Anexo I – PAINÉIS DE NGS

Nome do painel	Nº de genes	Lista de genes
CARDIOLOGIA / DOENÇAS VASCULARES (Os painéis de Cardiologia têm opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)		
Miocardiopatia hipertrófica (genes sarcoméricos)	8	*ACTC1, MYBPC3, MYH7, MYL2, MYL3, TNNI3, TNNT2, TPM1*
Miocardiopatia hipertrófica (painel básico)	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*
Miocardiopatia hipertrófica (painel alargado)	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*
Miocardiopatia dilatada (painel básico)	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*
Miocardiopatia dilatada (painel alargado)	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*
Miocárdio não-compactado	16	*ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1*
Miocardiopatia arritmogénica do ventrículo direito	17	*CASQ2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*
Miocardiopatia e arritmia	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*
Miocardiopatias	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*
Síndrome do QT Longo	15	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*
Síndrome de Brugada	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*
Taquicardia ventricular polimórfica catecolaminérgica	8	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN*
Arritmias cardíacas	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*
Morte súbita sem cardiopatia estrutural	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*
Síndrome de Ehlers-Danlos	20	*ADAMTS2, ATP7A, B3GALT6, B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, KIF22, PLOD1, SLC39A13, TNXB, ZNF469, PRDM5, SCN9A*

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Síndrome de Marfan e Marfan-like	9	*COL3A1, FBN1, FBN2, SKI, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2*
Patologias da aorta	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*
Doenças da aorta/tecido conjuntivo	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*
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*
DERMATOLOGIA		
Dermatoses pigmentares reticuladas	17	*ABCB6, ADAM10, ADAR, CTC1, DKC1, KRT14, KRT5, NHP2, NOP10, POFUT1, POGLUT1, POLA1, SASH1, TERC, TERT, TINF2, WRAP53*
Displasias ectodérmicas	31	*ABCC9, BCS1L, CDH3, DLX3, DSP, EDA, EDA2R, EDAR, EDARADD, ERCC2, EVC, EVC2, GJB2, GJB6, HOXC13, IKBKG, IFT122, IKBKG, JUP, KCTD1, KRT74, KRT85, MSX1, NFKBIA, PORCN, RMRP, SHOC2, TP63, TRAF6, WDR35, WNT10A*
Doença de Galli-Galli	7	*ADAM10, ADAR, KRT14, KRT5, POFUT1, POGLUT1, PSENEN*
Epidermólise bolhosa	15	*COL17A1, COL7A1, DSP, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, PLEC1, TGM5*
Epidermólise bolhosa (painel alargado)	32	*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, PLEC1, SERPINB8, TGM5*
Paquioníquia congénita	8	*AAGAB, GJB6, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3*
Queratodermia 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, RFWD3, RTEL1, SLX4, TERC, TERT, TINF2, UBE2T, USB1, WRAP53, WRN, XPA, XPC, XRCC2*
DOENÇAS DO SISTEMA IMUNITÁRIO		
Doenças auto-inflamatórias	29	*ADA2 (=CECR1), AP3B1, BLOC1S6, CARD14, CD27, ELANE, IL1RN, IL36RN, ITK, LPIN2, LYST, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PRF1, PSMB8, PSTPIP1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, TMEM173, TNFRSF1A, UNC13D, XIAP*
Via alterna do complemento/ Síndrome hemolítico urémico	9	*C3, CD46, CFB, CFH, CFHR1, CFHR5, CFI, DGKE, THBD*
ENDOCRINOLOGIA / DOENÇAS METABÓLICAS		
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, ADCK4, 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, C10orf2, C12orf65, CA5A, CASP8, CAT, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COASY, COMT, COQ2, COQ4, COQ6, COQ8A, 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, 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, PTRF, 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, TXNRD2, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, 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*

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Hipercolesterolemia familiar	11	*APOB, ABCG5, ABCG8, APOC2, APOC3, APOE, LDLR, LDLRAP1, LIPA, LPL, PCSK9*
Hiperglicémia não cetónica	2	*AMT, GLDC*
Hipomagnesémia	29	*BSND, CASR, CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, EGFR, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, MMGT1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7, CASR, CLCNKB, HNF1B, KCNJ10, PCBD1, SARS2, SLC12A3, TRPM6*
Leucodistrofias	93	*ABCD1, ACADS, ACO2, ACOX1, ADAR, ADGRG1, ADSL, ALDH3A2, AP4B1, ARSA, ASPA, B3GALNT2, COX7B, CSF1R, CYP27A1, DARS, DARS2, DDOST, DHFR, DNM1L EARS2, EIF2B1, EIF2B1-5, 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*
MODY	15	*ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, SLC16A1*
Obesidade	47	*ADRB2, ADRB3, AGRP, ALMS1, AQP7, ARL6, CARTPT, ENPP1, FFAR4, FTO, GHRL, DYRK1B, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, UCP3, PPARG, SIM1, SLC6A14, UCP1, UCP2, UCP3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CEP290, GNAS, MAGEL2, MKKS, MKS1, NTRK2, PHF6, SDCCAG8, SIM1, TRIM32, TTC8, VPS13B, WDPCP*
Obesidade não sindromática	27	*ADRB2, ADRB3, AGRP, ALMS1, AQP7, ARL6, CARTPT, ENPP1, FFAR4, FTO, GHRL, DYRK1B, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, UCP3, PPARG, SIM1, SLC6A14, UCP1, UCP2, UCP3*
Obesidade sindromática	20	*BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CEP290, GNAS, MAGEL2, MKKS, MKS1, NTRK2, PHF6, SDCCAG8, SIM1, TRIM32, TTC8, VPS13B, WDPCP*
Síndrome de Kallmann	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*
FARMACOGENÉTICA		
Painel de Farmacogenética personalizado (Antes de requisitar este teste, por favor contacte-nos; é necessário o envio prévio de um kit de recolha de saliva.)	27 (111 alelos)	*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
GASTROENTEROLOGIA / HEPATOLOGIA		
Colestase intra-hepática familiar	3	*ABCB11, ABCB4, ATP8B1*
Colestase neonatal intra-hepática	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*
Hemocromatose	7	*FTH1, FTL, HAMP, HFE, HJV (=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	3	*FANCA, FANCC, FANCG*
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*
Doença poliquística renal	3	*PKD1, PKD2, PKHD1*
Doença poliquística renal (painel alargado)	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*
Glicosúria renal familiar	2	*SLC2A2, SLC5A2*
Nefrite intersticial autossómica dominante (Temos também disponível a pesquisa da insC no gene *MUC1* e a pesquisa de grandes rearranjos no gene *HNF1B* por MLPA - consultar Anexo II.)	4	*HNF1B, REN, SEC61A1, UMOD*

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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	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*
NEUROLOGIA / DOENÇAS NEUROMUSCULARES		
Alzheimer familiar e Demência frontotemporal	15	*APOE, APP, CHMP2B, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, TARDBP, TREM2, VCP*
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, ADCK3, ANO10, ATM, MRE11A, MTPAP, MTTP, PIK3R5, POLG, SACS, SETX, SIL1, SPTBN2, SYNE1, SYT14, TDP1, ZNF592*
Ataxias (painel alargado)	157	*ABCB7, ABHD12, ACO2, ADCK3, AFG3L2, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP1A3, ATP8A2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C5ORF42, CA8, CACNA1A, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COASY, 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, MRE11A, 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, UBA5, VAMP1, VLDLR, WDPCP, 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	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, PARK2, PARK7, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PINK1, PLA2G6, PLP1, PNKD, POLG, 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 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*
Doenças do tecido conjuntivo	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*
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, 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*
Doença de Parkinson	10	*ATP13A2, FBXO7, LRRK2, PARK7, PINK1, PRKN, SLC6A3, SNCA, TAF1, VPS35*
Encefalopatia epilética	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*
Enxaqueca	10	*ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1*
Epilepsia noturna do lobo frontal	6	*CHRNA2, CHRNA4, CHRNB2, CRH, DEPDC5, KCNT1*

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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, 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, LARGE, 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, SEPN1, 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*
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, C10orf2, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, EXOSC8, FAM134B, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF2, SCN10A, SCN11A, SCN9A, SH3TC2, SLC12A6, SLC52A3, SLC5A7, SMN1, SPTLC1, SPTLC2, SYT2, TFG, TRPV4, TTR, VCP, WNK1, YARS*
Paraparésias 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, 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*
Oftalmoplegia externa progressiva	16	*DGUOK, DNA2, MGME1, MFN2, MPV17, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP*
Retinopatia pigmentar	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*
Síndrome de Usher e síndrome de Alström	14	*ADGRV1, ALMS1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN*
ONCOLOGIA		
Cancro da mama/ovário (Para a pesquisa de mutações germinais - em amostra de sangue periférico - inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene *BRCA2*. Para a pesquisa de mutações somáticas - em amostra de tumor - solicitamos a referência à percentagem de infiltração tumoral na amostra para estudo.)	2	*BRCA1, BRCA2*
Cancro da mama hereditário (Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene *BRCA2*. Painel com opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)	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*
Síndrome de Lynch	4	*MLH1, MSH2, MSH6, PMS2*
Cancro colorrectal (Painel com opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)	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, SAMD7, STK11, TELO2, TGFBR2, TP53, XAF1*

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Cancro do pâncreas	18	*APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53*
Cancro da próstata hereditário (Inclui a pesquisa prévia da mutação fundadora c.156_157insAlu no gene *BRCA2*.)	20	*ATM, BRCA1, BRCA2, HOXB13, CHEK2, RAD51C, RAD51D, PALB2, ATR, NBN, GEN1, MLH1, MSH2, MSH6, PMS2, MRE11A, BRIP1, FAM175A, EPCAM, TP53*
Paraganglioma e feocromocitoma	16	*EGLN1, EPAS1, FH, IDH1, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
Paragangliomas familiares	3	*SDHD, SDHB, SDHC*
Tumores renais hereditários (painel básico)	10	*AKT1, BAP1, FLNC, FH, MET, PTEN, PIK3CA, STK11, SDHB, VHL*
Tumores renais hereditários (painel alargado)	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*
OTORRINOLARINGOLOGIA		
Surdez sindrómica e não-sindrómica	128	*ABHD12, ACTG1, 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, GPR98, 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*
Síndrome de Usher	13	*ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, WHRN*
PEDIATRIA		
Artrogripose 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, TGFBR1, TGFBR2, TMCO1, TTR, TWIST1, WDR19, WDR35*
Displasia epifisária múltipla	10	*COL2A1, COL9A1, COL9A2, COL9A3, COMP, COMP, MATN3, MATN3, SLC26A2, UFSP2*
Displasias esqueléticas	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*
Microssomia hemifacial	13	*CHD7, DHODH, EFTUD2, EYA1, GNAI3, PLCB4, POLR1C, POLR1D, SALL1, SF3B4, SIX1, SIX5, TCOF1*
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, CEP290, IFT27, LZTFL1, MKKS, MKS1, PTHB1, SDCCAG8, TRIM32, TTC8, WDPCP*
Síndrome de Cornelia de Lange	5	*HDAC8, NIPBL, RAD21, SMC1A, SMC3*
Síndrome de Noonan/Rasopatias	20	*A2ML1, BRAF, CBL, FGD1, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
Síndrome de Klippel-Feil	3	*GDF, GDF6, MEOX1*
Síndrome de Treacher Collins	3	*TCOF1, POLR1C, POLR1D*
PNEUMOLOGIA		
Défice de surfactante (Painel com opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)	7	*ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD*
Doença pulmonar (painel alargado) (Painel com opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)	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*
Doença respiratória do recém-nascido (Painel com opção de CNVs, mediante custo adicional. Se pretender CNVs, por favor assinale essa opção na requisição.)	58	*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, DYX1C1, FGFR2, FLNA, FOXF1, GAS2L2, GAS8, HEATR2, HYDIN, INVS, LRRC6, MCIDAS, NKX2-1, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SFTPA1, SFTPB, SFTPC, SFTPD, SPAG1, TBX4, TMEM173, TTC25, ZMYND10*

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Ciliopatias	164	*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, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CDH23, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP53, CFTR, CLRN1, CRB1, CRELD1, CRX, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, EVC, EVC2, EXOC8, 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, LRRC6, LZTFL1, 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, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TSC1, TSC2, TTC21B, TTC25, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WHRN, XPNPEP3, ZIC3, ZMYND10, ZNF423*
Discinesias ciliares primárias	44	*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, DYX1C1, GAS2L2, GAS8, HEATR2, HYDIN, INVS, LRRC6, MCIDAS, NME8, OFD1, 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 (excepto o painel de Farmacogenética), permitindo a adição 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.

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