNN1A, SCNN1B, SERPINA1, SFTP1A, SFTP1B, SFTP1C, SFTP1D, SLC34A2, SLC6A5, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2, ZEB2</i>
Neonatal respiratory diseases	56	<i>ABCA3, ARMC4, C21orf59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFTR, COPA, CSF2RA, CSF2RB, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FGFR2, FLNA, FOXF1, GAS2L2, GAS8, HYDIN, INV5, LRRK6, MCIDAS, NKX2-1, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SFTP1A, SFTP1B, SFTP1C, SFTP1D, SPAG1, TBX4, TMEM173, TTC25, ZMYND10</i>
Ciliopathies	176	<i>ACVR2B, ADGRV1, AHI1, API1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21orf59, C21orf71, C5orf42, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC172, CCDC28B, CCDC39, CCDC40, CCDC65, CCDC96, CCNO, CDH23, CENPF, CEP104, CEP120, CEP164, CEP290, CEP295, CEP41, CEP83, CFAP300, CFAP53, CFTR, CLRN1, CRB1, CRELD1, CRX, CSPP1, DCDC2, DEUP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYN2C1, EVC, EVC2, EXOC6B, EXOC8, FAM166B, FOXH1, GAS2L2, GAS8, GLI2, GUCY2D, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT43, IFT80, IMPDH1, INPP5E, INV5, IQCB1, KCNJ13, KIAA0556, KIAA0586, KIF14, KIF7, LCA5, LEFTY2, LRAT, LRRC34, LRRC6, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, MYO7A, NEK1, NEK8, NKX2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCDH15, PDE6D, PIEZO2, PIH1D3, PKD2, PKHD1, RD3, RDH12, RPE65, RPGR, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCNN1A, SCNN1B, SDCCAG8, SPAG1, SPATA7, TCTN1, TCTN2, TMEM107, TMEM138, TMEM17, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TSC1, TSC2, TTC21B, TTC23, TTC25, TTC6, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WHRN, XPNPEP3, ZIC3, ZMYND10, ZNF423</i>
Primary ciliary dyskinesia	41	<i>ARMC4, C21orf59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRK56, LRRK6, MCIDAS, NEK10, NME8, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC25, ZMYND10</i>
Familial pulmonary fibrosis	21	<i>ABCA3, AP3B1, CSF2RA, CSF2RB, DCK1, FOXF1, HPS1, HPS4, MUC5B, NKX2-1, RTEL1, PARN, SLC7A7, SFTP1A, SFTP1B, SFTP1C, SFTP1D, TERC, TERT, TINF2</i>
Myasthenia and respiratory failure	2	<i>SLC52A2, SLC52A3</i>

NOTAS

- Código SNS para todos os painéis: 34900;
- Todos os painéis são personalizáveis, permitindo a adição/exclusão de outros genes de interesse;
- Possibilidade de re-análise bioinformática para outro painel de genes ou para exoma clínico;
- Para outros painéis não listados, por favor contacte-nos previamente.