REQUISIÇÃO DOENÇAS GENÉTICAS
FARMACOGENÉTICA

Anexo I – PAINÉIS DE NGS

Nome do painel	Nº de genes	Lista de genes
CARDIOLOGIA (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)	85	*A2ML1, AARS2, ACADVL, ACTA1, ACTC1, ACTN2, AGL, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COA5, CPT2, CRYAB, CSRP3, DES, ELAC2, FHL1, FHOD3, FLNC, FOXRED1, FXN, GAA, GATA4, GLA, GLB1, GUSB, HRAS, JPH2, KCNQ1, KLF10, KRAS, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEXN, NF1, NRAS, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RASA1, RIT1, RRAS, RYR2, SCO2, SHOC2, SLC25A3, SLC25A4, SOS1, SOS2, SPRED1, 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)	100	*ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CAV3, CAVIN4, CHRM2, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DMPK, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, GATA4, GATA6, GATAD1, GLA, HAMP, HCN4, HFE, HFE2, IDH2, ILK, 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, 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*
Miocárdio não-compactado	16	*ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1*
Miocardiopatia arritmogénica	17	*CASQ2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*
Miocardiopatia e arritmia	225	*A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALG10, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CBL, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CPT2, CRYAB, CSRP3, CTF1, CTNNA3, DES, DLD, DMD, DMPK, 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, MRPL44, 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, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*
Miocardiopatias	166	*A2ML1, AARS2, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, AGL, AKAP9, ALG10, ALMS1, ANK2, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CACNB2, CALM1, CALR3, CASQ2, CAV3, CAVIN4, CBL, CHRM2, COA5, CPT2, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, 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, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RRAS, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SNTA1, SOS1, SOS2, SPRED1, SYNE1, SYNE2, TAZ, TCAP, TFR2, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*
Síndrome do QT Longo	15	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*
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*
Taquicardia ventricular polimórfica catecolaminérgica	8	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN*
Arritmias cardíacas	201	*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, 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, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, 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, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

Morte súbita	295	*A2ML1, AARS2, ABCA1, ABCA12, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, AKT1, ALG10, ALG10B, ALMS1, 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, CHRM2, CLCF1, COA5, COA6, COL3A1, COL5A1, COL5A2, COQ2, COX15, COX6B1, CPT1A, CPT2, CREB3L3, CRLF1, CRYAB, CSRP3, CTF1, CTNNA3, CYP27A1, DEPDC5, DES, DLD, DMD, DMPK, 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, MRPL44, 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, PPP1R17, PRDM16, PRKAG2, PRKG1, PRRT2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RRAS, 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, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, USF1, VCL, XK*
Síndrome de Ehlers-Danlos	20	*ADAMTS2, ATP7A, B3GALT6, B4GALT7, 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, TGFBR1, TGFBR2*
Patologias da aorta	29	*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, TNXB*
Doenças da aorta / tecido conjuntivo (painel básico)	63	*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, TNXB, ZNF469*
Doenças da aorta / tecido conjuntivo (painel alargado)	115	*ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ALPL, ARHGAP31, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BGN, BMP1, 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, EOGT, FBLN5, FBN1, FBN2, FKBP10, FKBP14, FLCN, FLNA, FOXE3, GAA, GGCX, 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, RIN2, ROBO3, SERPINF1, SERPINH1, SGMS2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, SPARC, TAB2, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM38B, TNXB, TPSAB1, 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*
DERMATOLOGIA		
Cútis laxa	15	*ALDH18A1, ATP6V0A2, ATP7A, C1R, COL11A1, EFEMP2, ELN, FBLN5, FLNA, GORAB, LTBP4, PLAC8, PTDSS1, PYCR1, RIN2*
Dermatoses pigmentares reticuladas	17	*ABCB6, ADAM10, ADAR, CTC1, DKC1, KRT14, KRT5, NHP2, NOP10, POFUT1, POGLUT1, POLA1, SASH1, TERC, TERT, TINF2, WRAP53*
Displasias ectodérmicas	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*
Doença de Galli-Galli	7	*ADAM10, ADAR, KRT14, KRT5, POFUT1, POGLUT1, PSENEN*
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*
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		
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, 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*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

Doenças auto-inflamatórias	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*
Febres recorrentes	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*
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	9	*C3, CD46, CFB, CFH, CFHR1, CFHR5, CFI, DGKE, THBD*
DOENÇAS METABÓLICAS		
Doença de Tangier	2	*ABCA1, LCAT*
Doenças mitocondriais (genes nucleares)	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, 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*
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*
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*
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, 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*
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, 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*
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 sindromática	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 sindromática	20	*BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS9, CEP290, GNAS, MAGEL2, MKKS, MKS1, NTRK2, PHF6, SDCCAG8, SIM1, TRIM32, TTC8, VPS13B, WDPCP*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

Resistência à insulina	12	*ABCC8, AGPAT2, BSCL2, CAVIN1, GCK, GLUD1, HADH, HNF4A, IGF1R, INSR, KCNJ11, UCP2*
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		
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, 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 renal poliquística	6	*DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1*
Doença renal poliquística (painel alargado)	33	*ABCC8, ALG8, BICC1, BLK, CEL, 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*
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*
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		
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, MTTP, PIK3R5, POLG, SACS, SETX, SIL1, SPTBN2, SYNE1, SYT14, TDP1, ZNF592*
Ataxias (painel alargado)	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,*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

		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
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, 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*
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*
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, 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*
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)	48	*ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, COQ2, DNAJC13, DNAJC6, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, NR4A2, PARK7, PDGFB, PDGFRB, PINK1, PLA2G6, PODXL, PRKAG2, PRKN, PRKRA, RAB39B, RIC3, SLC18A2, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCAIP, SPR, SYNJ1, TAF1, TBP, TH, TMEM230, TRPM7, UCHL1, VPS13A, VPS13C, VPS35, XPR1*
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*
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*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

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, 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*
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*
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*
ONCOLOGIA		
Genes *BRCA1* e *BRCA2* – Pacote 1 [insAlu (sangue) + NGS (sangue ou FFPE) + MLPA (sangue)] (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* (Inclui a pesquisa da mutação fundadora c.156_157insAlu no gene *BRCA2* e MLPA em amostra de sangue. Inclui confirmação da origem da mutação - germinal ou somática.)
Genes *BRCA1* e *BRCA2* – Pacote 2 [insAlu (sangue) + NGS (sangue ou FFPE)] (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* (Inclui a pesquisa da mutação fundadora c.156_157insAlu no gene *BRCA2* em amostra de sangue. Inclui confirmação da origem da mutação - germinal ou somática.)
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 colo-rectal hereditário (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, SMAD7, STK11, TELO2, TGFBR2, TP53, XAF1*
Cancro do pâncreas hereditário	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, MRE11, BRIP1, FAM175A, EPCAM, TP53*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

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	241	*ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BMP5, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CDC14A, CDC6, CDH23, CDKN1C, CDT1, CEACAM16, CEMIP, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DFNA5, DFNB59, DHODH, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, ECE1, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, ELMOD3, EPS8, EPS8L2, ERCC2, ERCC3, ESPN, ESRRB, EVC, EYA1, EYA4, FAS, FGF10, FGF3, FGFR3, FOXI1, FOXI3, FRAS1, FREM2, GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB4, GJB6, GNAI3, GPSM2, GRHL2, GRIP1, GRXCR1, GRXCR2, GSC, GSTP1, HAAO, HAL, HARS2, HGF, HMX1, HOMER2, HOXA2, HOXB1, HSD17B4, HSPA9, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KDM6A, KIT, KITLG, KMT2D, LARS2, LEMD3, 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, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTOR, P2RX2, PAX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PEX7, PHYH, PLCB4, PMP22, PNPT1, POLR1A, POLR1C, POLR1D, PORCN, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, RPS28, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC4A11, 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	*ADGRV1, ALMS1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, TIMM8A, USH2A, USH1C, USH1G, 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	8	*COL2A1, COL9A1, COL9A2, COL9A3, COMP, 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, BBS9, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP*
Síndrome de Cornelia de Lange	5	*HDAC8, NIPBL, RAD21, SMC1A, SMC3*
Síndrome de Noonan/Rasopatias	24	*A2ML1, BRAF, CBL, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
Síndrome de Klippel-Feil	3	*GDF3, 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.)	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*

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt

Ciliopatias	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*
Discinesias ciliares primárias	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*
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/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.

GenoMed, Diagnósticos de Medicina Molecular SA
Instituto de Medicina Molecular
Faculdade de Medicina da Universidade de Lisboa

Edifício Egas Moniz
Av. Prof. Egas Moniz
1649-028 LISBOA
PORTUGAL

Tel: (351) 21 799 95 01
Fax: (351) 21 799 95 00
E-mail: genomed@genomed.pt
www.genomed.pt