

Anexo I – PAINÉIS DE NGS

Nome do painel	Nº de genes	Lista de genes
CARDIOLOGIA		
Arritmias cardíacas	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, PRKAG2, PSEN1, PSEN2, RASGEF1, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, 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, TSMF, TTN, TTR, TXNRD2, VCL, XK
Doenças da aorta/tecido conjuntivo (painel básico)	64	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, IPO8, KCNJ8, LOX, LTBP2, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1, TGFB2, TNXB, ZNF469
Doenças da aorta/tecido conjuntivo (painel alargado)	121	ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, 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, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COX7B, CREB3L1, CRTAP, DCC, DLL4, DOCK6, DSE, EFEMP2, ELN, EMILIN1, EOGT, FBLN5, FBN1, 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, PIEZO2, 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, TGFB2, TMEM38B, TNXB, TPSAB1, VCAN, WNT1, ZNF469
Hipertensão arterial pulmonar hereditária	11	ACVRL1, BMPR1B, BMPR2, CAV1, CBLN2, EIF2AK4, ENG, FOXF1, KCNA5, KCNK3, SMAD9
Hipertensão monogénica	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
Miocárdio não-compactado	16	ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1
Miocardíopatia arritmogénica	19	CASQ2, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TTN
Miocardíopatia dilatada (painel básico)	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
Miocardíopatia dilatada (painel alargado)	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, GATA4, GATA6, GATAD1, GLA, HAMP, HCN4, 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, PRKAG2, 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
Miocardíopatia hipertrófica (genes sarcoméricos)	8	MYBPC3, MYH7, TNNT2, TNNI3, TPM1, ACTC1, MYL2, MYL3
Miocardíopatia hipertrófica (painel básico)	26	ACTC1, ACTN2, ALPK3, CSRP3, GLA, KRAS, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, NEXN, PLN, PRKAG2, PTPN11, RAF1, RIT1, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR
Miocardíopatia hipertrófica (painel alargado)	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, FHL2, 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, PRKAG2, PTPN11, RAF1, RASA1, RIT1, RRAS, RYR2, SCO2, SHOC2, SLC25A3, SLC25A4, SOS1, SOS2, SPRED1, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSMF, TTN, TTR, VCL

Miocardiopatia e arritmia	228	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, CSRP3, CTF1, CTNNA3, DES, DLD, 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, GPD1L, 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, MLYCD, MRPL3, MRPL4, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLEKHM2, PLN, PMM2, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, 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
Miocardiopatias	169	A2ML1, AARS2, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, AGL, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALM1, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, CPT2, CRYAB, CSRP3, 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, GPD1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MED12, MIB1, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SNTA1, SOS1, SOS2, SPEG, SPRED1, SYNE1, SYNE2, TAZ, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK
Morte súbita	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, COQ2, COX15, COX6B1, CPT1A, CPT2, CREB3L3, CRLF1, CRYAB, CSRP3, CTF1, CTNNA3, CYP27A1, DEPDC5, 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, GNAI2, GNPTAB, GPD1, GPD1L, 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, LDLR, LDLRAP1, LIAS, LIPA, LIPI, LMF1, LMNA, LPL, LRP6, LRRC10, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MFAP5, MIB1, MLYCD, MRPL3, MRPL4, MRPS22, MTO1, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, 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, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB2, TGFB3, TGFB3, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, USF1, VCL, XK
Patologias da aorta	31	ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, GATA5, IPO8, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB3, TGFB3, TGFB2, TNXB
Síndrome de Brugada	42	ABCC9, AKAP9, ANK2, ANK3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, DPP6, FGF12, GPD1L, HCN4, IRX5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, PKP2, PXDNL, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLMAP, SNTA1, TRDN, TRPM4
Síndrome de Ehlers-Danlos	22	ADAMTS2, AEBP1, ATP7A, B3GALT6, B4GALT7, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, KIF22, PLOD1, PRDM5, SCN9A, SLC39A13, TNXB, ZNF469
Síndrome de Marfan e Marfan-like	9	COL3A1, FBN1, FBN2, SKI, SLC2A10, SMAD3, TGFB2, TGFB3, TGFB2
Síndrome QT Longo (genes KCNQ1, KCNH2, SCN5A)	3	KCNQ1, KCNH2, SCN5A
Síndrome QT Longo	17	AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN
Taquicardia ventricular polimórfica catecolaminérgica	8	ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN
DERMATOLOGIA		
Cútiis laxa	15	ALDH18A1, ATP6V0A2, ATP7A, C1R, COL11A1, EFEMP2, ELN, FBLN5, FLNA, GORAB, LTBP4, PLAC8, PTSS1, PYCR1, RIN2
Dermatoses pigmentares reticuladas	17	ABCB6, ADAM10, ADAR, CTC1, DKC1, KRT14, KRT5, NHP2, NOP10, POFUT1, POGlut1, POLA1, SASH1, TERC, TERT, TINF2, WRAP53
Disqueratose congénita	11	CTC1, DKC1, NOP10, NHP2, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53
Displasias ectodérmicas	30	ABCC9, BCS1L, CDH3, DLX3, DSP, EDA, EDA2R, EDAR, EDARADD, ERCC2, EVC, EVC2, GJB2, GJB6, HOXC13, IKBKKG, IFT22, JUP, KCTD11, KRT74, KRT85, MSX1, NFKBIA, PORCN, RMRP, SHOC2, TP63, TRAF6, WDR35, WNT10A

Doença de Galli-Galli	7	ADAM10, ADAR, KRT14, KRT5, POFUT1, POGLUT1, PSENE1
Epidermólise bolhosa	14	COL17A1, COL7A1, DSP, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5
Epidermólise bolhosa (painel alargado)	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
Paquioníquia congénita	8	AAGAB, GJB6, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3
Queratoderma palmoplantar	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
Síndrome de Rothmund-Thomson	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, RFW3, RTEL1, SLX4, TERC, TERT, TINF2, UBE2T, USB1, WRAP53, WRN, XPA, XPC, XRCC2

DOENÇAS DO SISTEMA IMUNITÁRIO

Doenças auto-inflamatórias	105	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, 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, MEKV, MYO5B, NCF1, NCF2, NCF4, NEUROG3, NFAT5, NLRC4, NLRP1, NLRP2, 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
Doença inflamatória intestinal	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, MEKV, 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
Febres recorrentes	35	ADAR, AP1S3, ASAH1, CARD14, DDX58, ELANE, HAX1, IFIH1, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEKV, MYO5B, NCF1, NCF2, NCF4, NEUROG3, NFAT5, NLRC4, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A, TREX1
Imunodeficiências primárias	502	ABCD4, ACD, ACKR1, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGA, AICDA, AIRE, AK2, ALG1, ALG12, ALPI, AMT, AP1S3, AP3B1, AP3D1, APOL1, ARHGAP1, ARPC1B, ATG4A, ATM, ATP6AP1, AUH, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, BTK, C10orf62, 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, CDCA7, CEBPA, CEBPE, CFB, 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, CSF3R, CTC1, CTLA4, CTNBL1, CTSP1, CTSC, CXCR2, CXCR4, CYBA, CYBB, DBR1, CD81, CD8A, CDC42, CDCA7, CEBPA, CEBPE, CFB, 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, CSF3R, CTC1, CTLA4, CTNBL1, CTSP1, CTSC, CXCR2, CXCR4, CYBA, CYBB, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNM2, DNMT3B, DOCK2, DOCK8, DSG1, EFL1, ELANE, EP300, EPG5, ERBIN, ERCC4, ETV6, EXTL3, FAAP24, FADD, FAM111A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FBXL4, FCGR3A, FCHO1, FCN3, FDX2, FERMT1, FERMT3, FH, FIBP, FMO3, FNIP1, FOXN1, FOXP3, FPR1, FUT8, G6PC1, G6PC3, G6PD, GATA1, GATA2, GCSH, GFI1, GINS1, GLDC, GSS, GTF2H5, HAVCR2, HAX1, HELLS, HMOX1, HPS5, HTRA2, HUWE1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKKBK, IKZF1, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB2, IL12RB3, IL17F, IL17RA, IL17RC, IL18BP2, 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, MEKV, MKL1, MLH1, MMLA, MMAB, MMACHC, MOGS, MPL, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MTR, MUT, MVK, MYD88, MYSM1, NBAS, NBN, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC3, NLRC4, NLRP1, NLRP2, NLRP3, NLRP7, NOD2, NOP10, NOS2, NRAS, NSMCE3, 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, PSENE1, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, REL, RELB, RELB, RFW3, RFX5, RFXANK, RFXAP, RHOG, RHOH, 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, SH3BP1, SKIV2L, SLC19A2, SLC29A3, SLC33A1, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC7A7, SLX4, SMARCAL1, SMARCD2, SNX10, SOCS1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK11, STK4, STN1, STX11, STXBP2, TAFAZZIN, TALDO1, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TBX21, TCF3, TCIRG1, TCN2, TDP2, TERT, TFR2, TFR3, TFR4, TGFB1, TGFB2, THBD, THG1L, TICAM1, TINF2, TIRAP, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TONSL, TOP2B, TP53, TPP1, TPP2, 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
Síndrome linfoproliferativa autoimune (ALPS)	19	ADA2, AICDA, CASP10, CASP8, CD40LG, CTLA4, FADD, FAS, FASLG, ITK, KRAS, LRBA, MAGT1, NRAS, PRKCD, RASGRP1, SH2D1A, STAT3, WAS
Via alterna do complemento/ Síndrome hemolítico-urémica	12	C3, CD46, CFB, CFH, CFHR1, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD, VTN

DOENÇAS METABÓLICAS

Dislipidemia / Hipercolesterolemia familiar	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
Doença de Tangier	2	ABCA1, LCAT

Doenças mitocondriais (genes nucleares)	373	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, COX42, 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, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHR, 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, 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, NDUFB1, 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, UQCRCQ, VARS2, WDR81, WFS1, XPNPEP3, YARS2
Glicogenoses	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
Hiperamoniémia	4	CA5A, CPS1, NAGS, OTC
Hiperglicinemia não-cetótica	2	AMT, GLDC
Leucodistrofias	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, 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
ENDOCRINOLOGIA		
Hipogonadismo hipogonadotrófico (Síndrome de Kallmann)	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
Hipomagnesémia	24	BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, EGFR, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, MGMT1, 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
Obesidade	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
Obesidade não sindrômica	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
Obesidade sindrômica	20	BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CEP290, GNAS, MAGEL2, MKKS, MKS1, NTRK2, PHF6, SDCCAG8, SIM1, TRIM32, TTC8, VPS13B, WDPCP
Pseudo-hipoaldosteronismo tipo 2	4	CUL3, KLHL3, WNK1, WNK4
Puberdade precoce central	4	KISS1, KISS1R, MKRN3, DLK1
Resistência à insulina	12	ABCC8, AGPAT2, BSCL2, CAVIN1, GCK, GLUD1, HADH, HNF4A, IGF1R, INSR, KCNJ11, UCP2
GASTROENTEROLOGIA		
Colestase intra-hepática familiar	3	ABCB11, ABCB4, ATP8B1
Colestase neonatal intra-hepática	58	ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, ATP7B, ATP8B1, BAA1, CC2D2A, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, GALE, GALK1, GALT, GBA, GBE1, HADHA, HNF1B, HSD3B7, INVS, JAG1, LIPA, MPI, MPV17, MVK, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX6, PKHD1, POLG, SLC25A13, SMPD1, TALDO1, TJP2, TRMU, UGT1A1, VIPAS39, VPS33B
Hemocromatose	7	FTH1, FTL, HAMP, HFE, HFE2, SLC40A1, TFR2
Pancreatite hereditária	8	CASR, CFTR, CLDN2, CPA1, CTRC, SPINK1, PRSS1, PRSS2

Porfirias hereditárias	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS
HEMATOLOGIA		
Anemia de Blackfan-Diamond	20	GATA1, RPL5, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL36, RPS7, RPS10, RPS15, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, TSR2
Anemia de Fanconi	21	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANGG, FANGI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2
Malformações vasculares e linfáticas	63	ACVRL1, ADAMTS13, ADAMTS3, AKT1, AKT3, ANTXR1, ATM, BMPR2, BRAF, CAV1, CBL, CCBE1, CCM2, DOCK6, ELMO2, ENG, EPHB4, FAT4, FLT4, FOXC2, GATA2, GDF2, GJC2, GLMN, GNAQ, KCNK3, KDR, KIF11, KRAS, KRIT1, LZTR1, MAP2K1, MAP3K3, MTOR, NF1, NF2, NRAS, PDCD10, PDGFRB, PIEZO1, PIK3CA, PTEN, PTPN11, RAF1, RASA1, RIT1, SHOC2, SMAD2, SMAD3, SMAD4, SMARCB1, SOS1, SOX18, SPRED1, STAMBIP, TEK, TGFB2, TGFB3, TGFB1, TGFB2, TSC1, TSC2, VEGFC
Síndrome hemofagocítica	7	DCLRE1C, PRF1, STX11, STXBP2, RAG1, RAG2, UNC13D
Telangiectasia hemorrágica hereditária (Doença de Osler-Weber-Rendu)	5	ACVRL1, ENG, GDF2, RASA1, SMAD4
Trombastenia de Glanzmann	2	ITGA2B, ITGB3
NEFROLOGIA		
Acidose tubular renal	3	ATP6V0A4, ATP6V1B1, SLC4A1
Cistinúria	2	SLC3A1, SLC7A9
Colestases	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
Doença renal crónica no jovem	85	ACE, ACTG2, ACTN4, AGT, AGTR1, AMN, ANKS6, ARHGDI1, 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, INVS, 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
Doença renal poliquística	6	DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1
Doença renal poliquística (painel alargado)	32	ABCC8, ALG8, BICC1, BLK, COL4A1, DNAJB11, DZIP1L, GANAB, GCK, HNF1B, HNF4A, INS, INVS, KCNJ11, KLF11, LRP5, NEUROD1, NOTCH2, NPHP3, OFD1, PAX4, PDX1, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63, TSC1, TSC2, UMOD, VHL
Glicosúria renal familiar	2	SLC2A2, SLC5A2
Hiperoxalúria primária	3	AGXT, GRHPR, HOGA1
Litíase renal	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
Nefrite intersticial autossómica dominante	4	HNF1B, REN, SEC61A1, UMOD Temos também disponível a pesquisa da insC no gene MUC1 e MLPA no gene HNF1B por MLPA - consultar Anexo II.
Nefrite túbulo-intersticial	23	ADAMTS9, ANKS6, CEP164, CEP83, DCDC2, DNAJB11, GATM, GLIS2, HNF1B, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, REN, SEC61A1, TMEM67, TTC21B, UMOD, WDR19, XPNPEP3, ZNF423
Nefrocalcinose	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
Nefronoftise	17	ANKS6, CEP164, CEP290, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423
Síndrome de Alport	4	COL4A3, COL4A4, COL4A5, COL4A6
Síndrome de Bartter e Síndrome de Gitelman	6	BSND, CLCNKA, CLCNKB, KCNJ1, SLC12A1, SLC12A3
Síndrome de Liddle	2	SCNN1B, SCNN1G
Síndrome nefrótica	58	AACTN4, AMN, APOE, APOL1, ARHGDI1, 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, OSGEP, PAX2, PDSS2, PLCE1, PODXL, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, WDR73, WT1, YRDC
Tubulopatias	48	AP2S1, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN10, CLDN16, CLDN19, CNM2, CTNS, CUL3, CYP24A1, FAH, GATM, GNA11, HNF1B, HNF4A, KCNJ1, KCNJ10, KCNJ16, KLHL3, MAGED2, NR3C2, REN, RRAAGD, SARS2, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC4A1, SLC4A4, SLC5A2, TRPM6, UMOD, VIPAS39, VPS33B, WNK4
NEUROLOGIA		

Angiopatia amiloide familiar	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
Ataxias recessivas	17	AFG3L2, ANO10, ATM, COQ8A, MRE11, MTPAP, MTPP, PIK3R5, POLG, SACS, SETX, SIL1, SPTBN2, SYNE1, SYT14, TDP1, ZNF592
Ataxias (painel alargado)	158	ABCB7, 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, MTPP, NDUFA6, 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, WDCPC, WDR81, WFS1, WWOX, ZFYVE26, ZNF423
Atrofia muscular espinhal	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
Cavernomas cerebrais múltiplos	3	CCM2, KRIT1, PDCD10
Distonias	82	ADAR, ADCY5, ANO3, ATP13A2, ATP1A3, ATP7B, BTBD, C19orf12, CACNA1B, COASY, COL4A1, CP, CTSF, DCAF17, DDC, DLAT, DRD2, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FBXO7, FTL, GALT, 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
Distrofia muscular de cinturas	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
Doença de Alzheimer familiar e Demência frontotemporal	15	APOE, APP, CHMP2B, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, TARDBP, TREM2, VCP
Doença de Charcot-Marie-Tooth	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
Doença de Moyamoya	3	ACTA2, GUCY1A3, RNF213
Doença de Parkinson (painel básico)	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
Doença de Parkinson (painel alargado)	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, VPS13C, VPS35, XPR1
Doenças do tecido conjuntivo	51	ABL1, ACTA2, ADAMTS2, ADAMTSL4, ARHGAP36, ATP7A, B3GALT6, B4GALT7, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL5A3, COL6A1, COL6A2, COL6A3, DSE, EFEMP2, ELN, EMILIN1, FBN1, FBN2, FKBP14, FLCN, FLNA, GAA, HRAS, KCNJ8, KIF22, MED12, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD3, SMAD4, TGFB2, TGFB3, TGFB3R1, TGFB3R2, TNXB, ZNF469
Doenças neuromusculares	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, RAPS, 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, TGFB3R1, TGFB3R2, TIA1, TK2, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, ZC4H2, ZMPSTE24

Doenças neuromusculares e musculares	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, CHRN1, CHRN2, CHRNE, CHRNA3, 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, 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, TGFB3R1, TGFB3R2, TIA1, TK2, TMEM5, TNNI2, TNNI3, TNNI3, TPM2, TPM3, TRIM32, TRPV4, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, VCP, VIPAS39, VMA21, VPS33B, YARS2, ZC4H2, ZMPSTE24
Encefalopatia epiléptica	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, STXB1, TBCE, TCF4, TREX1, UBE3A, ZEB2
Enxaqueca	10	ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1
Epilepsia noturna do lobo frontal	6	CHRNA2, CHRNA4, CHRNB2, CRH, DEPDC5, KCNT1
Esclerose lateral amiotrófica	5	FUS, SOD1, TARDBP, SETX, VCP
Esclerose lateral amiotrófica juvenil	3	ALS2, ERLIN2, SETX
Esclerose tuberosa	2	TSC1, TSC2
Hemorragia intraventricular no periparto	5	COL4A1, COL4A2, GLA, HTRA1, TREX1
Miopatias	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, CHRN1, CHRN2, 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, TNNI1, TPM2, TPM3, TRIM32, TTN, TWNK, TYMP, VCP, VMA21, YARS2
Miopatias metabólicas e Rabdomiólise	64	ABHD5, ACAD9, ACADM, 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, RBCK1, RRM2B, RYR1, SIL1, SLC22A5, SLC25A1, SLC25A20, SUCLA2, TANG02, TAZ, TK2, TSFM, TYMP, VMA21
Miopatias miofibrilares	8	BAG3, CRYAB, DES, DNAJB6, FHL1, FLCN, LDB3, MYOT
Miotonias não-distróficas	11	ATP2A1, CACNA1A, CACNA1S, CAV3, CLCN1, HINT1, HSPG2, KCNA1, KCNE3, KCNJ18, SCN4A
Neurodegeneração com acumulação cerebral de ferro (NBIA)	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
Neuropatias	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
Oftalmoplegia externa progressiva	16	DGUOK, DNA2, MGME1, MFN2, MPV17, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP
Paralisias periódicas	11	ATP1A2, ATP1A3, CACNA1S, DNM1L, KCNJ12 (=KCNJ18), KCNJ2, KCNJ5, MCM3AP, RYR1, SCN4A, SLC12A3
Paraparesias espásticas	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
Síndrome de Fahr	4	PDGFRB, PDGFB, SLC20A2, XPR1

OFTALMOLOGIA

Cataratas congénitas	41	AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTD1P1, 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
Distrofias maculares	28	ABCA4, 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
Retinopatia pigmentar	160	ABCA4, ABHD12, ADAM9, ADGRA3, AGL5, AIPL1, ARL2BP, ARL3, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C21orf2, C2orf71, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CC2D2A, CDH23, CDHR1, CEP290, CEP78, GERKL, 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, PDE6G, 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, RPRIP1, RPRIP1L, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRIM32, TRNT1, TRPM1, TTC8, TLL5, TTPA, TUB, TULP1, UNC119, USH1C, USH2A, WDR19, WFS1, WHRN, ZNF408, ZNF513

ONCOLOGIA

Genes <i>BRCA1</i> e <i>BRCA2</i> – Pacote 1 [insAlu (sangue) + NGS (sangue ou FFPE) + MLPA (sangue)]	2	<i>BRCA1, BRCA2</i> *Inclui a pesquisa da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> e MLPA em amostra de sangue. Inclui confirmação da origem da mutação - germinal ou somática. Nota: para mutações somáticas - em amostra de tumor - solicitamos a referência à percentagem de infiltração tumoral.
Genes <i>BRCA1</i> e <i>BRCA2</i> – Pacote 2 [insAlu (sangue) + NGS (sangue ou FFPE)]	2	<i>BRCA1, BRCA2</i> *Inclui a pesquisa da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> em amostra de sangue. Inclui confirmação da origem da mutação - germinal ou somática. Nota: para mutações somáticas - em amostra de tumor - solicitamos a referência à percentagem de infiltração tumoral.
Cancro da mama/ovário hereditário	30	<i>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</i> *Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> .
Cancro colo-rectal hereditário	39	<i>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, TLO2, TGFB2, TP53, XAF1</i> *Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> .
Cancro do pâncreas hereditário	18	<i>APC, ATM, BMPR1A, BRCA1, BRCA2*, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53</i> *Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> .
Cancro da próstata hereditário	20	<i>ATM, BRCA1, BRCA2*, HOXB13, CHEK2, RAD51C, RAD51D, PALB2, ATR, NBN, GEN1, MLH1, MSH2, MSH6, PMS2, MRE11, BRIP1, FAM175A, EPCAM, TP53</i> *Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene <i>BRCA2</i> .
Melanoma maligno familiar	5	<i>BAP1, CDKN2A, CDK4, MITF, POT1</i>
Paraganglioma e feocromocitoma	18	<i>EGLN1, EPAS1, FH, IDH1, KIF1B, MAX, MDH2, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL</i>
Paragangliomas familiares	3	<i>SDHD, SDHB, SDHC</i>
Síndrome de Lynch	4	<i>MLH1, MSH2, MSH6, PMS2</i>
Tumores renais hereditários (painel básico)	10	<i>AKT1, BAP1, FLCN, FH, MET, PTEN, PIK3CA, STK11, SDHB, VHL</i>
Tumores renais hereditários (painel alargado)	25	<i>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</i>

OTORRINOLARINGOLOGIA

Surdez síndrómica e não-síndrómica	241	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BMP5, BSND, BTB, 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, GNAI3, GSPM2, 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, LHFPL5, LHX3, 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, OSBP2, OTOA, OTOF, OTOG, OTOGL, OTOR, P2RX2, PAX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PEX7, PHYH, PLCB4, PMP22, PNPT1, POLR1A, POLR1C, POLR1D, PORCN, POU3F4, POU4F3, PRPS1, PTPRO, RDX, RIPOR2, ROR1, RPS28, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC4A11, SLITRK6, 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, TRIOBP, TSPEAR, TWNK, TWSG1, TYR, USH1C, USH1G, USH2A, WFS1, WHRN
Síndrome de Usher e síndrome de Alström	14	<i>ADGRV1, ALMS1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN</i>

Síndrome de Usher	13	ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN
Síndrome de Waardenburg	7	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR
PEDIATRIA		
Artrogrípse distal	9	ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2, TPM2, TNNI2, TNNT3
Craniossinostoses	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, TGFBF1, TGFBF2, TMC01, TTR, TWIST1, WDR19, WDR35
Displasia epifisária múltipla	8	COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, UFSP2
Displasias esqueléticas	106	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, GNPAT, HSPG2, IFT140, IFT172, IFT80, IHH, IKBK, 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
Microsomia hemifacial	13	CHD7, DHODH, EFTUD2, EYA1, GNAI3, PLCB4, POLR1C, POLR1D, SALL1, SF3B4, SIX1, SIX5, TCOF1
Osteogenesis Imperfecta	2	COL1A1, COL1A2
Raquitismo hipofosfatémico	11	ALPL, CLCN5, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR
Síndrome de Bardet-Biedl	20	ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
Síndrome de Cornelia de Lange	5	HDAC8, NIPBL, RAD21, SMC1A, SMC3
Síndrome de Klippel-Feil	3	GDF3, GDF6, MEOX1
Síndrome de Noonan/Rasopatias	25	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
Síndrome de Treacher Collins	3	TCOF1, POLR1C, POLR1D
PNEUMOLOGIA		
Défice de surfactante	7	ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD
Doença pulmonar (painel alargado)	67	ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRN1, CHRN2, 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
Doença respiratória do recém-nascido	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, LRR6, MCIDAS, NKX2-1, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SFTPA1, SFTPB, SFTPC, SFTPD, SPAG1, TBX4, TMEM173, TTC25, ZMYND10
Ciliopatias	176	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, 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, DYNC2H1, EVC, EVC2, EXOC6B, EXOC8, FAM166B, FOXH1, GAS2L2, GAS8, GDF1, GLIS2, GUCY2D, HYDIN, HYL1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IMPDH1, INPP5E, INVS, IQCB1, KCNJ13, KIAA0556, KIAA0586, KIF14, KIF7, LCA5, LEFTY2, LRAT, LRR6, LRR6, 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, TMEM138, TMEM173, 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
Discinesias ciliares primárias	41	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, LRR6, LRR6, MCIDAS, NEK10, NME8, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC25, ZMYND10
Fibrose pulmonar familiar	21	ABCA3, AP3B1, CSF2RA, CSF2RB, DKC1, FOXF1, HPS1, HPS4, MUC5B, NKX2-1, RTEL1, PARN, SLC7A7, SFTPA1, SFTPA2, SFTPB, SFTPC, SFTPD, TERC, TERT, TINF2
Miastenia e insuficiência respiratória	2	SLC52A2, SLC52A3

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.