REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

GENOMED
Está nos genes.

Attachment I – NGS PANELS

Panel name	Gene number	Gene list
CARDIOLOGY / VASCULAR DISEASES (Cardiology panels may include CNVs analysis, with an additional cost. If you want that, please select that option in our requisition).		
Hypertrophic cardiomyopathy (sarcomeric genes)	8	*ACTC1, MYBPC3, MYH7, MYL2, MYL3, TNNI3, TNNT2, TPM1*
Hypertrophic cardiomyopathy (basic panel)	25	*ACTC1, ACTN2, CSRP3, GLA, KRAS, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, NEXN, PLN, PRKAG2, PTPN11, RAF1, RIT1, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR*
Hypertrophic cardiomyopathy (extended panel)	68	*AARS2, ACTA1, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CASQ2, CAV3, COA5, CRYAB, CSRP3, DES, FHL1, FHOD3, FLNC, FOXRED1, FXN, GAA, GLA, GLB1, GUSB, HRAS, JPH2, KCNQ1, KLF10, KRAS, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MRPL3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SCO2, SHOC2, SLC25A3, SLC25A4, SOS1, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, VCL*
Dilated cardiomyopathy (basic panel)	32	*ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DSG2, DMD, DSP, EMD, EYA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PSEN1, PSEN2, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL*
Dilated cardiomyopathy (extended panel)	47	*ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, CTF1, DES, DMD, DSG2, DSP, EMD, EYA4, FHL2, FHOD3, FKTN, FLNC, GATAD1, ILK, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYOZ1, MYPN, NEXN, PKP2, PLN, PSEN1, PSEN2, RBM20, SCN5A, SGCA, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL*
Left ventricular noncompaction	16	*ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1*
Arrhythmogenic right ventricular cardiomyopathy	17	*CASQ2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*
Cardiomyopathy and arrhythmia	196	*A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTF1, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FGF12, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, ILK, JPH2, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLN, PMM2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCA, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*
Cardiomyopathies	103	*AARS2, ABCC9, ACTA1, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FHL2, FHOD3, FKTN, FLNC, FOXRED1, FXN, GAA, GATAD1, GLA, GLB1, GUSB, HRAS, ILK, JPH2, JUP, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MIB1, MRPL3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NRAS, OBSCN, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SCO2, SGCA, SGCD, SHOC2, SLC25A3, SLC25A4, SOS1, TAZ, TCAP, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRIM63, TSFM, TTN, TTR, VCL*
Long QT syndrome	15	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*
Brugada syndrome	31	*ANK2, ANK3, ABCC9, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CAV3, CLASP2, DPP6, FGF12, GPD1L, HCN4, IRX5, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, PXDNL, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4*
Catecholaminergic polymorphic ventricular tachycardia	8	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN*
Cardiac arrhythmias	189	*A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, JPH2, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOM1, MYOT, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NRAS, OBSCN, PDHA1, PHKA1, PITX2, PKP2, PLN, PMM2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*
Sudden death without structural cardiopathy	65	*ABCC9, ACTC1, AKAP9, ANK2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CAVIN4, DES, DSC2, DSG2, DSP, EMD, FGF12, FHL2, FLNC, GAA, GJA5, GLA, GPD1L, HCN4, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, LAMP2, LMNA, NKX2-5, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLMAP, SNTA1, TBX5, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTR*
Ehlers-Danlos syndrome	20	*ADAMTS2, ATP7A, B3GALT6, B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, KIF22, PLOD1, PRDM5, SCN9A, SLC39A13, TNXB, ZNF469*

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Marfan and Marfan-like syndrome	9	*COL3A1, FBN1, FBN2, SKI, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2*
Aortic diseases	28	*ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, GATA5, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2*
Aortic / connective tissue diseases	62	*ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, 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, KCNJ8, LOX, LTBP2, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469*
Heritable pulmonary 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*
DERMATOLOGY		
Cutis laxa	15	*ALDH18A1, ATP6V0A2, ATP7A, C1R, COL11A1, EFEMP2, ELN, FBLN5, FLNA, GORAB, LTBP4, PLAC8, PTDSS1, PYCR1, RIN2*
Reticulate pigmentary dermatoses	17	*ABCB6, ADAM10, ADAR, CTC1, DKC1, KRT14, KRT5, NHP2, NOP10, POFUT1, POGLUT1, POLA1, SASH1, TERC, TERT, TINF2, WRAP53*
Ectodermal dysplasia	30	*ABCC9, BCS1L, CDH3, DLX3, DSP, EDA, EDA2R, EDAR, EDARADD, ERCC2, EVC, EVC2, GJB2, GJB6, HOXC13, IKBKG, IFT122, JUP, KCTD1, KRT74, KRT85, MSX1, NFKBIA, PORCN, RMRP, SHOC2, TP63, TRAF6, WDR35, WNT10A*
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		
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*
Autoinflammatory diseases	104	*ADA, ADA2, ADAM17, ADAR, AICDA, AP1S3, AP3B1, ASAH1, BLOC1S6, BTK, CARD14, CASP10, CASP8, CD27, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DDX58, DKC1, DOCK8, ELANE, EPCAM, FADD, 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, NLRP12, NLRP3, NOD2, NRAS, OTULIN, PIK3CD, PIK3R1, PLCG2, PRF1, PRKCD, PSMB8, PSTPIP1, RAB27A, RAG1, RAG2, RASGRP1, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, RTEL1, SAMHD1, SH2D1A, SI, SKIV2L, SLC29A3, SLC37A4, SLC7A7, STAT1, STAT3, STIM1, STX11, STXBP2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A, TREX1, TTC37, TTC7A, UNC13D, WAS, XIAP, ZAP70*
Recurrent fevers	35	*ADAR, AP1S3, ASAH1, CARD14, DDX58, ELANE, HAX1, IFIH1, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A, TREX1*
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	9	*C3, CD46, CFB, CFH, CFHR1, CFHR5, CFI, DGKE, THBD*
METABOLIC DISEASES		
Tangier disease	2	*ABCA1, LCAT*
Mitochondrial diseases (nuclear genes)	374	*AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, 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, DMPK, 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, GCSH, 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,*

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		KARS, KIF1B, KRT5, L2HGDH, LARS2, LIAS, LIPT1, LONP1, LRPPRC, LYRM4, LYRM7, MAOA, MAOB, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICU1, MIP, MLH1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSRB3, MTFMT, 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, RECQL4, RMND1, RNASEH1, RNASEL, RPIA, RPL35A, RPS14, RRM2B, SARDH, SARS2, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC37A4, SLC9A6, SNAP29, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, STOM, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TARS2, TCIRG1, TIMM44, TIMM8A, TK2, TMEM126A, TMEM70, TMLHE, TPI1, TPK1, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TWNK, TXNRD2, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, 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*
Familial hypercholesterolemia	11	*APOB, ABCG5, ABCG8, APOC2, APOC3, APOE, LDLR, LDLRAP1, LIPA, LPL, PCSK9*
Nonketotic hyperglycinemia	2	*AMT, GLDC*
Leukodystrophies	92	*ABCD1, ACADS, ACO2, 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, MTTP, 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		
Hypomagnesemia	24	*BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, EGFR, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, MMGT1, NIPA2, PCBD1, SARS2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7*
MODY	15	*ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, 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*
Kallmann syndrome	29	*ANOS1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, NSMF, POLR3B, PROK2, PROKR2, SEMA3A, SEMA3E, SEMA7A, SOX10, SPRY4, SRA1, TAC3, TACR3, WDR11*
PHARMACOGENETICS		
Personalized pharmacogenetics panel (Before requesting, please contact us. It is necessary to send a kit to collect saliva.)	27 (111 alleles)	*CYP1A2**1C, *1D, *1E, *1F, *1J, *1K, *1L, *1V, *1W; *CYP2B6**4, *5, *6, *7, *9, *16, *18; *CYP2C cluster* G/A; *CYP2C9**2, *3, *4, *5, *6, *8, *11; *CYP2C19**2, *3, *4, *4B, *10, *17; *CYP2D6**2A, *2, *3, *4, *4N, *4M, *5, *6, *6C, *7, *8, *9, *10, *11, *12, *13, *14A, *14B, *15, *17, *18, *19, *29, *31, *34, *35, *36, *39, *41, *42, *59, *63, *64, *68, *69, *70, *91, *109; *CYP3A4**1B, *22; *CYP3A5**3, *6, *7; *CYP4F2**3; *COMT* Val158Met; *DPYD**2A, Asp949Val, *13; *DRD2* -241A>G; *F2* 20210G>A; *F5* Leiden, *GRIK4* c.83-10039T>C; *HLA-A**31:01; *HLA-B**15:02, *HLA-B**57:01, *HLA-B**58:01; *HTR2A* c.614-2211T>C; *HTR2C* c.-759C>T; *IL28B* (IFNL4) c.151-152G>A; *MTHFR* 677C>T, 1298A>C; *NUDT15* Arg139Cys; *OPRM1* Asn40Asp; *SLC6A4* c.-1810A>G, -1791_-1749del43; *SLCO1B1**5, *17, *21; *TPMT**2, *3A, *3B, *3C, *4; *UGT1A1**6, *28; *VKORC1* c.442C>T, -1639G>A*
GASTROENTEROLOGY / HEPATOLOGY		
Familial intrahepatic cholestasis	3	*ABCB11, ABCB4, ATP8B1*
Neonatal intrahepatic cholestasis	54	*ABCB11, ABCB4, ACAD9, AKR1D1, ASAH1, ATP8B1, BAAT, BCS1L, CC2D2A, CFTR, CLDN1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, GBA, GBE1, HSD3B7, INVS, JAG1, LIPA, MKS1, MPV17, MYO5B, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKHD1, POLG, POLG2, RRM2B, SERPINA1, SLC25A13, TJP2, TRMU, VIPAS39, VPS33B*
Hemochromatosis	7	*FTH1, FTL, HAMP, HFE, HFE2, SLC40A1, TFR2*
Hereditary pancreatitis	8	*CASR, CFTR, CLDN2, CPA1, CTRC, SPINK1, PRSS1, PRSS2*
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	3	*FANCA, FANCC, FANCG*

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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*
NEPHROLOGY		
Renal tubular acidosis	3	*ATP6V0A4, ATP6V1B1, SLC4A1*
Cystinuria	2	*SLC3A1, SLC7A9*
Polycystic kidney disease	3	*PKD1, PKD2, PKHD1*
Polycystic kidney disease (extended panel)	31	*ABCC8, ALG8, BICC1, BLK, CEL, DNAJB11, DZIP1L, GANAB, GCK, HNF1B, HNF4A, INS, INVS, KCNJ11, KLF11, LRP5, NEUROD1, NOTCH2, NPHP3, OFD1, PAX4, PDX1, PKD1, PKD2, PKHD1, PRKCSH, SEC63, TSC1, TSC2, UMOD, VHL*
Familial renal glucosuria	2	*SLC2A2, SLC5A2*
Autosomal dominant interstitial nephritis (We also have available the analysis of the insC in the *MUC1* gene and CNVs analysis in the *HNF1B* gene - see attachment II.)	4	*HNF1B, REN, SEC61A1, UMOD*
Nephronophthisis	17	*ANKS6, CEP164, CEP290, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423*
Alport syndrome	4	*COL4A3, COL4A4, COL4A5, COL4A6*
Bartter and Gitelman syndromes	6	*BSND, CLCNKA, CLCNKB, KCNJ1, SLC12A1, SLC12A3*
Liddle syndrome	2	*SCNN1B, SCNN1G*
Nephrotic syndrome	48	*ACTN4, ADCK4, ALG1, ANLN, APOL1, ARHGAP24, ARHGDIA, CD151, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, CRB2, CUBN, DGKE, EMP2, EXT1, FAT1, GATA3, INF2, ITGA3, ITGB4, LAMB2, LMNA, LMX1B, MAFB, MYH9, MYO1E, NEU1, NPHS1, NPHS2, NUP107, NUP93, NXF5, PAX2, PDSS2, PLCE1, PMM2, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WDR73, WT1, ZMPSTE24*
NEUROLOGY		
Alzheimer disease and frontotemporal dementia	15	*APOE, APP, CHMP2B, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, TARDBP, TREM2, VCP*
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)	157	*ABCB7, ABHD12, ACO2, AFG3L2, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP1A3, 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, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, WDPCP, WDR81, WFS1, WWOX, ZFYVE26, ZNF423*
Spinal muscular atrophy	31	*AR, ASAH1, 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	81	*ADAR, 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, HNRNPDL, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TRAPPC11, TRIM32, TTN*
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*
Connective tissue diseases	49	*ABL1, ACTA2, ADAMTS2, ADAMTSL4, ATP7A, B3GALT6, B4GALT7, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL5A3, COL6A1, COL6A2, COL6A3, DSE, EFEMP2, ELN, FBN1, FBN2, FKBP14, FLCN, FLNA, GAA, HRAS, KCNJ8, KIF22, MED12, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469*

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Neuromuscular diseases	207	*ACAD9, ACADM, ACADVL, ACTA1, ADAMTS10, ADGRG6, AGL, AGRN, ALDOA, ANO5, ANTXR2, ASXL1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CAV3, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CNBP, CNTNAP1, COL12A1, COL13A1, 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, FLNC, GAA, GBA, GBE1, GFPT1, GLDN, GLE1, GMPPB, GNE, GYG1, GYS1, HADHA, HADHB, HSPB1, HSPB8, HSPG2, INPP5K, IRF6, ISCU, 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, PIEZO2, 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, TGFBR1, TGFBR2, TIA1, TK2, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, ZC4H2, ZMPSTE24*
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, CHRND, 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, PLOD1, PLOD2, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRG4, PRKAG2, PUS1, PYGM, RAPSN, RBCK1, 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, TGFBR1, TGFBR2, TIA1, TK2, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, YARS2, ZC4H2, ZMPSTE24*
Parkinson's disease	10	*ATP13A2, FBXO7, LRRK2, PARK7, PINK1, PRKN, SLC6A3, SNCA, TAF1, VPS35*
Epileptic encephalopathy	49	*ACY1, ADSL, ALDH7A1, AMT, ARHGEF9, ARX, CDKL5, CNTNAP2, CPT2, FOLR1, FOXG1, GABRG2, GAMT, GCSH, GLDC, GRIN2A, GRIN2B, KCNA2, KCNJ10, 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, CHRNB2, 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, CHRND, CHRNE, 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, 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, RBCK1, 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*
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, SCN11A, SCN9A, SH3TC2, SLC12A6, SLC52A3, SLC5A7, SMN1, SPTLC1, SPTLC2, SYT2, TFG, TRPV4, TTR, TWNK, VCP, WNK1, YARS*
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, FARS2, GAD1, GBA2, GJC2, GRID2, HSPD1, IBA57, KIF1A, KIF1C, KIF5A, KLC2, 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*

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REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

OPHTHALMOLOGY		
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, TDRD7, VIM*
Progressive external ophthalmoplegia	16	*DGUOK, DNA2, MGME1, MFN2, MPV17, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP*
Retinitis pigmentosa	131	*ABCA4, ABHD12, ADGRA3, AGBL5, AIPL1, ARL2BP, ARL3, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CACNA1F, CC2D2A, CDH23, CDHR1, CEP290, CERKL, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GNPTG, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, INPP5E, INVS, IQCB1, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, MKKS, MVK, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, OFD1, PCDH15, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX26, PEX7, PHYH, PITPNM3, PLA2G5, POMGNT1, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RD3, RDH11, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRIM32, TRNT1, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, WDR19, WFS1, WHRN, ZNF408, ZNF513*
ONCOLOGY		
BRCA1 and *BRCA2* genes – Pack 1 [insAlu (blood) + NGS (blood or FFPE) + MLPA (blood)] (For somatic mutations, in the tumor sample, we request the tumor infiltration percentage).	2	*BRCA1, BRCA2* (Includes the research of the founder mutation c.156_157insAlu in the *BRCA2* gene and MLPA in the blood sample. This includes the confirmation of the mutation's origin (germinal or somatic)).
BRCA1 and *BRCA2* genes – Pack 2 [insAlu (blood) + NGS (blood or FFPE)] (For somatic mutations, in the tumor sample, we request the tumor infiltration percentage).	2	*BRCA1, BRCA2* (Includes the research of the founder mutation c.156_157insAlu in the *BRCA2* gene in the blood sample. This includes the confirmation of the mutation's origin (germinal or somatic)).
Hereditary breast cancer (Includes Portuguese founder mutation (c.156_157insAlu) research in the *BRCA2* gene. This panel may include CNVs analysis, with an additional cost. If it is of your interest, please select that option in our requisition).	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*
Lynch syndrome	4	*MLH1, MSH2, MSH6, PMS2*
Hereditary colorectal cancer (This panel may include CNVs analysis, with an additional cost. If it is of your interest, please select that option in our requisition).	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, TELO2, TGFBR2, TP53, XAF1*
Hereditary pancreatic cancer	18	*APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53*
Hereditary prostate cancer (Includes Portuguese founder mutation (c.156_157insAlu) research in the *BRCA2* gene).	20	*ATM, BRCA1, BRCA2, HOXB13, CHEK2, RAD51C, RAD51D, PALB2, ATR, NBN, GEN1, MLH1, MSH2, MSH6, PMS2, MRE11, BRIP1, FAM175A, EPCAM, TP53*
Paraganglioma and pheochromocytoma	16	*EGLN1, EPAS1, FH, IDH1, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
Familial paraganglioma	3	*SDHD, SDHB, SDHC*
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*
OTORHINOLARYNGOLOGY		
Syndromic and nonsyndromic deafness	128	*ABHD12, ACTG1, ADGRV1, ALMS1, ANKH, ATP6V1B1, BSND, CABP2, CACNA1D, CCDC50, CD151, CDH23, CDKN1C, CEACAM16, CHD7, CHSY1, CIB2, CLDN14, CLIC5, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DLX5, DSPP, EDN3, EDNRB, ESPN, ESRRB, EYA1, EYA4, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, HGF, HOXB1, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, LHFPL5, LOXHD1, LRP2, LRTOMT, MANBA, MARVELD2, MIR96, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NDP, NLRP3, OTOA, OTOF, OTOG, OTOGL, PAX3, PCDH15, PDZD7, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLITRK6, SMPX, SNAI2, SOX10, STRC, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMC2, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, TYR, USH1C, USH1G, USH2A, WFS1, WHRN*
Usher and Alström syndrome	14	*ADGRV1, ALMS1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN*
Usher syndrome	13	*ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN*
PEDIATRICS		
Distal arthrogryposis	9	*ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2, TPM2, TNNI2, TNNT3*
Craniosynostosis	42	*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, TGFBR1, TGFBR2, TMCO1, TTR, TWIST1, WDR19, WDR35*
Multiple epiphyseal dysplasias	8	*COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, UFSP2*

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Skeletal dysplasias	105	*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, ENPP1, ESCO2, EVC, EVC2, FAM20C, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GDF5, GNPAT, 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, SERPINF1, SERPINH1, SHOX, SLC26A2, SLC34A3, SLC39A13, SMAD4, SMARCAL1, SOX9, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TRAPPC2, TRPV4, TTC21B, VDR, WDR19, WDR35, WISP3, WNT5A*
Hemifacial microsomia	13	*CHD7, DHODH, EFTUD2, EYA1, GNAI3, PLCB4, POLR1C, POLR1D, SALL1, SF3B4, SIX1, SIX5, TCOF1*
Hypophosphatemic rickets	11	*ALPL, CLCN5, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR*
Bardet-Biedl syndrome	20	*ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP*
Cornelia de Lange syndrome	5	*HDAC8, NIPBL, RAD21, SMC1A, SMC3*
Noonan syndrome/RASopathies	23	*A2ML1, BRAF, CBL, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
Klippel-Feil syndrome	3	*GDF3, GDF6, MEOX1*
Treacher Collins syndrome	3	*TCOF1, POLR1C, POLR1D*
PNEUMOLOGY		
Surfactant dysfunction (This panel may include CNVs analysis, with an additional cost. If it is of your interest, please select that option in our requisition).	7	*ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD*
Pulmonary disease (extended panel) (This panel may include CNVs analysis, with an additional cost. If it is of your interest, please select that option in our requisition).	67	*ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH1, DNAH11, DNAH5, DNAI1, 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, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC6A5, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2, ZEB2*
Neonatal respiratory diseases (This panel may include CNVs analysis, with an additional cost. If it is of your interest, please select that option in our requisition).	56	*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, INVS, LRRC6, MCIDAS, NKX2-1, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SFTPA1, SFTPB, SFTPC, SFTPD, SPAG1, TBX4, TMEM173, TTC25, ZMYND10*
Ciliopathies	174	*ACVR2B, ADGRV1, AHI1, AIPL1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21orf59, C2orf71, C5orf42, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC172, CCDC28B, CCDC39, CCDC40, CCDC65, CCDC96, CCNO, CDH23, CENPF, CEP104, CEP120, CEP164, CEP290, CEP295, CEP41, CEP83, CFAP53, CFTR, CLRN1, CRB1, CRELD1, CRX, CSPP1, DCDC2, DEUP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, EVC, EVC2, EXOC8, FAM166B, FOXH1, GAS2L2, GAS8, GDF1, GLIS2, GUCY2D, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IMPDH1, INPP5E, INVS, 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, SCNN1G, SDCCAG8, SPAG1, SPATA7, TCTN1, TCTN2, TCTN3, 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*
Primary ciliary dyskinesia	42	*ARMC4, C21orf59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS2L2, GAS8, HYDIN, INVS, LRRC6, MCIDAS, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC25, ZMYND10*
Familial pulmonary fibrosis	21	*ABCA3, AP3B1, CSF2RA, CSF2RB, DKC1, FOXF1, HPS1, HPS4, MUC5B, NKX2-1, RTEL1, PARN, SLC7A7, SFTPA1, SFTPA2, SFTPB, SFTPC, SFTPD, TERC, TERT, TINF2*
Myasthenia and respiratory failure	2	*SLC52A2, SLC52A3*

NOTES

- Portuguese SNS codes for all these panels is 34900.
- All panels can be personalized (except the “Pharmacogenetics” panel), allowing the addiction and exclusion of gene(s).
- Bioinformatics’ reanalysis can be performed for other panel or clinical exome.
- For the analysis of other panels, please contact us previously.

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