Genomed

Catálogo de pruebas genómicas
por especialidad

Neurología

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

Panel de esclerosis lateral amiotrófica (ELA)/demencia

incluye genes que causan la enfermedad de Alzheimer, la demencia y la demencia frontotemporal, así como genes utilizados para el diagnóstico diferencial con superposición en cualquier punto de la historia natural de la enfermedad. Los genes dentro de este panel se han seleccionado cuidadosamente para aumentar el rendimiento del diagnóstico. Se incluyen enfermedades procesables que se superponen con el fenotipo (como la enfermedad de Wilson, la enfermedad de Niemann-Pick y la deficiencia de hexosaminidasa A). Este panel no detecta la enfermedad de Huntington.

Incluye 105 genes: ABCA7, ALS2, ANG, ANXA11, APOE, APP, ARSA, ATL1, ATP7B, ATXN2, BSCL2, C9orf72, CCNF, CHCHD10, CHMP2B, CP, CSF1R, CYLD, CYP27A1, DCTN1, ERBB4, EWSR1, FIG4, FTL, FUS, GLE1, GRN, HEXA, HNRNPA1, HNRNPA2B1, HSPD1, ITM2B, KIF5A, MAPT, MATR3, 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, NEFH, NEK1, NOTCH3, NPC1, OPTN, PANK2, PFN1, PRNP, PRPH, PSEN1, PSEN2, REEP1, SETX, SIGMAR1, SLC52A3, SNCA, SOD1, SORL1, SPAST, SPG11, SQSTM1, TAF15, TARDBP, TBK1, TFG, TREM2, TUBA4A, TYROBP, UBE3A, UBQLN2, VAPB, VCP, WASHC5

Genes de expansión de repetición: C9orf72, ATXN2, PRNP

Panel de expansión de repetición de ataxia

Incluye genes relevantes para trastornos neurológicos hereditarios caracterizados por ataxia, incluida la ataxia espinocerebelosa (dominante y recesiva), ataxia cerebelosa, ataxia episódica y ataxia pontocerebelosa. Estos trastornos normalmente comparten síntomas superpuestos y solo pueden diferenciarse claramente mediante pruebas genéticas moleculares. Nuestro panel de ataxia es la mejor opción para un paciente que muestra un desequilibrio en la marcha y una marcha descoordinada (ataxia). Las formas más comunes de ataxia hereditaria son causadas por expansión repetida.

Incluye 13 genes: ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP

Genes de expansión de repetición: ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP

Panel completo de ataxia/paraplejía espástica

Incluye 492 genes: ATXN1, ATXN2, ATXN3, ATXN7, BEAN1, CACNA1A, PPP2R2B, TBP, ATXN10, ATN1, NOP56, FXN, ATXN8OS, AARS2, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADVL, ACO2, ADAR, ADPRS, AFG3L2, AGK, AGTPBP1, AHI1, AIFM1, AIMP1, ALAS2, ALDH18A1, ALDH5A1, ALS2, AMACR, AMPD2, ANO10, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARL13B, ARL6, ARL6IP1, ARSA, ATCAY, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP2B4, ATP7B, ATP8A2, ATRX, AUH, B4GALNT1, B9D1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BICD2, BLOC1S1, BOLA3, BSCL2, BTD, C19orf12, CA8, CACNA1G, CACNB4, CAMTA1

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

CAPN1, CARS2, CASK, CC2D2A, CCDC88C, CCT5, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPB, CLPP, COA6, COA7, COA8, COASY, COL4A1, COL4A2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, CP, CPT1C, CSPP1, CSTB, CWF19L1, CYC1, CYP27A1, CYP2U1, CYP7B1, DAB1, DARS1, DARS2, DBT, DDHD1, DDHD2, DGUOK, DHPS, DLAT, DLD, DNA2, DNAJC19, DNAJC5, DNM1L, DNMT1, DOCK3, DSTYK, EARS2, EBF3, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELOVL4, ELOVL5, ENTPD1, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FARS2, FASTKD2, FAT2, FBXL4, FDX2, FDXR, FGF14, FH, FLAD1, FLVCR1, FOXRED1, FTL, GAD1, GALC, GARS1, GBA, GBA2, GCDH, GCH1, GFAP, GFER, GFM1, GFM2, GJB1, GJC2, GLRX5, GOSR2, GRID2, GRM1, GSS, GTPBP3, HACE1, HARS2, HEPACAM, HEXA, HEXB, HIBCH, HMGCL, HSPD1, HTRA2, IARS2, IBA57, INPP5E, IRF2BPL, ISCA2, ISCU, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KIF7, L1CAM, LAMA1, LAMP2, LARS2, LIAS, LIPT1, LMNB1, LRPPRC, LYRM7, LYST, MAG, MARS1, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MKKS, MKS1, MLC1, MPC1, MPV17, MRE11, MRPL3, MRPL44, MRPS16, MRPS22, 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, MTO1, MTPAP, MTRFR, MTTP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NIPA1, NKX6-2, NPC1, NPC2, NPHP1, NR2F1, NT5C2, NUBPL, OFD1, OPA1, OPA3, OPHN1, OTC, PANK2, PARS2, PAX6, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX10, PEX2, PEX7, PGAP1, PHYH, PIK3R5, PLA2G6, PLK1, PLP1, PMPCA, PNKD, PNKP, PNPLA6, PNPT1, POLG, POLG2, POLR3A, POLR3B, PRICKLE1, PRKCG, PRRT2, PUM1, QARS1, RAB3GAP2, RAP1GDS1, RARS1, RARS2, REEP1, REEP2, RMND1, RNASEH1, RNASEH2B, RNF216, RPGRIP1L, RRM2B, RTN2, RUBCN, SACS, SAMD9L, SARS2, SCN1A, SCN2A, SCO1, SCO2, SCYL1, SDHA, SDHAF1, SELENOI, SERAC1, SETX, SFXN4, SIL1, SLC16A2,SLC17A5, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A19, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC9A6, SNX14, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTBN2, STUB1, SUCLA2, SUCLG1, SURF1, SYNE1, TACO1, TARS2, TBC1D24, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TFG, TGM6, TIMM8A, TK2, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TPK1, TPP1, TRIM32, TRIT1, TRMT10C, TRNT1, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC8, TTPA, TTR, TUBB4A, TUFM, TWNK, TYMP, UBA5, UBAP1, UBE3A, UBTF, UCHL1, UNC80, UQCC2, UQCRB, UQCRC2, UQCRQ, USP8, VAMP1, VARS2, VCP, VLDLR, VPS37A, VRK1, WASHC5, WDR45, WDR45B, WDR81, WFS1, WWOX, YARS2, ZFYVE26, ZFYVE27, ZNF423

Genes de expansión de repetición: ATXN1, ATXN2, ATXN3, ATXN7, BEAN1, CACNA1A, PPP2R2B, TBP, ATXN10, ATN1, NOP56, FXN, ATXN8OS

Panel de ataxia/paraplejía espástica

Incluye 481 genes: AARS2, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADVL, ACO2, ADAR, ADPRS, AFG3L2, AGK, AGTPBP1, AHI1, AIFM1, AIMP1, ALAS2, ALDH18A1, ALDH5A1, ALS2, AMACR, AMPD2, ANO10, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARL13B, ARL6, ARL6IP1, ARSA, ATCAY, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP2B4, ATP7B, ATP8A2, ATRX, AUH, B4GALNT1, B9D1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BICD2, BLOC1S1, BOLA3, BSCL2, BTD, C19orf12, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CASK, CC2D2A, CCDC88C, CCT5, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPB, CLPP, COA6, COA7, COA8, COASY, COL4A1, COL4A2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, CP, CPT1C, CSPP1, CSTB, CWF19L1, CYC1, CYP27A1, CYP2U1, CYP7B1, DAB1, DARS1, DARS2, DBT, DDHD1, DDHD2, DGUOK, DHPS, DLAT, DLD, DNA2, DNAJC19, DNAJC5, DNM1L, DNMT1, DOCK3, DSTYK, EARS2, EBF3, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELOVL4, ELOVL5, ENTPD1, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FARS2, FASTKD2, FAT2, FBXL4, FDX2, FDXR, FGF14, FH, FLAD1, FLVCR1, FOXRED1, FTL, FXN, GAD1, GALC, GARS1, GBA, GBA2, GCDH, GCH1, GFAP, GFER, GFM1, GFM2, GJB1, GJC2, GLRX5, GOSR2, GRID2, GRM1, GSS, GTPBP3, HACE1, HARS2, HEPACAM, HEXA, HEXB, HIBCH, HMGCL, HSPD1, HTRA2, IARS2, IBA57, INPP5E, IRF2BPL, ISCA2, ISCU, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KIF7, L1CAM, LAMA1, LAMP2, LARS2, LIAS, LIPT1, LMNB1, LRPPRC, LYRM7, LYST, MAG, MARS1, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MKKS, MKS1, MLC1, MPC1, MPV17, MRE11, MRPL3, MRPL44, MRPS16, MRPS22, 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, MTO1, MTPAP, MTRFR, MTTP, NARS2, NDUFA1, NDUFA10,

Neurología

NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NIPA1, NKX6-2, NPC1, NPC2, NPHP1, NR2F1, NT5C2, NUBPL, OFD1, OPA1, OPA3, OPHN1, OTC, PANK2, PARS2, PAX6, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX10, PEX2, PEX7, PGAP1, PHYH, PIK3R5, PLA2G6, PLK1, PLP1, PMPCA, PNKD, PNKP, PNPLA6, PNPT1, POLG, POLG2, POLR3A, POLR3B, PRICKLE1, PRKCG, PRRT2, PUM1, QARS1, RAB3GAP2, RAP1GDS1, RARS1, RARS2, REEP1, REEP2, RMND1, RNASEH1, RNASEH2B, RNF216, RPGRIP1L, RRM2B, RTN2, RUBCN, SACS, SAMD9L, SARS2, SCN1A, SCN2A, SCO1, SCO2, SCYL1, SDHA, SDHAF1, SELENOI, SERAC1, SETX, SFXN4, SIL1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A19, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC9A6, SNX14, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTBN2, STUB1, SUCLA2, SUCLG1, SURF1, SYNE1, TACO1, TARS2, TBC1D24, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TFG, TGM6, TIMM8A, TK2, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TPK1, TPP1, TRIM32, TRIT1, TRMT10C, TRNT1, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC8, TTPA, TTR, TUBB4A, TUFM, TWNK, TYMP, UBA5, UBAP1, UBE3A, UBTF, UCHL1, UNC80, UQCC2, UQCRB, UQCRC2, UQCRQ, USP8, VAMP1, VARS2, VCP, VLDLR, VPS37A, VRK1, WASHC5, WDR45, WDR45B, WDR81, WFS1, WWOX, YARS2, ZFYVE26, ZFYVE27, ZNF423

Panel de la enfermedad de Parkinson

Identifica todas las variantes genéticas fisiopatológicamente relevantes para el desarrollo y el tratamiento de la EP. Los rasgos característicos de la EP incluyen la pérdida neuronal en áreas específicas de la sustancia negra y la acumulación generalizada de proteína sinucleína intracelular. La enfermedad se caracteriza por tres síntomas motores centrales: temblor, rigidez muscular y bradicinesia.

Incluye 115 genes: APP, ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, CHCHD10, CHCHD2, CLN3, COASY, CP, CSF1R, CYP27A1, DCAF17, DCTN1, DNAJB2, DNAJC12, DNAJC13, DNAJC5, DNAJC6, EIF4G1, FBXO7, FTL, FUS, GBA, GCH1, GIGYF2, GRN, HTRA2, JAM2, KIF5A, LRP10, LRRK2, LYST, MAPT, 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, MYORG, NPC1, NPC2, NUS1, PANK2, PARK7, PDE10A, PDE8B, PDGFB, PDGFRB, PINK1, PLA2G6, POLG, POLG2, PRKN, PRKRA, PSEN1, PSEN2, PTS, RAB39B, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SMPD1, SNCA, SNCB, SPG11, SPR, SYNJ1, TAF1, TARDBP, TENM4, TH, TMEM230, TWNK, UCHL1, VPS13A, VPS13C, VPS35, WDR45, XPR1, ZFYVE26

CentoNeuro

Diseñado para detectar una gran variedad de trastornos neurológicos, desde casos de UCI neonatal hasta demencia o trastornos del movimiento en adultos. Este panel incluye genes relacionados con enfermedades neurológicas, como esclerosis lateral amiotrófica, demencia, Parkinson, enfermedades neuromusculares, Charcot-Marie-Tooth, distonía, epilepsia, autismo, discapacidad intelectual, migraña, paraplejía espástica, ataxia, síndrome de Leigh, peroxisomal enfermedades, encefalopatías epilépticas y trastornos del movimiento, entre otras. Limitaciones: Si existe una alta sospecha diagnóstica de distrofia muscular de Duchenne, recomendamos que el médico ordene un análisis de deleción/duplicación por MLPA dirigido al gen DMD como un servicio adicional.

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

Incluye 1902 genes: AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA7, ABCB6, ABCB7, ABCC6, ABCC8, ABCD1, ABCD3, ABCD4, ABHD12, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACHE, ACO2, ACOX1, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTG2, ACTL6B, ACTN4, ACVRL1, ACY1, ADA, ADAM10, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADAT3, ADCY5, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRS, ADSL, AFF2, AFF3, AFG3L2, AGA, AGK, AGL, AGPS, AGRN, AGTPBP1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AK2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMMECR1, AMPD1, AMPD2, AMT, ANG, ANK2, ANK3, ANKLE2, ANKRD11, ANO10, ANO3, ANO5, ANTXR2, ANXA11, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOE, APP, APTX, ARFGEF2, ARG1, ARHGAP31, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARL6IP1, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN2, ASXL1, ASXL3, ATAD1, ATCAY, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B3, ATP2B4, ATP5F1A, ATP5F1E, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT1, B4GAT1, B9D1, B9D2, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BDNF, BEST1, BICD2, BIN1, BLOC1S1, BLOC1S3, BLOC1S6, BOLA3, BRAF, BRAT1, BRWD3, BSCL2, BSND, BTD, BVES, C12orf4, C12orf57, C19orf12, C1QBP, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1H, CACNA1S, CACNA2D2, CACNB2, CACNB4, CAD, CAMK2A, CAMK2B, CAMK2G, CAMTA1, CAPN1, CAPN3, CARD11, CARS2, CASK, CASQ1, CASR, CAT, CAV1, CAV3, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCDC115, CCDC22, CCDC40, CCDC78, CCDC88A, CCDC88C, CCM2, CCNF, CCT5, CD320, CD59, CD96, CDH11, CDH15, CDK5RAP2, CDKL5, CDON, CEL, CENPF, CENPJ, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CERS1, CERT1, CFAP418, CFL2, CHAMP1, CHAT, CHCHD10, CHCHD2, CHD1, CHD2, CHD3, CHD7, CHD8, CHKB, CHL1, CHMP1A, CHMP2B, CHRNA1, CHRNA2, CHRNA4, CHRNA7, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHST14, CHSY1, CIB2, CIC, CILK1, CISD2, CIT, CLCN1, CLCN2, CLCN4, CLCNKA, CLCNKB, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNBP, CNGB3, CNKSR2, CNNM2, CNPY3, CNTN4, CNTNAP1, CNTNAP2, COA5, COA6, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL12A1, COL13A1, COL18A1, COL2A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COLGALT1, COLQ, COMT, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CP, CPA6, CPLX1, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRADD, CRBN, CREBBP, CRIPT, CRLF1, CRPPA, CRYAB, CSF1R, CSMD1, CSNK2B, CSPP1, CSRP3, CST3, CSTB, CTC1, CTCF, CTDP1, CTNNA2, CTNNA3, CTNNB1, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUL3, CUL4B, CUL7, CUX1, CUX2, CWF19L1, CX3CR1, CYB5A, CYB5R3, CYC1, CYCS, CYFIP2, CYLD, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, CYP2U1, CYP7B1, D2HGDH, DAB1, DAG1, DARS1, DARS2, DBT, DCAF17, DCTN1, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHH, DHODH, DHPS, DHTKD1, DHX30, DIABLO, DIAPH1, DIAPH3, DIP2B, DKC1, DLAT, DLD, DLG3, DLG4, DLGAP2, DLL3, DLX3, DMD, DMGDH, DMPK, DMXL2, DNA2, DNAJB2, DNAJB6, DNAJC12, DNAJC13, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DOCK3, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPF2, DPM1, DPM2, DPM3, DPP6, DPYD, DPYS, DRD3, DST, DSTYK, DTNBP1, DVL3, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, EDC3, EDN3, EDNRB, EEF1A2, EFHC1, EFTUD2, EGF, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4G1, ELAC2, ELOVL4, ELOVL5, ELP1, ELP2, EMC10, EMD, EML1, EMX2, ENO3, ENTPD1, EP300, EPB41L1, EPG5, EPHX2, EPM2A, EPRS1, ERBB4, ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EWSR1, EXOC6B, EXOSC3, EXOSC8, EXOSC9, EXT1, EZH2, F2, F5, FA2H, FADD, FAH, FAM126A, FAN1, FANCB, FARS2, FARSB, FASTKD2, FAT2, FBLN5, FBN1, FBN2, FBXL4, FBXO11, FBXO38, FBXO7, FDX2, FDXR, FECH, FEZF1, FGA, FGD1, FGD4, FGF10, FGF12, FGF14, FGFR2, FGFR3, FH, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNC, FLVCR1, FLVCR2, FMN2, FOLR1, FOXC1, FOXG1, FOXL2, FOXP1, FOXP2, FOXRED1, FRMD7, FRMPD4, FRRS1L, FTL, FTO, FTSJ1, FUCA1, FUS, FUT8, FXN, FXR1, FXYD2, G6PD, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRE, GABRG2, GAD1, GALC, GALNS, GALT, GAMT, GAN, GARS1, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GDI1, GDNF, GFAP, GFER, GFM1, GFM2, GFPT1, GIGYF2, GJA1, GJB1, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLO1, GLRA1, GLRB, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNAQ, GNAS, GNB1, GNB4, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, , GPAA1, GPC3, GPC4, GPC6, GPHN, GPI, GPT2, GPX1, GRHPR, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRN, GSN, GSR, GSS, GTPBP2, GTPBP3, GUF1, GUSB, GYG1, GYS1, HACE1, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, , HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HIKESHI, HINT1, HIVEP2, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNMT, HNRNPA1, HNRNPA2B1, HNRNPDL, HNRNPH2, HNRNPR, HNRNPU, HOGA1, HOXA1, HOXD10, HPCA, HPD, HPRT1, HPS1, HPS4, HPS5, HPS6, HRAS, HSD11B1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, HTRA1, HTRA2, HUWE1

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Neurología

HYAL1, HYDIN, IARS2, IBA57, IDH2, IDH3B, IDS, IDUA, IER3IP1, IFIH1, IFT140, IFT172, IFT27, IGBP1, IGF1, IGF1R, IGHMBP2, IL1RAPL1, IMPA1, INF2, INPP5E, INVS, IQSEC2, IRF2BPL, IRX5, ISCA1, ISCA2, ISCU, ITGA7, ITGB3, ITM2B, ITPA, ITPR1, IVD, JAG1, JAM2, JAM3, KANK1, KANSL1, KARS1, KAT6A, KAT6B, KAT8, KATNB1, KBTBD13, KCNA1, KCNA2, KCNB1, KCNC1, KCNC3, KCND3, KCNE3, KCNH1, KCNJ1, KCNJ10, KCNJ2, KCNK18, KCNK4, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD17, KCTD3, KCTD7, KDM4B, KDM5B, KDM5C, KDM6A, KIAA1109, KIDINS220, KIF11, KIF14, KIF1A, KIF1B, KIF1C, KIF21A, KIF2A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KLHL40, KLHL41, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT5, KRT8, KY, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMB2, LAMC3, LAMP2, LARGE1, LARS2, LAT, LBR, LDB3, LDHA, LEP, LGI1, LGI4, LHX3, LHX4, LIAS, LIMS2, LINS1, LIPA, LIPT1, LIPT2, LITAF, LMAN2L, LMBRD1, LMNA, LMNB1, LMOD3, LMX1B, LONP1, LPIN1, LRBA, LRP1, LRP10, LRP2, LRP4, LRPPRC, LRRK2, LRSAM1, LYRM7, LYST, LZTFL1, LZTR1, MACF1, MAF, MAG, MAGEL2, MAGI2, MAGT1, MAMLD1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MAPT, MARS1, MARS2, MASP1, MATR3, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCEE, MCM4, MCM6, MCOLN1, MCPH1, MDH2, MECP2, MECR, MED12, MED13, MED13L, MED17, MED23, MED25, MEF2C, MEGF10, MEIS2, METTL23, MFF, MFN2, MFRP, MFSD2A, MFSD8, MGAT2, MGME1, MIB1, MICU1, MID1, MIPEP, MITF, MKKS, MKS1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPDZ, MPI, MPV17, MPZ, MRE11, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MSMO1, MSR1, MSRB3, MSTO1, MSX1, MSX2, MTFMT, MTHFD1, MTHFR, MTHFS, MTM1, MTMR14, MTMR2, MTO1, MTOR, MTPAP, MTR, MTRFR, MTRR, MTTP, MUSK, MYBPC1, MYBPC3, MYCN, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO1E, MYO5A, MYO9A, MYO9B, MYORG, MYOT, MYPN, MYT1L, NAA10, NAA15, NACC1, NADK2, NAGA, NAGLU, NAGS, NALCN, NARS2, NAXD, NAXE, NBAS, NBEA, NCAPD3, NDE1, NDP, NDRG1, NDST1, 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, NEB, NECAP1, NECTIN1, NEDD4L, NEFH, NEK1, NEK10, NEU1, NEUROD2, NEXMIF, NF1, NFE2L2, NFIA, NFIB, NFIX, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHS, NIPA1, NIPBL, NKX6-2, NLGN3, NLGN4X, NLRP12, NLRP3, NNT, NOTCH1, NOTCH2, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPR2, NPRL2, NPRL3, NR2F1, NR3C2, NRXN1, NSD1, NSD2, NSDHL, NSMCE3, NSUN2, NT5C2, NTHL1, NTRK1, NTRK2, NUBPL, NUP133, NUP62, NUS1, OAT, OCLN, OCRL, ODAD4, OFD1, OGDH, OPA1, OPA3, OPHN1, OPTN, ORC1, OSGEP, OTC, OTUD6B, OXCT1, P4HA2, P4HB, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PAM16, PANK2, PARK7, PARS2, PAX3, PAX6, PAX7, PBX1, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH15, PCDH19, PCK2, PCNT, PCYT2, PDCD1, PDCD10, PDE10A, PDE6D, PDE8B, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PDYN, PER2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGAP2, PGK1, PGM1, PHACTR1, PHF6, PHF8, PHGDH, PHIP, PHKA1, PHOX2B, PHYH, PIEZO2, PIGA, PIGB, PIGC, PIGG, PIGH, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PIK3R5, PINK1, PIP5K1C, PITX1, PITX2, PKLR, PLA2G6, PLAA, PLCB1, PLCG2, PLEC, PLEKHG2, PLEKHG5, PLK1, PLK4, PLN, PLOD2, PLP1, PLPBP, PLXNB3, PMM2, PMP22, PMPCA, PMPCB, PNKD, PNKP, PNPLA2, PNPLA6, PNPLA8, PNPO, PNPT1, POGLUT1, POGZ, POLA1, POLG, POLG2, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PON1, POP1, PORCN, POT1, POU1F1, PPM1D, PPOX, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PREPL, PRF1, PRICKLE1, PRICKLE2, PRKAG2, PRKCA, PRKCG, PRKN, PRKRA, PRMT7, PRNP, PRODH, PROP1, PRPH, PRPS1, PRRT2, PRRX1, PRSS12, PRX, PSAP, PSAT1, PSEN1, PSEN2, PSMD12, PSPH, PTCH1, PTCHD1, PTEN, PTF1A, PTPN11, PTPN23, PTPRC, PTRH2, PTS, PUM1, PURA, PUS1, PUS3, PXDN, PYCR1, PYCR2, PYGM, PYROXD1, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB18, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAB7A, RAC1, RAD21, RAD50, RAF1, RAI1, RALA, RALGAPA1, RAP1GDS1, RAPSN, RARS1, RARS2, RBBP8, RBCK1, RBFOX1, RBM10, RBM8A, RDH11, REEP1, REEP2, RELN, RERE, REST, RET, RETREG1, RFT1, RHOBTB2, RIMS1, RIN2, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, RNF170, RNF216, ROBO2, ROGDI, ROR2, RORA, RORB, RPGRIP1L, RPIA, RPL10, RPL35A, RPS14, RPS6KA3, RRM2B, RTN2, RTN4IP1, RTTN, RUBCN, RUSC2, RXYLT1, RYR1, SACS, SALL1, SAMD9L, SAMHD1, SARS2, SASH1, SASS6, SATB2, SBDS, SBF1, SBF2, SC5D, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCO1, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHD, SEC23B, SECISBP2, SELENOI, SELENON, SEMA5A, SEMA6B, SEPSECS, SERAC1, SERPINI1, SET, SETBP1, SETD1A, SETD2, SETD5, SETX, SF3B1, SFXN4, SGCA, SGCB, SGCD, SGCE, SGCG, SGSH, SH3TC2, SHANK2, SHH, SHOC2, SHROOM4, SIGMAR1, SIK1, SIL1, SIN3A, SIX3, SKI, SLC12A3, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A3, SLC19A2, SLC19A3, SLC1A1, SLC1A2, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC27A4, SLC2A1, SLC2A10, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC39A14, SLC39A8, SLC3A1, SLC4A10, SLC4A4, SLC52A2, SLC52A3, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A4, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A6, SLC9A9, SLCO1B3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMCHD1, SMPD1, SMPD4, SMS, SNAI2, SNAP25, SNAP29, SNCA, SNCB, SNIP1,

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Neurología

SNTA1, SNX14, SNX27, SOBP, SOD1, SOD2, SON, SORL1, SOS1, SOX10, SOX11, SOX2, SOX3, SOX5, SPART, SPAST, SPATA5, SPEG, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRCAP, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAC3, STAG1, STAMBP, STAR, STAT1, STAT2, STIL, STIM1, STRA6, STRADA, STT3A, STUB1, STX1B, STXBP1, SUCLA2, SUCLG1, SUGCT, SUMF1, SUN2, SUOX, SURF1, SYN1, SYN2, SYNE1, SYNE2, SYNGAP1, SYNJ1, SYP, SYT2, SZT2, TACO1, TAF1, TAF13, TAF15, TAF2, TAF6, TAFAZZIN, TANGO2, TAOK1, TARDBP, TARS2, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBK1, TBL1XR1, TBR1, TBX1, TBX3, TCAP, TCF20, TCF4, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TECR, TECTA, TENM4, TET2, TFAP2A, TFAP2B, TFG, TFR2, TG, TGFB1, TGFB3, TGIF1, TGM6, TH, THAP1, THRA, THRB, TIA1, TIMM50, TIMM8A, TIMMDC1, TINF2, TK2, TLK2, TMCO1, TMEM106B, TMEM126A, TMEM126B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM230, TMEM231, TMEM237, TMEM240, TMEM43, TMEM67, TMEM70, TMLHE, TMTC3, TMX2, TNIK, TNK2, TNNI2, TNNT1, TNNT3, TNPO3, TOE1, TOP3A, TOR1A, TOR1AIP1, TPI1, TPK1, TPM2, TPM3, TPO, TPP1, TRAF3IP1, TRAF7, TRAK1, TRAPPC11, TRAPPC4, TRAPPC9, TREM2, TREX1, TRIM2, TRIM32, TRIM8, TRIO, TRIP12, TRIP4, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPC6, TRPM1, TRPM6, TRPS1, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TTBK2, TTC19, TTC21B, TTC8, TTI2, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWNK, TYMP, TYR, TYROBP, UBA1, UBA5, UBAP1, UBE2A, UBE3A, UBE3B, UBQLN2, UBR1, UBTF, UCHL1, UFM1, UGP2, UMPS, UNC80, UNG, UPB1, UPF3B, UQCC2, UQCRB, UQCRC2, UQCRQ, UROC1, USH2A, USP8, USP9X, USP9Y, VAMP1, VAMP2, VANGL1, VAPB, VARS1, VARS2, VCP, VDR, VHL, VIPAS39, VLDLR, VMA21, VPS11, VPS13A, VPS13B, VPS13C, VPS13D, VPS33B, VPS35, VPS37A, VPS53, VRK1, WAC, WARS2, WASF1, WASHC4, WASHC5, WDFY3, WDR26, WDR37, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WNK1, WNT1, WNT5A, WNT7A, WWOX, XK, XPNPEP3, XPR1, YAP1, YARS1, YARS2, YWHAE, YWHAG, YY1, ZBTB16, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFYVE26, ZFYVE27, ZIC1, ZIC2, ZIC3, ZMYND11, ZNF142, ZNF292, ZNF335, ZNF41, ZNF423, ZNF699, ZNF711, ZNF81

Panel de epilepsia

Si bien algunos tipos de convulsiones se clasifican fácilmente (es decir, parciales o generalizadas), otros no lo son o pueden convertirse más tarde en diferentes tipos (es decir, convulsiones parciales con generalización secundaria), lo que hace que las pruebas de panel específicas tengan menos probabilidades de lograr un diagnóstico. Nuestro panel de epilepsia es un panel dirigido por fenotipo que cubre diferentes tipos de síndromes convulsivos, que abarcan el síndrome de Dravet, la encefalopatía epiléptica infantil temprana, la epilepsia parcial, la epilepsia generalizada, la ausencia de epilepsia, el panel de epilepsia mioclónica y la hipomagnesemia. Este panel no incluye genes mitocondriales (es decir, genes que causan epilepsia mioclónica con fibras rojas irregulares -MERRF-). Si la sospecha clínica está orientada a trastornos metabólicos o mitocondriales, solicite CentoMito Comprehensive.

Incluye 784 genes: AARS1, AARS2, ABAT, ABCC8, ABCD1, ABCD3, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADGRG1, ADSL, AFF3, AFG3L2, AGA, AGK, AGPS, AIFM1, AIMP1, AIMP2, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMPD2, AMT, ANK3, ANTXR2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APP, APTX, ARG1, ARHGAP31, ARHGEF9, ARSA, ARSB, ARV1, ARX, ASAH1, ASL, ASNS, ASPA, ASS1, ASXL1, ATM, ATP13A2, ATP1A2, ATP6AP1, ATP6V0A2, ATP6V1A, ATP7A, ATP7B, ATPAF2, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BEST1, BOLA3, BRAT1, BTD, C12orf57, C19orf12, CA5A, CACNA1A, CACNA1E, CACNA1H, CACNA2D2, CACNB4, CARS2, CASK, CAV1, CBS, CCDC115, CCDC88A, CDKL5, CERS1, CHD2, CHMP2B, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CIC, CLCN2, CLCN4, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CLTC, CNNM2, CNPY3, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPA6, CPLX1, CPS1, CPT1A, CPT2, CSF1R, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCX, DEAF1, DEGS1, DENND5A, DEPDC5, DGUOK, DHCR7, DHDDS, DHFR, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL3, DNAJC5, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYM, DYRK1A, EARS2, ECHS1, EEF1A2, EFHC1, EGF, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EMC10, EML1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8,

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

ETFA, ETFB, ETFDH, ETHE1, F2, F5, FA2H, FAH, FAM126A, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGF12, FH, FHL1, FIG4, FKRP, FLVCR2, FOLR1, FOXG1, FOXRED1, FRRS1L, FTL, FUCA1, FUT8, FXYD2, GAA, GABBR2, GABRA1, GABRA2, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALT, GAMT, GAN, GBA, GBE1, GCDH, GCH1, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLUD1, GLUL, GM2A, GMPPA, GNAO1, GNAQ, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GRIA2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GTPBP2, GTPBP3, GUF1, GUSB, HACE1, HADHA, HADHB, HCFC1, HCN1, HEPACAM, HEXA, HEXB, HGSNAT, HIBCH, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPR, HNRNPU, HRAS, HSD17B4, HSPD1, HTRA1, IARS2, IBA57, IDS, IDUA, IER3IP1, IFIH1, IQSEC2, ISCA2, ITPA, IVD, JAG1, JAM3, KAT8, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD3, KCTD7, KIAA1109, KIF2A, KIF5A, KMT2E, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LAT, LDB3, LGI1, LIAS, LIPA, LIPT1, LMNB1, LRPPRC, LYRM7, LYST, MAF, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MBD5, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MECR, MED17, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, 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, MTHFS, MTOR, MTR, MTRFR, MYO5A, MYOT, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDE1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFE2L2, NFU1, NGLY1, NHLRC1, NOTCH1, NOTCH3, NPC1, NPC2, NPR2, NPRL2, NPRL3, NRXN1, NSD2, NUBPL, NUS1, OAT, OCLN, OCRL, OPA1, OPA3, OSGEP, OTC, OTUD6B, P4HTM, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PHACTR1, PHGDH, PHYH, PIGA, PIGB, PIGH, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PLA2G6, PLAA, PLCB1, PLCG2, PLEKHG2, PLK1, PLP1, PMM2, PMP22, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMK, POMT1, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRF1, PRICKLE1, PRICKLE2, PRMT7, PRODH, PRPS1, PRRT2, PSAP, PSEN1, PTEN, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB27A, RAI1, RALA, RALGAPA1, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF216, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETD1A, SGCE, SGSH, SIK1, SLC12A3, SLC12A5, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SLC6A1, SLC6A8, SLC7A7, SLC9A6, SMC1A, SMPD1, SNIP1, SNTA1, SNX27, SON, SOX10, SPART, SPATA5, SPG11, SPG7, SPTAN1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAMBP, STAT1, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TBC1D24, TBCD, TBCE, TBCK, TCF4, TDP2, TGFB1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM199, TMEM70, TMX2, TNK2, TPK1, TPP1, TRAK1, TRAPPC4, TRAPPC9, TREM2, TREX1, TRIM8, TRPM6, TRPV4, TSC1, TSC2, TSEN54, TSFM, TTC19, TUBA1A, TUBB2A, TUBB2B, TUBB4A, TUBG1, TUFM, TUSC3, TWNK, TYMP, TYROBP, UBE2A, UBE3A, UFM1, UGP2, UMPS, UPB1, UQCRQ, VAMP2, VARS1, VARS2, VCP, VPS11, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR73, WFS1, WWOX, YWHAG, ZEB2, ZFYVE26, ZNF142, ZNF335

Panel de discapacidad intelectual

Incluye genes asociados con discapacidades intelectuales que cubren todos los mecanismos de herencia, así como autismo sindrómico y no sindrómico, microcefalia, trastornos de migración neuronal, regresión del desarrollo y Aicardi Goutierres. La detección del síndrome X frágil es posible ya que nuestro panel incluye la expansión repetida de FMR1.

Incluye 817 genes: ABAT, ABCA7, ABCD1, ACE, ACHE, ACSL4, ACTB, ACTG1, ACTN4, ADA, ADAR, ADAT3, ADCY5, ADGRG1, ADK, ADNP, ADSL, AFF2, AHDC1, AHI1, AIMP1, AKT3, ALDH18A1, ALDH5A1, ALDH7A1, ALG11, ALG13, ALX4, AMMECR1, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKLE2, ANKRD11, AP1S1, AP1S2, AP2M1, AP3B1, AP4B1, AP4M1, ARFGEF2, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARX, ASAH1, ASCL1, ASH1L, ASPM, ASTN2, ASXL1, ASXL3, ATP13A2, ATP1A1, ATP1A3, ATP6AP2, ATP6V0A2, ATP7A, ATP8A2, ATR, ATRX, AUTS2,

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

B3GALNT2, B4GAT1, B9D1, B9D2, BBS4, BCAP31, BCKDK, BCL11A, BCOR, BCS1L, BDNF, BLOC1S1, BLOC1S3, BLOC1S6, BRAF, BRCA2, BRWD3, C12orf4, C12orf57, C19orf12, CA2, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1H, CACNB2, CAMK2A, CAMK2B, CAMK2G, CAMTA1, CARD11, CASK, CBS, CC2D1A, CC2D2A, CCDC22, CCDC40, CCDC88C, CDH11, CDH15, CDK5RAP2, CDKL5, CDON, CENPF, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CERT1, CHAMP1, CHD1, CHD2, CHD3, CHD7, CHD8, CHL1, CHMP1A, CIB2, CIC, CIT, CLCN4, CLN8, CLP1, CLTC, CNGB3, CNKSR2, CNTN4, CNTNAP2, COASY, COL4A1, CPT2, CRADD, CRBN, CREBBP, CRIPT, CRPPA, CSMD1, CSPP1, CTC1, CTCF, CTNNA3, CTNNB1, CUL3, CUL4B, CUL7, CUX1, CUX2, CX3CR1, CYB5R3, CYP11B1, CYP27A1, DARS1, DARS2, DCAF17, DCX, DDC, DDX3X, DEAF1, DHCR7, DIP2B, DKC1, DLG3, DLG4, DLGAP2, DLX3, DMXL2, DNM1, DNMT3A, DOCK8, DPF2, DPP6, DPYD, DRD3, DST, DTNBP1, DVL3, DYM, DYNC1H1, DYRK1A, EBF3, EDC3, EDN3, EDNRB, EEF1A2, EFTUD2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELOVL4, ELP2, EMC10, EMX2, EP300, EPB41L1, ETFB, EXOC6B, EXOSC3, EXOSC9, EXT1, EZH2, FAM126A, FAN1, FANCB, FBN1, FBXO11, FGA, FGD1, FGFR2, FGFR3, FH, FKRP, FKTN, FLNA, FLVCR1, FMN2, FMR1, FOLR1, FOXC1, FOXG1, FOXL2, FOXP1, FOXP2, FRMPD4, FTL, FTO, FTSJ1, G6PD, GABBR2, GABRA5, GABRB3, GAD1, GAMT, GATAD2B, GATM, GCK, GDI1, GDNF, GFAP, GIGYF2, GJC2, GK, GLI2, GLRB, GMPPA, GMPPB, GNAO1, GNAS, GNB1, GPC3, GPC4, GPC6, GPHN, GPT2, GPX1, GRIA1, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC2, HIVEP2, HNMT, HNRNPH2, HNRNPU, HOXA1, HPRT1, HPS1, HPS4, HPS5, HPS6, HRAS, HSD11B1, HSD17B10, HSPD1, HUWE1, HYDIN, IDS, IER3IP1, IFIH1, IGBP1, IGF1, IGF1R, IL1RAPL1, IMPA1, INPP5E, INVS, IQSEC2, IRF2BPL, IRX5, ITGA7, ITGB3, ITPR1, JAG1, KANK1, KANSL1, KAT6A, KAT6B, KATNB1, KCNB1, KCNC1, KCNC3, KCND3, KCNJ10, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KDM4B, KDM5B, KDM5C, KDM6A, KIF11, KIF14, KIF1A, KIF5C, KIF7, KIFBP, KIRREL3, KMT2A, KMT2C, KMT2D, KMT5B, KNL1, KPTN, KRAS, L1CAM, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LEP, LINS1, LMAN2L, LMX1B, LRBA, LRP1, LRP2, LZTR1, MACF1, MAGEL2, MAGT1, MAN1B1, MAOA, MBD5, MBOAT7, MBTPS2, MCCC2, MCM4, MCM6, MCPH1, MECP2, MED12, MED13, MED13L, MED17, MED23, MEF2C, MEGF10, MEIS2, METTL23, MFRP, MFSD2A, MGAT2, MIB1, MID1, MITF, MKKS, MKS1, MLC1, MPDZ, MSMO1, MSR1, 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, MTHFR, MTM1, MTOR, MTR, MYCN, MYO1E, MYO5A, MYO9B, MYT1L, NAA10, NAA15, NACC1, NAGA, NALCN, NBEA, NCAPD3, NDE1, NDP, NDST1, NDUFA1, NECTIN1, NEK10, NEXMIF, NF1, NFIA, NFIB, NFIX, NHEJ1, NHS, NIPA1, NIPBL, NLGN3, NLGN4X, NOTCH2, NPHP1, NPHP3, NR2F1, NR3C2, NRXN1, NSD1, NSDHL, NSMCE3, NSUN2, NTRK1, NUP133, NUS1, OCLN, OCRL, ODAD4, OFD1, OPHN1, ORC1, OTC, P4HA2, PACS1, PAFAH1B1, PAH, PAK3, PANK2, PAX3, PAX6, PBX1, PCDH15, PCDH19, PCNT, PDCD1, PDE6D, PDHA1, PER2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGK1, PHF6, PHF8, PHIP, PIGA, PIGC, PIGG, PIGL, PIGN, PIGO, PIGV, PIK3R2, PITX1, PITX2, PLA2G6, PLCB1, PLK1, PLK4, PLN, PLP1, PNKP, POGZ, POLA1, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PON1, PORCN, POT1, PPM1D, PPOX, PPP2R1A, PPP2R5D, PPT1, PQBP1, PRICKLE1, PRKCA, PRKN, PRODH, PRPS1, PRSS12, PSMD12, PTCH1, PTCHD1, PTEN, PTPN11, PTPRC, PTS, PURA, PUS3, PXDN, PYCR1, PYCR2, QARS1, QRICH1, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAI1, RAP1GDS1, RARS2, RBBP8, RBFOX1, RBM10, RELN, RERE, REST, RET, RIMS1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, ROBO2, ROGDI, ROR2, RPGRIP1L, RPL10, RPS6KA3, RTTN, RUSC2, RXYLT1, SALL1, SAMHD1, SASH1, SASS6, SATB2, SBF1, SC5D, SCN1A, SCN2A, SCN4A, SCN8A, SCN9A, SCO2, SDCCAG8, SDHA, SEPSECS, SET, SETBP1, SETD2, SETD5, SF3B1, SGCA, SHANK2, SHH, SHROOM4, SIL1, SIN3A, SIX3, SLC12A5, SLC16A2, SLC1A1, SLC25A12, SLC25A15, SLC25A19, SLC27A4, SLC2A1, SLC35A2, SLC35A3, SLC4A10, SLC4A4, SLC6A1, SLC6A17, SLC6A3, SLC6A4, SLC6A8, SLC7A7, SLC9A6, SLC9A9, SLCO1B3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMPD1, SMS, SNAI2, SNAP25, SNAP29, SNIP1, SOBP, SOD1, SOX10, SOX11, SOX2, SOX3, SOX5, SPAST, SRCAP, SRD5A3, ST3GAL3, STAG1, STAMBP, STIL, STRA6, STXBP1, SYN1, SYN2, SYNE1, SYNGAP1, SYNJ1, SYP, TAF13, TAF2, TAF6, TAOK1, TBC1D20, TBC1D23, TBC1D24, TBCE, TBL1XR1, TBR1, TBX1, TBX3, TCF20, TCF4, TCOF1, TCTN1, TCTN2, TCTN3, TECR, TECTA, TET2, TFAP2A, TGIF1, THRA, THRB, TIMM8A, TINF2, TLK2, TMCO1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMLHE, TMTC3, TNIK, TOE1, TPO, TRAF7, TRAPPC9, TREX1, TRIO, TRIP12, TRMT10A, TRPC6, TRPM1, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TTC21B, TTI2, TTN, TUBA1A, TUBA8, TUBB2B, TUBB3, TUBB4A, TUBGCP4, TUBGCP6, TUSC3, TYR, UBE2A, UBE3A, UBE3B, UBR1, UNC80, UPF3B, UROC1, USH2A, USP9X, USP9Y, VDR, VLDLR, VPS13B, VPS53, VRK1, WAC, WASHC4, WDFY3, WDR26, WDR45, WDR62, WDR81, WNT1, WNT5A, WWOX, YWHAE, YY1, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMYND11, ZNF292, ZNF335, ZNF41, ZNF423, ZNF699, ZNF711, ZNF81, MECR.

Genes de expansión de repetición: FMR1

Gen med

Laboratorio de diagnóstico clínico y genómico

Neurología

Panel neuromuscular

Es ideal para pacientes con enfermedades musculares. Incluye genes que causan enfermedades neurológicas y cubre trastornos, como miopatías metabólicas, distrofias musculares, Charcot-Marie-Tooth, síndromes miasténicos congénitos, miopatías congénitas, miopatías miofibrilares, miopatías nemalínicas y otros síndromes con hipotonía, miotonía o debilidad. La artrogriposis se incluye para el diagnóstico diferencial de los trastornos neuromusculares de aparición temprana. Si existe una alta sospecha diagnóstica de distrofia muscular de Duchenne, recomendamos que el médico ordene un análisis de deleción/duplicación por MLPA dirigido al gen DMD como un servicio adicional.

Incluye 354 genes: AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTG2, ADGRG6, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, AMPD1, ANO5, ARHGEF9, ASAH1, ASCC1, ATAD1, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, BVES, CACNA1S, CAPN3, CASK, CASQ1, CAV1, CAV3, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTNAP1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COX6A1, CPT2, CRLF1, CRPPA, CRYAB, CSRP3, CTDP1, DAG1, DCTN1, DES, DGUOK, DHCR24, DHTKD1, DMD, DMPK, DNA2, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBLN5, FBN2, FBXO38, FDX2, FGD4, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNC, GAA, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GJB1, GLDN, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GPHN, GYG1, GYS1, HADHA, HADHB, HINT1, HK1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, ISCU, ITGA7, KARS1, KAT6B, KBTBD13, KCNA1, KCNE3, KCNJ2, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL7, KY, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LGI4, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MAGEL2, MAMLD1, MARS1, MATR3, MED25, MEGF10, MFN2, MICU1, MPV17, MPZ, 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, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NALCN, NDRG1, NEB, NTRK1, OPA1, OPA3, PAX7, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PRKAG2, PRPS1, PRX, PYGM, PYROXD1, QARS1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SLC6A5, SMCHD1, SMN1, SMN2, SMPD4, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, STAC3, STIM1, SUCLA2, SUN2, SYNE1, SYNE2, SYT2, TAFAZZIN, TANGO2, TBCK, TCAP, TFG, TGFB3, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIP4, TRPV4, TSEN2, TSFM, TTN, TWNK, TYMP, UBA1, VAMP1, VAPB, VCP, VIPAS39, VMA21, VPS33B, VRK1, WNK1, XK, YARS1, ZC4H2.