Exomas

Exoma Clínico
WES - Sequenciação completa do exoma
WES Trio

Painéis NGS baseados em exoma

Doença de Parkinson e parkinsonismo (painel NGS baseado em exoma com análise de CNVs de 73 genes - ADORA1, ANO3, APP, ATP13A2, ATP1A3, ATP6AP2, C19orf12, CHCHD10, CHCHD2, CLN3, COASY, CSF1R, DCTN1, DNAJC12, DNAJC13, DNAJC6, EIF4G1, FBXO7, FTL, GBA, GCH1, GIGYF2, GNAL, GRN, HTRA2, KIF5A, LRRK2, LYST, MAPT, NR4A2, OPA3, PANK2, PARK7, PDE10A, PDE8B, PDGFB, PDGFRB, PINK1, PLA2G6, POLG, PRKAR1B, PRKN, PRKRA, PSEN1, PTRHD1, RAB39B, SGCE, SLC18A2, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCAIP, SNCB, SPG11, SPR, SYNJ1, TAF1, TARDBP, TENM4, TH, THAP1, TOR1A, TUBB4A, TWNK, UCHL1, VCP, VPS13A, VPS13C, VPS35, WDR45, XPR1)
Síndrome Ehlers-Danlos (painel NGS baseado em exoma com análise de CNVsde 26 genes - ABCC6, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, FBLN5, FKBP14, PLOD1, PRDM5, PYCR1, SLC39A13, TNXB, ZNF469)
Síndrome Ehlers-Danlos autossómico dominante (painel NGS baseado em exoma com análise de CNVs de 9 genes - C1R, C1S, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, SLC39A13)
Síndrome Ehlers-Danlos autossómico recessivo (painel NGS baseado em exoma com análise de CNVs de 19 genes - ABCC6, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, EFEMP2, FBLN5, FKBP14, PLOD1, PRDM5, PYCR1, SLC39A13, TNXB, ZNF469)
Charcot-Marie-Tooth e outras neuropatias periféricas hereditárias (painel NGS baseado em exoma com análise de CNVs de 134 genes - AAAS, AARS1, ABCA1, ABHD12, AIFM1, ALS2, ANG, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12ORF65, CCT5, CHCHD10, CLP1, COX6A1, CTDP1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, EXOSC3, FAM126A, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, GALC, GAN, GARS1, GDAP1, GJB1, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PLEKHG5, PLP1, PMP2, PMP22, PNKP, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, SYT2, TDP1, TFG, TRIM2, TRIP4, TRPA1, TRPV4, TTR, TUBB3, TWNK, TYMP, UBA1, VAPB, VCP, VRK1, WNK1, YARS1)

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Obesidade não sindrómica (painel NGS baseado em exoma com análise de CNVs de 25 genes - ADCY3, ADRB2, ADRB3, AGRP, CARTPT, CEP19, DYRK1B, ENPP1, GHRL, KSR2, LEP, LEPR, MC3R, MC4R, NR0B2, NTRK2, PCSK1, POMC, PPARG, PPP1R3A, SDC3, SH2B1, SIM1, TUB, UCP3)
Obesidade sindrómica (painel NGS baseado em exoma de 274 genes com análise de CNVs - ABCA4, ADNP, AFF4, AGBL5, AIP, AKT2, ALMS1, ANOS1, APC2, ApoE, ARHGEF18, ARL13B, ARL2BP, ARL6, ARMC5, ARNT2, ARVCF, ATRX, BAZ1B, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, BEST1, BLK, BRAF, C8orf37, CA4, CACNA1S, CANT1, CCDC141, CDH23, CDHR1, CEP164, CEP19, CEP290, CERKL, CHD7, CLIP2, CLRN1, CNGA1, CNGB1, CNNM2, COMT, CRB1, CREBBP, CRX, CTNNB1, CTSH, CUL4B, CYP19A1, CYP7A1, DEAF1, DHDDS, DPYD, DUSP6, DYNC2I2, DYRK1B, EGF, EHMT1, EIF2S3, ELN, EP300, ERMARD, EYS, FAM161A, FEZF1, FGF17, FGF8, FGFR1, FGFR3, FLII, FLRT3, FMR1, FOXP1, FSCN2, FTO, GABRA3, GABRD, GATA4, GHR, GNAS, GP1BB, GTF2I, GTF2IRD1, GUCA1B, H6PD, HACE1, HCRT, HDAC4, HDAC8, HERC2, HESX1, HGSNAT, HIRA, HLA-DQB1, HLA-DRB1, HS6ST1, HSD11B1, HUWE1, IDH3B, IFT140, IFT172, IFT27, IFT74, IGF1, IGFALS, IL17RD, IMPDH1, IMPG2, INPP5E, IQSEC2, JMJD1C, KCNAB2, KCNJ18, KDM6A, KIDINS220, KIF7, KISS1R, KIZ, KLHL7, KMT2A, KMT2D, LAS1L, LEP, LEPR, LIMK1, LIPE, LRAT, LZTFL1, MAGEL2, MAK, MAN1B1, MC4R, MEGF8, MERTK, MKKS, MKRN3, MKS1, MLXIPL, MOG, MTFMT, MYT1L, NDN, NEK2, NIPBL, NPAP1, NPHP1, NR0B2, NR2E3, NRL, NSD1, NSMF, NTRK2, OFD1, OTX2, P2RY11, PAX6, PCARE, PCNT, PCSK1, PDE11A, PDE4D, PDE6A, PDE6B, PDE6G, PDSS1, PHF6, PNPLA6, POMC, POMGNT1, PPARG, PRCD, PRDM16, PRKAR1A, PRMT7, PROK2, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PTEN, RAB23, RAD21, RAI1, RAP1A, RAP1B, RBMX, RBP3, RDH12, REEP6, RERE, RFC2, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RREB1, SAG, SDCCAG8, SEC24C, SEMA3A, SEMA4A, SETD2, SETD5, SH2B1, SHANK3, SHOX, SIM1, SIN3A, SKI, SLC25A4, SLC7A14, SLC7A7, SMAD4, SMC1A, SMC3, SNORD115-1, SNORD116-1, SNRNP200, SNRPN, SOX10, SOX2, SOX3, SPATA7, SPG11, SPRY4, SRY, STX16, TACR3, TBL2, TBX1, TBX3, THOC2, TMEM67, TNFSF4, TOPORS, TRAF3IP1, TRAPPC9, TRIM32, TTC8, TUB, TULP1, UBE3A, UFD1, USH2A, USP8, VPS13B, WDPCP, WDR11, WNT4, WT1, XRCC4, XYLT1, ZBTB20, ZDHHC15, ZNF365, ZNF408, ZNF513)
Doenças do tecido conjuntivo (painel NGS baseado em exoma com análise de CNVs de 80 genes - ACTA2, ABCC6, ABL1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COX7B, CRTAP, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, HCCS, LOX, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NDUFB11, NOTCH1, P3H1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, UPF3B, ZDHHC9, ZNF469)
Hemocromatose hereditária (painel NGS baseado em exoma com análise de CNVs de 11 genes e análise de CNVs - BMP2, CP, FTH1, FTL, HAMP, HFE, HJV, SLC11A2, SLC40A1, TFR2, TH)

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Malformações cerebrais (painel NGS baseado em exoma com análise de CNVs de 132 genes - ACTB, ACTG1, ADGRG1, AKT1, AKT3, AMPD2, ARFGEF2, ARID1A, ARID2, ARX, ASPM, ATP6V0A2, B3GALNT2, CASK, CCND2, CDC42, CDK5, CDK5RAP2, CDKL5, CDON, CENPE, CENPJ, CEP152, CHMP1A, CHN1, CIT, CLP1, COL4A1, COL4A2, CRPPA, CTNNA2, CUL4B, DCHS1, DCX, DEPDC5, DISP1, DLL1, DYNC1H1, DYRK1A, ECEL1, EXOSC3, EZH2, FAT4, FGF8, FKRP, FKTN, FLNA, FOXH1, GLDC, GLI2, GMPPB, GPSM2, HDAC8, HOXA1, HOXB1, IFIH1, KATNB1, KIF11, KIF21A, KIF2A, KIF5C, KIF7, KIFBP, LAMB1, LAMC3, LARGE1, LGI1, MACF1, MCPH1, MECP2, MTOR, MYMK, NDE1, NEDD4L, NODAL, NSDHL, OCLN, OFD1, OPHN1, PAFAH1B1, PHF6, PHOX2A, PIK3CA, PIK3R2, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, PTCH1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, RTTN, RXYLT1, SALL4, SEPSECS, SHH, SIX3, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SRD5A3, SRPX2, STAMBP, STIL, TBC1D20, TBC1D23, TDGF1, TGIF1, TMTC3, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, VLDLR, VRK1, WDR62, ZEB2, ZIC2)
Craniossinostoses (painel NGS baseado em exoma com análise de CNVs de 28 genes - CDC45, CYP26B1, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT122, IFT43, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, RAB23, RECQL4, RSPRY1, SKI, TCF12, TGFBR1, TGFBR2, TWIST1, WDR19, WDR35, ZIC1)
Doenças do movimento (painel NGS baseado em exoma com análise de CNVs de 640 genes - ADAR, AAAS, AARS1, AARS2, AASS, ABCA1, ABCA7, ABCB7, ABCD1, ABCD4, ABHD12, ACAT1, ACER3, ACOX1, ACP2, ACP5, ACTB, ACVR1, ACY1, ADAMTS13, ADCY5, ADD3, ADGRG1, ADH1C, ADRA2B, AFG3L2, AHI1, ALAS2, ALDH18A1, ALDH5A1, ALDH6A1, ALG6, ALS2, AMACR, AMPD2, ANG, ANO10, ANO3, ANO5, AP1S2, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOB, APTX, ARG1, ARHGAP31, ARHGEF28, ARL13B, ARL6, ARL6IP1, ARSA, ARSI, ARV1, ARX, ASAH1, ASL, ASS1, ATCAY, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATR, AUH, B4GALNT1, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCL11B, BCS1L, BEAN1, BICD2, BIN1, BRAT1, BSCL2, BTD, C12orf65, C19orf12, CA2, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CASQ1, CAV3, CBS, CC2D2A, CCDC28B, CCDC78, CCDC88C, CCT5, CEP290, CEP41, CFL2, CHCHD10, CHMP2B, CIT, CIZ1, CKAP2L, CLCN2, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNTN1, COASY, COL12A1, COL4A1, COL6A1, COL6A2, COL6A3, COMT, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, CP, CPLANE1, CPS1, CPT1C, CRAT, CRLF1, CRYAB, CSF1R, CSTB, CTC1, CTSD, CTSF, CWF19L1, CYB5R3, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS2, DBT, DCAF17, DCTN1, DDB2, DDC, DDHD1, DDHD2, DDX3X, DES, DGAT2, DLAT, DLD, DMXL2, DNA2, DNAJC12, DNAJC13, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNM2, DNMT1, DOCK6, DPYS, DRD2, DRD5, DYNC1H1, DYSF, EARS2, ECHS1, ECM1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELAC2, ELOVL4, ELOVL5, ELP2, EMC1, ENTPD1, EPM2A, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, FA2H, FAR1, FASTKD2, FBXL4, FBXO7, FGF14, FGFR1, FHL1, FKBP14, FLNC, FLRT1, FLVCR1, FOXG1, FOXRED1, FRRS1L, FTL, FUCA1, FUS, FXN, GAD1, GALC, GAMT, GAN, GARS1, GBA, GBA2, GBE1, GCDH, GCH1, GCLC, GFAP, GIGYF2, GJA1, GJB1, GJC2, GLB1, GLRA1, GLUD2, GLYCTK, GM2A, GNA11, GNAL, GNAO1, GNAS, GNB1, GNE, GOSR2, GPR88, GRID2, GRIK2, GRM1, GRN, GTF2E2, GTF2H5, HACD1, HACE1, HARS1, HEPACAM, HEXA, HEXB, HIBCH, HIKESHI, HIVEP2, HNRNPA1, HNRNPA2B1, HNRNPH2, HPCA, HPRT1, HSD17B4,

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HSPB1, HSPB3, HSPB8, HSPD1, HTRA2, HTT, IBA57, IFIH1, IFNG, IGHMBP2, ISCU, ISG15, ITM2B, ITPR1, IVD, JAM3, JPH3, KANK1, KBTBD13, KCNA1, KCNA4, KCNC3, KCND3, KCNJ10, KCNMA1, KCNQ2, KCTD17, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KIF5C, KIF7, KLC4, KLHL40, KLHL41, KLHL9, KMT2B, KRIT1, KY, L1CAM, L2HGDH, LAMP2, LARS2, LDB3, LIPT1, LIPT2, LMNB1, LMOD3, LRPPRC, LRRK2, LYST, MAG, MAPT, MARS1, MARS2, MAT1A, MATR3, MCCC2, MCEE, MCOLN1, MDH2, MECP2, MECR, MEGF10, MFF, MFSD8, MGP, MICU1, MKKS, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MMUT, MORC2, MPV17, MR1, MRE11, MRPS34, MSTN, MTHFR, MTM1, MTMR14, MTO1, MTPAP, MTTP, MYF6, MYH14, MYH2, MYH7, MYOT, NADK2, NALCN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEB, NEDD4, NEFH, NEU1, NHLRC1, NHP2, NIPA1, NKX2-1, NKX6-2, NLRP3, NME1, NOL3, NOP56, NPC1, NPC2, NPHP1, NT5C2, NUP62, OCLN, OFD1, OPA1, OPA3, OPTN, OTC, PABPN1, PAH, PANK2, PARK7, PAX6, PCCA, PCCB, PDE10A, PDE2A, PDE8B, PDGFB, PDGFRB, PDHA1, PDHX, PDSS1, PDSS2, PDYN, PET100, PEX2, PEX6, PEX7, PFN1, PGAP1, PHYH, PIK3R5, PINK1, PLA2G6, PLEKHG2, PLEKHG5, PLP1, PMPCA, PNKD, PNKP, PNP, PNPLA6, PNPLA8, PNPT1, PODXL, POLG, POLR1C, POLR3A, POLR3B, PPP1R15B, PPP2R2B, PPT1, PQBP1, PRKCG, PRKN, PRKRA, PRNP, PRPH, PRRT2, PSEN1, PSMB8, PTEN, PTS, QDPR, RAB18, RAB29, RAB39B, RAB3GAP1, RAB3GAP2, RBBP8, REEP1, REEP2, RELN, RETREG1, RLIM, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF125, RNF216, RTN2, RUBCN, SACS, SAMHD1, SCN1A, SCN2A, SCN8A, SCO2, SCP2, SDHA, SDHAF1, SDHD, SELENOI, SERAC1, SETX, SFXN4, SGCE, SIGMAR1, SIL1, SLC16A2, SLC19A3, SLC1A3, SLC20A2, SLC25A15, SLC25A26, SLC2A1, SLC30A10, SLC30A9, SLC33A1, SLC39A14, SLC39A4, SLC46A1, SLC52A2, SLC52A3, SLC6A17, SLC6A19, SLC6A3, SLC6A8, SLC9A1, SMPD1, SNAP25, SNCA, SNX14, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTBN2, SQSTM1, STAMBP, STN1, STUB1, STXBP1, SUCLA2, SUOX, SURF1, SYNE1, SYNJ1, SYT1, SYT14, TACO1, TAF1, TAF2, TANGO2, TBC1D24, TBCD, TECPR2, TELO2, TENM4, TFG, TGM6, TH, THAP1, THOC2, TIMM8A, TIMMDC1, TINF2, TMEM126B, TMEM230, TOR1A, TOR1AIP1, TPI1, TPK1, TPP1, TRAPPC11, TREM2, TREX1, TRIM32, TSEN2, TSEN54, TSFM, TTBK2, TTC19, TTPA, TTR, TUBB4A, TWNK, TXN2, TYROBP, UBA5, UBQLN2, UBTF, UCHL1, UNC13A, UPB1, UQCRQ, UROC1, USP18, USP8, VAC14, VAMP1, VCP, VPS13A, VPS13C, VPS13D, VPS35, VPS37A, VWA3B, WARS2, WASHC5, WDR45, WDR73, WWOX, XK, XPNPEP3, XPR1, XRCC4, ZBTB20, ZC4H2, ZFYVE26, ZFYVE27, ZNF592)

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Doença neuromuscular (painel NGS baseado em exoma com análise de CNVs de 417 genes - AAAS, AARS1, ABCA1, ABHD12, ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTG2, ADAMTS10, ADCY6, ADGRG6, ADSS1, AGL, AGRN, AIFM1, ALDOA, ALG14, ALG2, ALS2, AMPD1, ANG, ANO5, ANTXR2, ANXA11, ARHGEF10, ASAH1, ASCC1, ASXL1, ATL1, ATL3, ATP13A2, ATP1A1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, BVES, C12orf65, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCNF, CCT5, CFL2, CHAT, CHCHD10, CHCHD2, CHKB, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CLTCL1, CNBP, CNTN1, CNTNAP1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPT2, CRLF1, CRPPA, CRYAB, CTDP1, DAG1, DAO, DCAF8, DCTN1, DES, DGAT2, DHTKD1, DMD, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DST, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, ELP3, EMD, ENO3, EPG5, ERBB3, ERBB4, ERCC6, ERCC8, ERLIN1, ETFA, ETFB, ETFDH, EWSR1, EXOSC3, EXOSC8, EXOSC9, FAM126A, FAM20C, FBLN5, FBN2, FBXO38, FGD4, FGFR2, FGFR3, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNB, FLNC, FLVCR1, FUS, GAA, GALC, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GJB1, GLA, GLDN, GLE1, GLT8D1, GMPPB, GNB4, GNE, GRN, GSN, GYG1, GYS1, HACD1, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HNRNPA1, hnRNPA2B1, HNRNPDL, HOXD10, HSBP1, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, INPP5K, IRF6, ISCU, ITGA7, JPH1, KARS1, KAT6B, KBTBD13, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL7, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MAP3K20, MAPT, MARS1, MATR3, MB, MED25, MEGF10, MFN2, MICU1, MME, MORC2, MPV17, MPZ, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYMK, MYO18B, MYO9A, MYOT, MYPN, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK1, NGF, NTRK1, OPA1, OPTN, ORAI1, PAX7, PDK3, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD1, PLOD2, PLP1, PMP2, PMP22, PNKP, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRDM12, PREPL, PRG4, PRKAG2, PRPH, PRPS1, PRUNE1, PRX, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, RBM7, REEP1, RETREG1, RIPK4, RNF170, RRM2B, RXYLT1, RYR1, RYR3, SACS, SBF1, SBF2, SCARF2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SCP2, SELENON, SETX, SGCA, SGCB, SGCD, SGCG, SH3TC2, SIGMAR1, SIL1, SKI, SLC12A6, SLC18A3, SLC22A5, SLC25A1, SLC25A4, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMAD3, SMAD4, SNAP25, SOD1, SOX10, SPART, SPAST, SPEG, SPG11, SPTLC1, SPTLC2, SQSTM1, STAC3, STIM1, SUCLA2, SURF1, SYNE1, SYNE2, SYT2, TAF15, TARDBP, TAZ, TBCD, TBCE, TBK1, TCAP, TECPR2, TFG, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TNXB, TOR1AIP1, TPM2, TPM3, TRAPPC11, TREM2, TRIM2, TRIM32, TRIM63, TRIP4, TRPV4, TSEN54, TSFM, TTN, TTR, TUBA4A, TUBB3, TWNK, TYMP, UBA1, UBQLN2, UNC13A, VAMP1, VAPB, VCP, VIPAS39, VMA21, VPS33B, VRK1, WARS1, WNK1, YARS1, ZC4H2, ZMPSTE24)
Doenças hereditárias do metabolismo de moléculas complexas (painel NGS baseado em exoma com análise de CNVs de 100 genes - ABCD1, ABCD3, ABHD5, ACOX1, ADAMTSL2, AGA, AGPS, AGXT, AMACR, ANTXR2, ARSA, ARSB, ASAH1, ATP13A2, CAT, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSF, DHCR7, DNAJC5, DNM1L, DYM, EBP, EHHADH, FAR1, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GRHPR, GUSB, HEXA, HEXB,

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HGSNAT, HRAS, HSD17B4, HYAL1, IDS, IDUA, LAMP2, LIPA, LYST, MAN2B1, MANBA, MCOLN1, MFF, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PNPLA2, PPT1, PSAP, RAI1, SCP2, SGSH, SLC17A5, SMPD1, SUGCT, SUMF1, TCF4, TPO, TPP1, TRIM37, VPS33A)
Doenças hereditárias do metabolismo intermediário (painel NGS baseado em exoma com análise de CNVs de 45 genes - ACAT1, ACY1, AGXT, ARG1, ASL, ASPA, ASS1, AUH, BCKDHA, BCKDHB, BTD, CBS, CTNS, CYP27A1, DBT, DHCR7, EBP, ETHE1, FAH, FMO3, GCDH, GCH1, HLCS, HMGCL, HPD, HSD17B10, L2HGDH, MAT1A, MCCC1, MCCC2, MLYCD, MMACHC, MMADHC, MMUT, MTHFR, NSDHL, PAH, PCCA, PCCB, SLC25A13, SLC25A15, SLC3A1, SLC7A7, SLC7A9, TAT)
Hiperglicemia (painel NGS baseado em exoma com análise de CNVs de 51 genes - ABCC8, AGPAT2, AGPS, AIRE, AKT2, ALMS1, APPL1, AQP2, AVP, AVPR2, BLK, BSCL2, CAV1, CEL, CIDEC, COQ2, COQ9, DCAF17, DMXL2, DNAJC3, DYRK1B, EIF2AK3, EIF2S3, ENPP1, FOXC2, FOXP3, GATA4, GATA6, GCK, GLIS3, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, KCNJ11, KLF11, MNX1, NEUROD1, NEUROG3, NKX2-2, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC19A2, WFS1, ZFP57)
Hipoglicemia (painel NGS baseado em exoma com análise de CNVs de 75 genes - ABCC8, ACAD9, ACADM, ACADS, ACADVL, ACAT1, ACSF3, AGL, AKT2, AKT3, ALDOB, BCKDHA, BCKDHB, CA5A, CACNA1C, CPT1A, CPT2, DBT, DGUOK, DIS3L2, DLD, ETFA, ETFB, ETFDH, FBP1, G6PC1, GALT, GBE1, GCK, GHR, GK, GLUD1, GPC3, GYS2, HADH, HADHA, HADHB, HMGCL, HMGCS2, HNF1A, HNF4A, HRAS, INSR, KCNJ11, KMT2B, KMT2D, MLYCD, MPI, MPV17, NNT, NSD1, OXCT1, PC, PCK1, PCK2, PGM1, PHKA2, PHKB, PHKG2, PIK3CA, PMM2, POLG, PRKAG2, PYGL, RNF125, SLC16A1, SLC22A5, SLC25A13, SLC25A20, SLC2A1, SLC2A2, SLC37A4, SLC5A1, TANGO2, UCP2)
Genes nucleares associados a doenças mitocondriais (painel NGS baseado em exoma com análise de CNVs de 342 genes - AARS2, AASS, ABAT, ABCB7, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH3A2, ALDH4A1, ALDH6A1, AMACR, AMPD1, AMT, APTX, ATL1, ATP5F1A, ATP5F1E, ATP5MC3, ATP7B, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C12ORF65, C19orf12, C1QBP, CA5A, CARS2, CEP89, CHAT, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COA8, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, COX8A, CPS1, CPT1A, CPT2, CRAT, CYB5R3, CYC1, CYCS, CYP27A1, D2HGDH, DARS1, DARS2, DBT, DDHD1, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, EPRS1, ERCC6, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FBXL4, FDX2, FDXR, FH, FLAD1, FOXRED1, GAMT, GARS1, GATM, GCDH, GCK, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GTPBP3, GYG2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HK1, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH1, IDH2, IDH3B, ISCA1, ISCA2, ISCU, IVD, KARS1, KIF1B, KIF5A, L2HGDH, LAMP2, LARS1, LARS2, LIAS, LIPT1, LIPT2, LRPPRC, LYRM4, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFF, MFN2, MGME1, MICOS13, MICU1, MLYCD, MMAA, MMAB, MMADHC, MMUT, MPC1, MPV17, MRPL12, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MSTO1, MTFMT, MTO1, MTPAP, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1,

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NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NR2F1, NUBPL, OAT, OGDH, OPA1, OPA3, OTC, OXCT1, PANK2, PARK7, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PINK1, PNKD, PNPLA2, PNPLA8, PNPT1, POLG, POLG2, PPOX, PRKAG2, PRKN, PUS1, QARS1, RARS1, RARS2, REEP1, RMND1, RNASEH1, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC33A1, SLC52A2, SLC6A8, SPART, SPAST, SPG7, STAR, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TANGO2, TARS2, TAZ, TFAM, TIMM44, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TPK1, TRAP1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WFS1, WWOX, XPNPEP3, YARS2, YWHAE)
Leucodistrofias e leucoencefalopatias (painel NGS baseado em exoma com análise de CNVs de 123 genes - AARS2, ABCD1, ACOX1, ADAR, ADGRG1, ADSL, AIMP1, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, COA8, COL4A1, COL4A2, COLGALT1, COQ2, COX10, COX15, CSF1R, CTC1, CTSA, CYP27A1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DDHD2, DEGS1, DGUOK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, ERCC6, ERCC8, ETFDH, FA2H, FAM126A, FOLR1, FOXC1, FUCA1, GALC, GAN, GBE1, GCDH, GFAP, GFM1, GJA1, GJB1, GJC2, GLA, GLB1, HEPACAM, HEXA, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA2, L2HGDH, LMNB1, LYRM7, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NDUFA1, NDUFS1, NDUFS2, NDUFS4, NDUFS8, NOTCH3, NUBPL, PEX1, PEX10, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLEKHG2, PLP1, POLG, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RRM2B, SAMHD1, SCO1, SCP2, SDHAF1, SLC16A2, SLC17A5, SOX10, STN1, SUCLA2, SUMF1, SURF1, TBCK, TREM2, TREX1, TUBB4A, TWNK, TYMP, TYROBP, VPS11)
Encefalopatias epiléticas (painel NGS baseado em exoma com análise de CNVs de 257 genes - AARS1, ACY1, ADGRV1, ADSL, AKT3, ALDH7A1, ALG11, ALG13, AMT, AP3B2, ARFGEF2, ARHGEF15, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6AP2, ATP6V0A2, ATRX, AUH, BCKDK, BRAT1, BSCL2, CACNA1A, CACNA1D, CACNA1H, CACNA2D2, CACNB4, CAD, CDK5, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CIC, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNTNAP2, COL4A1, COL4A2, COX15, CPA6, CPT2, CRADD, CSF1R, CSTB, CTSD, CTSF, CYFIP2, DARS2, DCX, DENND5A, DEPDC5, DIAPH1, DNAJC5, DNM1, DNM1L, DOCK7, DPYD, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EML1, EMX2, EPG5, EPM2A, ERBB4, ETHE1, FADD, FARS2, FBXL4, FGF12, FGFR1, FLNA, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GLI2, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GUF1, HCFC1, HCN1, HCN2, HEPACAM, HNRNPU, HTR2A, HTRA1, HUWE1, IQSEC2, IRF3, ITPA, KANSL1, KARS1, KATNB1, KCNA2, KCNB1, KCNH2, KCNH5, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, KPNA7, LAMB1, LGI1, LIAS, LIPT2, MAGI2, MAPK10, MBD5, MDH2, MECP2, MEF2C, MFF, MFSD8, MLC1, MOCS1, MOCS2, MTOR, NAPB, NAXE, NDE1, NDP, NECAP1, NEDD4L, NEXMIF, NHLRC1, NOTCH3, NPRL2, NR2F1, NRXN1, NTRK2, PAFAH1B1, PCDH19, PHGDH, PIGA, PIGP, PIGQ, PIGV, PIK3CA, PIK3R2, PLCB1, PNKP, PNPO, POLG, PPP3CA, PPT1, PRICKLE1, PRRT2, PSAT1, PSPH, PTCH1, PTEN, PURA, QARS1, RANBP2, RBFOX1, RBFOX3, RELN, RNASET2, ROGDI,

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RRM2B, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SCO2, SERAC1, SERPINI1, SHH, SIK1, SIX3, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC6A9, SLC9A6, SPTAN1, SRGAP2, SRPX2, ST3GAL3, ST3GAL5, STRADA, STX1B, STXBP1, SUCLA2, SYN1, SYNGAP1, SYNJ1, SZT2, TANGO2, TBC1D24, TBC1D7, TBCD, TBCE, TCF4, TMTC3, TNK2, TPK1, TPP1, TRAF3, TRAPPC12, TRIO, TSC1, TSC2, TUBA1A, UBA5, UBE3A, WDR45, WWOX, YWHAG, ZEB2)
Surdez (painel NGS baseado em exoma com análise de CNVs de 306 genes - ABHD12, ABHD5, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, AMMECR1, ANKH, AQP4, ARSB, ASAH1, ATP1A2, ATP2B2, ATP6V1B1, ATP6V1B2, BCAP31, BCS1L, BDP1, BMP4, BMP5, BSN, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CDC14A, CDC42, CDC6, CDH23, CDKN1C, CDT1, CEACAM16, CEMIP, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYL1, CRYM, DACT1, DCDC2, DHODH, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, DVL1, DVL3, ECE1, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, ELMOD3, EPS8, EPS8L2, ERAL1, ERCC2, ERCC3, ESPN, ESRRB, EVC, EYA1, EYA4, FAS, FBXO2, FGF10, FGF3, FGFR1, FGFR2, FGFR3, FKBP14, FOXC1, FOXI1, FOXI3, FRAS1, FREM2, GATA3, GDF6, GFER, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB4, GJB6, GLI3, GNAI3, GPSM2, GRHL2, GRIP1, GRXCR1, GRXCR2, GSC, GSDME, GSTP1, GUSB, HAAO, HAL, HARS, HARS1, HARS2, HGF, HMX1, HMX2, HOMER2, HOXA1, HOXA2, HOXB1, HSD17B, HSD17B4, HSPA9, HTRA2, IGF1, ILDR1, JAG1, KARS, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KDM6A, KIT, KITLG, KMT2D, LARS2, LEMD3, LHFPL5, LHX3, LOXHD1, LRP2, LRTOMT, MAFB, MANBA, MARVEL, MARVELD2, MASP1, MCM2, MET, MITF, MSRB3, MTAP, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO1F, MYO3A, MYO6, MYO7A, NARS2, NDP, NEU1, NF2, NKX3-2, NLRP3, NR2F1, OFD1, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTOR, P2RX2, PAX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PEX7, PHEX, PHYH, PJVK, PLCB4, PMP22, PNPT1, POLD1, POLR1A, POLR1C, POLR1D, PORCN, POU3F4, POU4F3, PRPS1, PRRX1, PTPRQ, RDX, RIPOR2, ROR1, RPGR, RPS28, RPS6KA3, S1PR2, SALL1, SALL4, SCARB2, SDHD, SEMA3E, SERAC1, SERPIN, SERPINB6, SF3B4, SGPL1, SIX1, SIX5, SLC17A, SLC17A8, SLC19A2, SLC26A, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC4A1, SLC4A11, SLC52A2, SLC52A3, SLC9A1, SLITRK, SLITRK6, SMAD4, SMPX, SMS, SNAI2, SOBP, SOX10, SOX2, SOX9, SPATA5, SPINK5, ST3GAL5, STRC, SYNE4, TBC1D2, TBC1D24, TBL1X, TBX1, TCF21, TCOF1, TECTA, TFAP2A, TFCP2, THRA, THRB, TIMM8A, TJP2, TMC1, TMC2, TMEM13, TMEM132E, TMIE, TMPRSS, TMPRSS3, TMPRSS5, TNC, TNFRSF11B, TPRN, TRIOBP, TRMU, TRPV4, TSPEAR, TWNK, TWSG1, TYR, USH1C, USH1G, USH2A, WFS1, WHRN, XPA, YAP1)

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Défice intelectual (painel NGS baseado em exoma com análise de CNVs de 1244 genes - AAAS, AARS1, AASS, ABCC6, ABCC9, ABCD1, ABCD4, ABHD5, ACAD9, ACADM, ACADS, ACAT1, ACO2, ACOX1, ACSL4, ACTB, ACTG1, ACTL6A, ACY1, ADAR, ADGRG1, ADK, ADNP, ADRA2B, ADSL, AFF2, AFF4, AFG3L2, AGA, AGL, AGPS, AGTR2, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AKR1C2, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALS2, ALX3, ALX4, AMER1, AMPD2, AMT, ANK3, ANKH, ANKRD11, AP1S1, AP1S2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARFGEF2, ARG1, ARHGAP31, ARHGEF2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASL, ASPA, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD3A, ATIC, ATL1, ATM, ATP10A, ATP13A2, ATP1A2, ATP1A3, ATP6AP2, ATP6V0A2, ATP6V1B2, ATP7A, ATP8A2, ATR, ATRX, AUH, AUTS2, AVPR1A, AVPR2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCL11A, BCOR, BCS1L, BDNF, BIN1, BLM, BMP4, BOLA3, BPNT2, BRAF, BRAT1, BRIP1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTD, BUB1B, C12orf57, C12ORF65, C19ORF12, C2CD3, C8orf37, CA2, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1H, CACNB4, CACNG2, CAMK2A, CAMK2B, CAMTA1, CANT1, CARS2, CASK, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC115, CCDC22, CCDC78, CCDC8, CCDC88C, CCND2, CDC42, CDC45, CDC6, CDH11, CDH15, CDH3, CDK13, CDK16, CDK5RAP2, CDKL5, CDKN1C, CDON, CDT1, CENPF, CENPJ, CEP104, CEP135, CEP152, CEP290, CEP41, CEP57, CEP63, CEP83, CERT1, CHAMP1, CHD2, CHD4, CHD7, CHD8, CHMP1A, CHRNA2, CHRNA4, CHRNB2, CHSY1, CIC, CISD2, CIT, CKAP2L, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLIC2, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLTC, CNKSR2, CNNM2, CNTNAP2, CNTNAP5, COA8, COASY, COG1, COG4, COG5, COG7, COG8, COL11A1, COL18A1, COL1A1, COL2A1, COL4A1, COL4A2, COL4A3BP, COL9A1, COLEC11, COQ4, COQ8A, COQ9, COX10, COX14, COX15, COX6B1, COX7B, CP, CPA6, CPLANE1, CPS1, CRADD, CRB2, CRBN, CREBBP, CRPPA, CSF1R, CSNK2A1, CSPP1, CSTB, CTC1, CTCF, CTDP1, CTNNB1, CTNND2, CTSA, CTSD, CTSF, CUL4B, CUL7, CYB5R3, CYC1, CYP1B1, CYP27A1, CYP2U1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCC, DCHS1, DCX, DDC, DDHD2, DDOST, DDX11, DDX3X, DEAF1, DENND5A, DEPDC5, DHCR24, DHCR7, DHFR, DHTKD1, DHX30, DIAPH1, DIP2B, DIS3L2, DKC1, DLAT, DLD, DLG3, DLGAP2, DMD, DMXL2, DNAJC12, DNAJC19, DNM1, DNMT3A, DNMT3B, DOCK4, DOCK6, DOCK7, DOCK8, DOLK, DPAGT1, DPM1, DPM3, DPP10, DPP6, DPYD, DYM, DYNC1H1, DYRK1A, EBF3, EBP, EDA, EDNRA, EDNRB, EED, EEF1A2, EFHC1, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2S3, ELAC2, ELOVL4, ELP2, EML1, EMX2, EP300, EPB41L1, EPG5, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERMARD, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXT2, EXTL3, EYA1, EZH2, FAAH2, FAH, FAM111A, FAM126A, FAM20C, FANCB, FANCG, FAR1, FAT4, FBN1, FBN2, FBXL4, FBXO11, FGD1, FGF12, FGF14, FGFR1, FGFR2, FGFR3, FH, FIG4, FKRP, FKTN, FLNA, FLVCR1, FLVCR2, FMN2, FOLR1, FOXC1, FOXE3, FOXG1, FOXP1, FOXP2, FOXRED1, FRAS1, FREM2, FRMPD4, FTCD, FTL, FTO, FTSJ1, FUCA1, G6PC3, GAA, GABRA1, GABRB3, GABRG2, GAD1, GALC, GALE, GALNS, GALT, GAMT, GAN, GATAD2B, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDI1, GFAP, GFER, GFM1, GHR, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLIS3, GLRA1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPAA1, GPC3, GPHN, GPSM2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GRN, GSPT2, GSS, GTF2H5,

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GTPBP3, GUSB, H1-4, HACE1, HADH, HADHA, HAX1, HCCS, HCFC1, HCN1, HDAC4, HDAC6, HDAC8, HECW2, HEPACAM, HERC1, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HIVEP2, HLCS, HMGB3, HNRNPH2, HNRNPK, HNRNPU, HOXA1, HOXC13, HPD, HPGD, HPRT1, HPS1, HRAS, HSD17B10, HSD17B4, HSPD1, HSPG2, HTRA2, HTT, HUWE1, HYLS1, IARS1, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT172, IFT43, IGBP1, IGF1, IGF1R, IGF2, IKBKG, IL11RA, IL1RAPL1, IMMP2L, IMPA1, INPP5E, INPP5K, INSR, IQSEC2, IRF6, IRX5, ITGA7, ITPR1, IVD, JAG1, JAM3, KANSL1, KARS1, KAT6A, KAT6B, KATNAL2, KCNA2, KCNB1, KCNC1, KCNC3, KCNH1, KCNJ10, KCNJ11, KCNJ6, KCNK9, KCNMA1, KCNQ2, KCNQ5, KCNT1, KCTD1, KCTD13, KCTD7, KDM1A, KDM5B, KDM5C, KDM6A, KIAA0586, KIAA1109, KIDINS220, KIF11, KIF1A, KIF1BP, KIF21A, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KLF8, KLHL15, KMT2A, KMT2C, KMT2D, KMT5B, KNL1, KPTN, KRAS, KRIT1, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARP7, LAS1L, LBR, LGI1, LGI4, LHX3, LIG4, LINS1, LMBRD1, LONP1, LRP2, LRP5, LRPPRC, LYST, LZTR1, MAB21L2, MACF1, MAF, MAGEL2, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MAPK8IP3, MARS2, MASP1, MAT1A, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MDH2, MECP2, MECR, MED12, MED13L, MED17, MED23, MEF2C, MEGF8, MFF, MFSD2A, MFSD8, MGAT2, MGP, MICU1, MID1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPDZ, MPI, MPLKIP, MPV17, MPZ, MRAP, MRPS22, MSMO1, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTO1, MTOR, MTR, MTRR, MVK, MYCN, MYO5A, MYO7A, MYT1L, NAA10, NACC1, NAGA, NAGLU, NALCN, NANS, NBN, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA9, NDUFAF2, NDUFAF3, NDUFAF5, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECTIN1, NEDD4L, NEGR1, NEU1, NEXMIF, NF1, NFIA, NFIX, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPBL, NKX2-1, NLGN3, NLGN4X, NONO, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR2F1, NRAS, NRXN1, NRXN2, NSD1, NSDHL, NSUN2, NT5C2, NTNG1, NTRK1, NUBPL, NUP107, NUP62, OBSL1, OCLN, OCRL, OFD1, OGT, OPA3, OPHN1, ORC1, ORC4, ORC6, OTC, OTUD6B, OTX2, PACS1, PAFAH1B1, PAH, PAK3, PANK2, PARK7, PARN, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCDH9, PCGF2, PCNT, PDCD10, PDE10A, PDE4D, PDHA1, PDHX, PDSS1, PDSS2, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PHC1, PHF6, PHF8, PHGDH, PHKA2, PHKG2, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIK3CA, PIK3R2, PINK1, PIP5K1B, PITX2, PITX3, PLA2G6, PLAA, PLCB1, PLCE1, PLK4, PLP1, PLPBP, PMM2, PNKP, PNPLA6, PNPT1, POGZ, POLG, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMT1, POMT2, PON3, PORCN, POU1F1, PPM1D, PPOX, PPP1CB, PPP2R1A, PPP2R5D, PPT1, PQBP1, PRICKLE1, PRKAR1A, PRKD1, PRMT7, PRODH, PRPS1, PRRT2, PRSS12, PRUNE1, PSAP, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTH1R, PTPN11, PTS, PUF60, PURA, PUS1, PYCR1, PYCR2, PYGL, QARS1, QDPR, RAB18, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAB40AL, RAC1, RAD21, RAF1, RAI1, RANBP2, RAPSN, RARB, RARS2, RBBP8, RBFOX1, RBM10, RBPJ, RECQL4, RELN, RERE, RFT1, RFX6, RIT1, RLIM, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, ROGDI, ROR2, RORA, RPGRIP1L, RPL10, RPS23, RPS6KA3, RRM2B, RTEL1, RTN4IP1, RTTN, RUNX2, RXYLT1, SACS, SAMD9, SAMHD1, SATB2, SC5D, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCO1, SCO2, SDCCAG8, SDHA, SDHAF1, SERAC1, SETBP1, SETD5, SETX, SGPL1, SGSH, SHANK1, SHANK2, SHANK3, SHH, SHOC2,

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SHROOM4, SIK1, SIL1, SIN3A, SIX3, SKI, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC2A1, SLC2A2, SLC30A9, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC39A14, SLC39A8, SLC45A1, SLC46A1, SLC4A4, SLC5A5, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A4, SLC6A8, SLC6A9, SLC7A7, SLC9A6, SLC9A9, SLX4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMG6, SMOC1, SMPD1, SMS, SNAP25, SNAP29, SNIP1, SNRPB, SNRPN, SNX14, SOBP, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX5, SOX9, SPART, SPAST, SPATA5, SPG11, SPR, SPRED1, SPTAN1, SPTBN2, SRCAP, SRD5A3, SRPX2, ST3GAL3, ST3GAL5, ST7, STAG1, STAMBP, STAT5B, STIL, STK3, STRA6, STRADA, STT3A, STT3B, STX11, STX1B, STXBP1, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SYP, SYT14, SZT2, TACO1, TAF1, TAF13, TAF2, TAF6, TANGO2, TAT, TAZ, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TCF12, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TECPR2, TECR, TET3, TFAP2A, TFAP2B, TGIF1, TH, THAP1, THOC2, THOC6, THRA, THRB, TIMM8A, TINF2, TK2, TM4SF20, TMCO1, TMEM126B, TMEM165, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TMLHE, TMTC3, TOE1, TP63, TPH2, TPK1, TPP1, TRAPPC9, TREX1, TRHR, TRIM32, TRIM37, TRIO, TRIP12, TRIT1, TRMT10A, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TTC19, TTC37, TTC8, TTI2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, UBA5, UBE2A, UBE3A, UBE3B, UBR1, UBTF, UMPS, UNC80, UPB1, UPF3B, UQCRB, UQCRQ, UROC1, USP27X, USP9X, VAMP1, VLDLR, VPS13B, VRK1, VSX2, WAC, WASHC5, WDPCP, WDR13, WDR19, WDR35, WDR45, WDR45B, WDR62, WDR73, WDR81, WNT5A, WRAP53, WWOX, XPNPEP3, XRCC4, YAP1, YY1, ZBTB16, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZFP57, ZFYVE26, ZIC1, ZIC2, ZMYM3, ZMYND11, ZNF148, ZNF335, ZNF41, ZNF507, ZNF674, ZNF711, ZNF804A, ZNF81, ZNHIT6, ZSWIM6)
Ciliopatias (painel NGS baseado em exoma com CNVs de 222 genes - ACVR2B, ADGRV1, AHI1, AIPL1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ARMC9, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF2, C21orf59, C2CD3, C2orf71, C5orf42, C8orf3, C8ORF37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC172, CCDC28B, CCDC39, CCDC40, CCDC65, CCDC96, CCNO, CCNQ, CDH23, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP295, CEP41, CEP83, CFAP298, CFAP41, CFAP53, CFTR, CLRN1, CPE, CPLANE, CPLANE1, CRB1, CRB2, CRELD1, CRX, CSPP1, DCDC2, DDX59, DEUP1, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H, DYNC2H1, DYNC2I, DYNC2L, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC8, FAM166B, FAM58A, FOXH1, GANAB, GAS2L2, GAS8, GDF1, GLI2, GLI3, GLIS2, GUCY2D, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT80, IFT81, IMPDH1, INPP5E, INVS, IQCB1, KATNIP, KCNJ13, KIAA05, KIAA0556, KIAA0586, KIAA07, KIAA0753, KIF14, KIF7, LCA5, LEFTY2, LRAT, LRRC34, LRRC6, LZTFL1, MAPKBP, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, MYO7A, NEK1, NEK8, NKX2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCDH15, PDE6D, PIEZO2, PIH1D3, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, RD3, RDH12, RPE65, RPGR, RPGRIP, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCAPER, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG, SDCCAG8, SPAG1, SPATA7, TCTEX1D2, TCTN1,

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TCTN2, TCTN3, TMEM10, TMEM107, TMEM13, TMEM138, TMEM17, TMEM21, TMEM216, TMEM23, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3I, TRAF3IP1, TRIM32, TSC1, TSC2, TTC21B, TTC23, TTC25, TTC6, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, USP9X, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WHRN, XPNPEP3, ZIC3, ZMYND10, ZNF423)
Baixa estatura (painel NGS baseado em exoma com CNVs de 166 genes - ACAN, ACTB, ACTG1, ADAMTS10, ADAMTS2, AGA, AGPS, ALDH18A1, ALMS1, AMER1, AMMECR1, ANTXR2, ARCN1, ARID1B, ATP8B1, ATR, B3GALT6, B3GAT3, BCS1L, BRAF, CANT1, CBL, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, CKAP2L, COL10A1, COL1A1, COL1A2, COL27A1, COL2A1, COL9A2, COL9A3, COMP, COX7B, CREBBP, CRIPT, CTNNB1, CTSA, CUL7, DHCR24, DHCR7, DLL3, DLX5, DONSON, DVL1, DVL3, EP300, ERCC4, ERCC6, EVC, EVC2, FAM111A, FBN1, FGD1, FGFR1, FGFR3, FLNB, FN1, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNAS, GORAB, HADH, HCCS, HDAC8, HESX1, HHAT, HOXD13, HRAS, HSPA9, IDS, IDUA, IGF1, IGF1R, IGFALS, INPP5K, INPPL1, INSR, IRS1, KAT6B, KIF11, KRAS, LARP7, LBR, LHX3, LHX4, LTBP3, LZTR1, MAP2K1, MAP2K2, MAP3K7, MBTPS2, MESP2, MYO18B, NDUFB11, NIN, NIPBL, NOTCH2, NPR2, NRAS, OBSL1, ORC1, ORC4, ORC6, OTX2, PCNT, PCYT1A, PDE4D, PEX7, PIEZO2, PIK3R1, PITX2, PNPLA6, POC1A, POU1F1, PRKAR1A, PROP1, PTDSS1, PTH1R, PTPN11, PYCR1, RAD21, RAF1, RASA2, RBBP8, RIT1, RNU4ATAC, RRAS, RTTN, RUNX2, SHOC2, SHOX, SIL1, SLC26A2, SLC39A13, SMAD4, SMARCA2, SMC1A, SMC3, SOS1, SOX2, SOX3, SRCAP, STAT5B, TBCE, TBX19, TBX3, TBX6, TCTN3, TKT, TRIM37, TRIP11, TRPV4, WNT5A, XRCC4, XYLT1)
Miocardiopatia dilatada (painel NGS baseado em exoma com CNVs de 128 genes - ABCC6, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, APOA1, BAG3, CASQ2, CASZ1, CAV3, CAVIN4, CHRM2, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, EYA4, FBXO32, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXD4, GATA4, GATA6, GATAD1, GATC, GBE1, GLA, GLB1, GSK3B, HAMP, HAND1, HCN4, HFE, HJV, IDH2, ILK, JPH2, JUP, KLHL24, LAMA4, LAMP2, LDB3, LEMD2, LMNA, LMOD2, LRRC10, MIB1, MLYCD, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, NRAP, PCCA, PCCB, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PRKAG2, PTPN11, QRSL1, RAB3GAP2, RAF1, RBCK1, RBM20, RMND1, RYR2, SCN5A, SDHA, SGCD, SLC22A5, SLC40A1, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TFR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TTN, TTR, TXNRD2, VCL, VPS13A)
Miocardiopatia hipertrófica (painel NGS baseado em exoma com CNVs de 116 genes - A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALPK3, ANKRD1, APOA1, ATP5F1E, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COA5, COX15, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EPG5, FBXL4, FHL1, FHOD3, FKTN, FLNC, FOXRED1, GAA, GATA4, GATAD1, GLA, GLB1, GSK3B, GUSB, HRAS, ILK, JPH2, JUP, KCNQ1, KLF10, KLHL24, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MIPEP, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NDUFAF2, NEBL, NEXN, NF1, NRAS, OBSCN, PDLIM3, PKP2, PLN, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RRAS, RYR2, SCN5A, SCO2, SGCD, SHOC2, SLC25A3, SLC25A4, SOS1, SOS2, SPRED1,

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TAZ, TCAP, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, VCL)
Síndrome de Alport (painel NGS baseado em exoma com CNVs de 11 genes - CD151, CFHR5, COL4A3, COL4A4, COL4A5, COL4A6, FN1, LMX1B, MYH9, MYO1E, NPHS2)
Síndrome de Bartter (painel NGS baseado em exoma com CNVs de 26 genes - ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4)
Síndrome nefrótico (painel NGS baseado em exoma com CNVs de 59 genes - ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, FAT1, FN1, INF2, ITGA3, ITGB4, KANK1, KANK2, LAGE3, LAMB2, LMX1B, MAFB, MAGI2, MYH9, MYO1E, NEU1, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OSGEP, PAX2, PDCN, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TP53RK, TPRKB, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5)

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