Genomed

Catálogo de pruebas genómicas
por especialidad

Desórdenes metabólicos

Gen med

Laboratorio de diagnóstico clínico y genómico

Desordenes metabólicos

CentoIEM

Los errores congénitos del metabolismo tienen un gran impacto en las enfermedades humanas. CentoIEM incluye una gran variedad de trastornos diferentes e incluye genes responsables de diversos fenotipos, incluido el metabolismo intermediario, como aminoacidopatías, acidurias orgánicas, trastornos del ciclo de la urea, intolerancia al azúcar, trastornos mentales y porfirias, entre otros. También se incluyen los procesos energéticos citoplasmáticos y mitocondriales y el metabolismo que afectan a los orgánulos celulares, como la síntesis lisosomal, peroxisomal, de glicosilación y de colesterol.

Incluye 744 genes: AARS2, ABCA1, ABCB4, ABCC2, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADA, ADAMTS10, ADAMTSL2, ADAR, ADGRG1, ADK, ADSL, AFG3L2, AGA, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AIMP1, AIMP2, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALS2, AMN, AMPD2, AMT, ANK1, ANTXR2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA2, APOA5, APOB, APOC2, APOE, APP, APPL1, APTX, AQP2, ARG1, ARL6IP1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATM, ATP13A2, ATP6V0A2, ATP7A, ATP7B, ATPAF2, ATRX, AUH, AVP, AVPR2, B3GALNT2, B4GALNT1, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BEST1, BICD2, BLK, BMP6, BOLA3, BRAT1, BSCL2, BTD, C19orf12, CA5A, CACNA1D, CAPN1, CASP10, CASP8, CAV1, CAVIN1, CBLIF, CBS, CCT5, CD320, CEL, CERS1, CETP, CISD2, CLCN2, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPOX, CPS1, CPT1A, CPT1C, CPT2, CSF1R, CTC1, CTH, CTLA4, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CYP11B1, CYP17A1, CYP19A1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DDC, DDHD1, DDHD2, DDOST, DGUOK, DHCR7, DHDDS, DIABLO, DKC1, DLAT, DLD, DLL3, DNAJC5, DNM1L, DOLK, DPM1, DPM2, DPM3, DPYD, DSTYK, DYM, EARS2, ECHS1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ENO3, ENPP1, ENTPD1, EPB42, EPHX2, EPM2A, EPRS1, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, F2, F5, FA2H, FADD, FAH, FAM126A, FARS2, FARSB, FAS, FASLG, FASTKD2, FBN1, FBP1, FBXL4, FDX2, FECH, FGF23, FH, FHL1, FLAD1, FOLR1, FOXA2, FOXP3, FOXRED1, FTL, FUCA1, G6PC1, G6PD, GAA, GABRB2, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GATA4, GATA6, GATM, GBA, GBA2, GBE1, GCDH, GCK, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJB1, GJC2, GK, GLA, GLB1, GLDC, GLIS3, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GRN, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HACE1, HADH, HADHA, HADHB, HAMP, HCFC1, HEPACAM, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIKESHI, HJV, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HSD3B2, HSPD1, HTRA1, HYAL1, IARS2, IBA57, IDS, IDUA, IER3IP1, IFIH1, IL2RA, INS, INSR, ISCA2, ITIH4, ITK, IVD, JAG1, JAM3, KCNC1, KCNJ10, KCNJ11, KCNT1, KCTD7, KDM6A, KHK, KIDINS220, KIF1A, KIF1C, KIF5A, KLF11, KMT2D, KRAS, L1CAM, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LAT, LCAT, LDB3, LDHA, LDLR, LDLRAP1, LIAS, LIPA, LIPC, LIPE, LIPI, LIPT1, LIPT2, LMBRD1, LMNA, LMNB1, LPIN1, LPL, LRBA, LRPPRC, LYRM7, LYST, MAG, MAGT1, MAN1B1, MAN2B1, MANBA, MARS1, MARS2, MCCC1, MCCC2, MCEE, MCOLN1, MECR, MFSD8, MGAT2, MGME1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPL44, MRPS22, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTFMT, MTHFR, MTPAP, MTR, MTRFR, MTRR, MYO5A, MYOT, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFE2L2, NFU1, NGLY1, NHLRC1, NIPA1, NKX2-2, NKX6-2, NOTCH3, NPC1, NPC2, NPR2, NRAS, NT5C2, NUBPL, OAT, OCLN, OCRL, OPA3, OSGEP, OTC, OXCT1, PAH, PANK2, PAX4, PC, PCCA, PCCB, PCK1, PCSK9, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIK3R1, PKLR, PLA2G6, PLAA, PLCG2, PLIN1, PLP1, PLPBP, PMM2, PMPCB, PNPLA6, PNPO, PNPT1, POLG, POLR1C, POLR3A, POLR3B, POR, PPARG, PPOX, PPP1R17, PPT1, PRF1, PRICKLE1, PRKAG2, PRKCD, PRODH, PSAP, PSEN1, PTF1A, PTS, PYCR2, PYGL, PYGM, QDPR, RAB11B, RAB27A, RAB3GAP2, RAI1, RARS1, RARS2, RBCK1, REEP1,REEP2, RFT1, RFX6, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPIA, RRM2B, RTN2, SACS,

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SAMHD1, SCARB2, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SERPINI1, SGSH, SI, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC3A1, SLC40A1, SLC4A1, SLC6A19, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNTA1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTA1, SPTB, SRD5A3, SSR4, STAT1, STAT3, STT3A, STT3B, STX11, STXBP2, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, SYNE1, TACO1, TAFAZZIN, TAT, TBC1D24, TCF4, TCN2, TECPR2, TFG, TFR2, TGFB1, TINF2, TK2, TMEM106B, TMEM165, TMEM70, TPK1, TPP1, TREM2, TREX1, TRMT10A, TRPV4, TSFM, TTC19, TUBB4A, TUFM, TUSC3, TWNK, TYMP, TYROBP, UBAP1, UCHL1, UCP2, UFM1, UGT1A1, UMPS, UNC13D, UNC80, UQCRQ, UROD, UROS, USH1C, VAMP1, VCP, VPS11, VPS37A, WARS2, WASHC5, WDR45, WDR45B, WFS1, ZFP57, ZFYVE26, ZFYVE27

CentoUCI

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, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, 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, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDCA8, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, 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, CTSA, CTSD, 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, DUOXA2, 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, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXE1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, 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, IGF1R, IGHMBP2, IGLL1, IGSF1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, IRS4, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, IYD, 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, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, , MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRFR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1

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NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, ODAD1, OPA3, OPHN1, ORC1, ORC4, OTC, OTX2, 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, PRKAR1A, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, 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, SECISBP2, 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, SLC26A4, 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, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAFAZZIN, TAT, TBC1D24, TBCE, TBL1X, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, THRB, TJP2, TMCO1, TMEM165, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRH, TRHR, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB1, TUBB2A, TWNK, UBA1, UBR1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423

CentoMito

Incluye 37 genes mitocondriales: MT-ND1, MT-ND2, MT-CO1, MT-CO2, MT-ATP8, MT-ATP6, MT-CO3, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-TF, MT-RNR1, MT-TV, MT-RNR2, MT-TL1, MT-TI, MT-TQ, MT-TM, MT-TW, MT-TA, MT-TN, MT-TC, MT-TY, MT-TS1, MT-TD, MT-TK, MT-TG, MT-TR, MT-TH, MT-TS2, MT-TL2, MT-TE, MT-TT, MT-TP

CentoMito integral

Cubre todo el genoma mitocondrial con detección de heteroplasmia hasta en un 5 % junto con genes nucleares relacionados con enfermedades mitocondriales. Las enfermedades mitocondriales son condiciones genéticas que ocurren cuando las mitocondrias no pueden producir suficiente energía para la célula. Las mutaciones genéticas relacionadas con las mitocondrias provocan síntomas principalmente en los órganos, donde el consumo energético es elevado. Estos órganos incluyen el ojo, el riñón, el páncreas, la sangre, el oído interno, el colon, el músculo esquelético, el corazón y el cerebro.

Incluye 450 genes: AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, ADAR, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMPD1, AMT, APTX, ATIC, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BAG3, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BTD, C19orf12, C1QBP, CA5A, CARS2, CAT, CAVIN1, CEL, CHAT, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COA7, COA8, COASY, COMT, COQ2, COQ4, COQ6, COQ7, 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, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2,

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Laboratorio de diagnóstico clínico y genómico

Desordenes metabólicos

FASTKD2, FBXL4, FDX2, FECH, FH, FKBP10, FLAD1, FOXRED1, FXN, GAMT, GARS1, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, IVD, KARS1, KRT5, KRT8, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MSRB3, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTFMT, MTHFD1, MTO1, MTPAP, MTRR, NADK2, NAGS, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NGLY1, NNT, NR2F1, NTHL1, NUBPL, NUP62, OAT, OGDH, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PMPCA, PMPCB, PNKD, PNPLA8, PNPO, PNPT1, POLG, POLG2, POP1, PPOX, PRODH, PSAP, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS1, RARS2, RDH11, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RPIA, RPL35A, RPS14, RRM2B, SACS, SARS2, SBDS, SCN1A, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC6A8, SLC9A6, SNAP29, SOD1, SOD2, SPAST, SPG7, SPR, SPTLC2, STAR, STAT2, STXBP1, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TAFAZZIN, TANGO2, TCIRG1, TFR2, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TMLHE, TOP3A, TPI1, TPK1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TWNK, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WDR81, WFS1, XPNPEP3, YARS2

Panel Diabetes y Obesidad

Recomendado para pacientes con anomalías en el metabolismo de la glucosa, como hipoglucemia hiperinsulinémica, diabetes neonatal, MODY, diabetes en adultos e hipercolesterolemia familiar, así como para pacientes que presentan resistencia a la insulina, desde el espectro leve hasta el severo (síndrome de Donohue), y para pacientes con hiperinsulinismo familiar. Los trastornos causados por errores de impresión o disomía uniparental, como la diabetes mellitus neonatal transitoria relacionada con 6q24 y el síndrome de Beckwith Wiedemann, no se detectan con este panel.

Incluye 265 genes: ABCA1, ABCC8, ABCG5, ABCG8, ACAT1, ACSF3, ADCY3, AFF4, AGL, AGRP, AIP, AIRE, AKT2, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG3, ALG6, ALG8, ALG9, ALMS1, ANGPTL3, APOA1, APOA5, APOB, APOC2, APOC3, APOE, APPL1, AQP2, ARL13B, ARL6, ARMC5, ATP6V0A2, AVP, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, BLK, CANT1, CC2D2A, CCDC28B, CEL, CEP164, CEP19, CEP290, CETP, CFAP418, CHD2, CISD2, CNOT1, COG1, COG4, COG5, COG6, COG7, COG8, CP, CPE, CREBBP, CUL4B, CYP27A1, DCAF17, DNAJC3, DOLK, DPM1, DPM2, DPM3, DYRK1B, EHMT1, EIF2AK3, EIF2B1, EIF2S3, ENO3, ENPP1, EPM2A, FBP1, FOXP3, GAA, GATA6, GBE1, GCK, GCKR, GH1, GHR, GHRHR, GHRL, GLI3, GLIS3, GLUD1, GMPPA, GNAS, GNE, GPC3, GPD1, GPIHBP1, GYS1, GYS2, H6PD, HADH, HEXA, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HSD11B1, IER3IP1, IFT172, IFT27, IGF1R, INPP5E, INS, INSR, ITCH, KCNJ11, KIDINS220, KIF7, KLF11, KMT2C, KSR2, LAMP2, LARGE1, LAS1L, LDHA, LDLR, LDLRAP1, LEP, LEPR, LIPA, LIPC, LIPE, LMF1, LMNA, LPL, LZTFL1, MAGEL2, MAN1B1, MC3R, MC4R, MCHR1, MEGF8, MGAT2, MKKS, MKRN3, MKS1, MOGS, MPDU1, MPI, MPV17, MRAP2, MTNR1B, MTTP, MYO5A, MYO7A, MYT1L, NDN, NEUROD1, NEUROG3, NGLY1, NHLRC1, NKX2-2, NPHP1, NPHP3, NR0B2, NSD1, NTRK2, OFD1, OXCT1, PAX4, PAX6, PC, PCBD1, PCK1, PCNT, PCSK1, PCSK9, PDE11A, PDE4D, PDX1, PFKM, PGAM2, PGK1, PGM1, PHF6, PHIP, PHKA1, PHKA2, PHKB, PHKG2, PIK3R1, PMM2, PNPLA6, POLD1, POMC, PPARG, PPP1R15B, PRKAG2, PRKAR1A, PRMT7, PROM1, PRPH2, PTEN, PTF1A, PYGL, PYGM, RAB23, RAI1, RBCK1, RDH5, RFT1, RFX6, RHO, RLBP1, RPGRIP1L, RPS6KA3, SDCCAG8, SETD2, SH2B1, SIM1, SLC16A1, SLC19A2, SLC29A3, SLC2A2, SLC35A1, SLC35A2, SLC35C1, SNRPN, SPG11, SRD5A3, SSR4, STAT1, STAT3, TBX3, THOC2, THRA, TMEM165, TMEM67, TRAF3IP1, TRAPPC9, TRIM32, TRMT10A, TTC21B, TTC8, TUSC3, UCP2, UCP3, VPS13B, WFS1, XRCC4, XYLT1, ZBTB20, ZFP57, ZMPSTE24, ZNF711