



Gen med

Catálogo de pruebas genómicas
por especialidad

Neonatología

Neonatología

Panel para Recién Nacidos

Panel completo de NGS que incluye genes seleccionados explícitamente para las pruebas genéticas de recién nacidos en estado crítico y niños menores de 24 meses en unidades de cuidados intensivos (UCI). Está diseñado para abordar múltiples condiciones genéticas que pueden estar presentes en el período del recién nacido o en la primera infancia, y muchas tienen fenotipos superpuestos e implicaciones inmediatas para el inicio del tratamiento.

Incluye 855 genes: AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMPI, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALGI, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTB, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM1, CHRNA1, CHRN1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSB, CTSN, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOX2A, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBPI, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKBP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALT, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAPI, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GPIBA, GPIBB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGFIR, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSB8, MITE, MKKS, MLC1, MLYCD, MMLA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMD1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUF51, NDUF52, NDUF53, NDUF54, NDUF56, NDUF57, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPH1, NPH3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1,

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PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STING1, STS, STT3B, STXBPI, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBCID24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423