GenoMed

Catálogo de pruebas genómicas por especialidad

Gen med

Laboratorio de diagnóstico clínico y genómico

Cardiología

CentoCardio

Incluye los genes más relevantes para arritmias, cardiopatías congénitas y miocardiopatías. Los síndromes incluyeron: QT largo y corto, síndrome de Brugada, taquicardia ventricular polimórfica catecolaminérgica, miocardiopatías dilatadas e hipertróficas y defectos cardíacos congénitos. Además, este panel incluye anomalías vasculares, como dolicoectasia y telangiectasia hemorrágica hereditaria. El panel no incluye análisis de PKD1.

ABCC9, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, AKAP9, ANK2, ANKRD1, ARHGAP31, ATM, B3GAT3, BAG3, BCOR, BMPR2, BRAF, CACNA1C, CACNB2, CALM1, CALM2, CASQ2, CAV3, CAVIN4, CBL, CDH2, CFAP53, CFC1, CHD7, CITED2, CLDN16, CLDN19, CNNM2, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, CREBBP, CRELD1, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EFEMP2, EGF, EHMT1, ELN, EMD, ENG, EP300, EVC, EVC2, EYA4, FBN1, FBN2, FHL1, FKTN, FLNA, FLNC, FOXC1, FOXF1, FOXH1, FXYD2, GAA, GATA4, GATA5, GATA6, GDF1, GDF2, GJA1, GJA5, GLA, GPC3, GPD1L, HCCS, HCN4, HFE, HRAS, HTRA1, ILK, JAG1, JPH2, JUP, KCNA1, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNK3, KCNQ1, KDM6A, KMT2D, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K2, MED12, MED13L, MEIS2, MFAP5, MIB1, MMP21, MMP3, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6,MYOZ2, MYPN, NEBL, NEXN, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NOTCH3, NPPA, NR2F2, NRAS, NSD1, PDLIM3, PKD1L1, PKD2, PKP2, PLN, PRDM16, PRKAG2, PRKG1, PSEN1, PSEN2, PTPN11, RAF1, RASA1, RBM10, RBM20, RIT1, RYR2, SALL1, SALL4, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, SKI, SLC12A3, SLC22A5, SLC25A4, SLC2A10, SLMAP, SMAD3, SMAD4, SMAD6, SMC3, SNTA1, SOS1, SOS2, SOX2, STRA6, SYNE1, SYNE2, TAB2, TAZ, TBX1, TBX20, TBX5, TCAP, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TREX1, TRIM63, TRPM4, TRPM6, TTN, TTR, VCL, ZEB2, ZFPM2, ZIC3SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, ESQUÍ, SLC12A3, SLC22A5, SLC25A4, SLC2A10, SLMAP, SMAD3, SMAD4, SMAD6, SMC3, SNTA1, SOS1, SOS2, SOX2, STRA6, SYNE1, SYNE2, TAB2, TAZ, TBX1, TBX20, TBX5, TCAP, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TREX1, TRIM63, TRPM4, TRPM6, TTN, TTR, VCL, ZEB2, ZFPM2, ZIC3SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, ESQUÍ, SLC12A3, SLC22A5, SLC25A4, SLC2A10, SLMAP, SMAD3, SMAD4, SMAD6, SMC3, SNTA1, SOS1, SOS2, SOX2, STRA6, SYNE1, SYNE2, TAB2, TAZ, TBX1, TBX20, TBX5, TCAP, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TREX1, TRIM63, TRPM4, TRPM6, TTN, TTR, VCL, ZEB2, ZFPM2, ZIC3

Panel Pulmonar

Incluye genes para el diagnóstico de hipoventilación central, disfunción del metabolismo del surfactante, hipertensión pulmonar, entre otras enfermedades pulmonares.

ABCA3, ABCC8, ACVRL1, AP3B1, ASCL1, BLOC1S3, BLOC1S6, BMPR1B, BMPR2, CAV1, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, ECE1, EDN3, EFEMP2, EIF2AK4, ELN, ENG, FBLN5, FBN1, FLCN, FOXF1, GDF2, GDNF, GLRA1, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA3, KCNA5, KCNK3, MECP2, NF1, NFU1, NKX2-1, NME8, NOP10, NOTCH3, PARN, PHOX2A, PHOX2B, POLD1, RAPSN, RASA1, RET, RSPH1, RSPH4A, RSPH9, RTEL1, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPA2, SFTPB, SFTPC, SLC6A5, SLC7A7, SMAD4, SMAD9, SMPD1, STAT3, STRA6, TERT, TINF2, TSC1, TSC2, ZEB2

Genes de expansión : PHOX2B

Gen med

Laboratorio de diagnóstico clínico y genómico

Cardiología

Estudios de un solo gen

Se recomienda la prueba de un solo gen en pacientes que tienen:

- Características clínicas distintivas
- Antecedentes familiares de un trastorno específico.
- Trastornos de un solo gen
- Posible trastorno epigenético
- Posibles trastornos de repetición triple
- Confirmación de prueba familiar

Displasia alveolocapilar con desalineación de las venas pulmonares FOXF1
Miocardiopatía arritmogénica del ventrículo derecho tipo 1 TGFB3
Miocardiopatía arritmogénica del ventrículo derecho tipo 10 DSG2
Miocardiopatía arritmogénica del ventrículo derecho tipo 11 DSC2
Miocardiopatía arritmogénica del ventrículo derecho tipo 12 JUP
Miocardiopatía arritmogénica del ventrículo derecho tipo 5 TMEM43
Miocardiopatía arritmogénica del ventrículo derecho tipo 8 DSP
Miocardiopatía arritmogénica del ventrículo derecho tipo 9 PKP2
Displasia arritmogénica del ventrículo derecho tipo 2 RYR2
Fibrilación auricular tipo 10 SCN5A
Fibrilación auricular tipo 11 GJA5
Fibrilación auricular tipo 12 ABCC9
Fibrilación auricular tipo 3 KCNQ1
Fibrilación auricular tipo 4 KCNE2
Fibrilación auricular tipo 6 NPPA
Fibrilación auricular tipo 7 KCNA5
Comunicación interauricular tipo 3 MYH6
Comunicación interauricular tipo 4 TBX20
Comunicación interauricular tipo 5 ACTC1
Comunicación interauricular tipo 6 TLL1
Comunicación interauricular tipo 8 CITED2
Comunicación interauricular tipo 9 GATA6
Comunicación auriculoventricular tipo 4 GATA4
Comunicación auriculoventricular tipo 5 GATA6
síndrome de Barth TAZ
Válvula aórtica bicúspide TIMP1
Síndrome de Brugada tipo 1 SCN5A
Síndrome de Brugada tipo 2 GPD1L
Síndrome de Brugada tipo 3 CACNA1C
Síndrome de Brugada tipo 4 CACNB2
Síndrome de Brugada tipo 5 SCN1B
Síndrome de Brugada tipo 6 KCNE3
Síndrome de Brugada tipo 7 SCN3B
Síndrome de Brugada tipo 8 HCN4
Síndrome de Brugada tipo 9 SLMAP
Defectos cardíacos, relacionados con CNOT3
Defectos cardíacos, relacionados con PPP1R8
Displesia valvular cardíaca, ligada al cromosoma X FLNA
Cardioencefalomiopatía infantil fatal por deficiencia de citocromo c oxidasa SCO2
Cardioencefalomiopatía infantil fatal por deficiencia de citocromo c oxidasa tipo 2 COX15
Cardioencefalomiopatía infantil fatal por deficiencia de citocromo c oxidasa tipo 3 COA5
Síndrome cardiofaciocutáneo BRAF
Síndrome cardiofaciocutáneo KRAS
Síndrome cardiofaciocutáneo tipo 3 MAP2K1
Síndrome cardiofaciocutáneo tipo 4 MAP2K2
Miocardiopatía hipertrófica apical y neuropatía relacionada con MT-ATP8
Miocardiopatía, dilatada MYBPC3
Miocardiopatía dilatada tipo 1 CRYAB
Miocardiopatía dilatada tipo 1A LMNA
Miocardiopatía dilatada tipo 1AA ACTN2
Miocardiopatía dilatada tipo 1BB DSG2
Miocardiopatía dilatada tipo 1C LDB3
Miocardiopatía dilatada tipo 1CC NEXN
Miocardiopatía dilatada tipo 1D TNNT2
Miocardiopatía dilatada tipo 1DD RBM20
Miocardiopatía dilatada tipo 1E SCN5A
Miocardiopatía dilatada tipo 1EE MYH6
Miocardiopatía dilatada tipo 1G TTN
Miocardiopatía dilatada tipo 1GG SDHA
Miocardiopatía dilatada tipo 1HH BOLSA3
Miocardiopatía dilatada tipo 1I DES
Miocardiopatía dilatada tipo 1J EYA4
Miocardiopatía dilatada tipo 1KK MYPN
Miocardiopatía dilatada tipo 1L SGCD
Miocardiopatía dilatada tipo 1LL PRDM16
Miocardiopatía dilatada tipo 1M CSRP3
Miocardiopatía dilatada tipo 1N TCAP
Miocardiopatía dilatada tipo 1O ABCC9
Miocardiopatía dilatada tipo 1P PLN
Miocardiopatía dilatada tipo 1R ACTC1
Miocardiopatía dilatada tipo 1S MYH7
Miocardiopatía dilatada tipo 1T TMPO
Miocardiopatía dilatada tipo 1U PSEN1
Miocardiopatía dilatada tipo 1V PSEN2
Miocardiopatía dilatada tipo 1W VCL
Miocardiopatía dilatada tipo 1X FKTN
Miocardiopatía dilatada tipo 1Y TPM1
Miocardiopatía dilatada tipo 1Z TNNC1
Miocardiopatía dilatada tipo 2A TNNI3
Miocardiopatía dilatada tipo 2B GATAD1
Miocardiopatía dilatada tipo 3B DMD
Miocardiopatía dilatada con ataxia ADNJC19
Miocardiopatía dilatada con hipogonadismo hipergonadotrópico LMNA
Miocardiopatía dilatada con pelo lanoso y queratodermia DSP
Miocardiopatía hipertrófica familiar CAV3
Miocardiopatía hipertrófica familiar tipo 1 MYH7
Miocardiopatía hipertrófica familiar tipo 10 MYL2
Miocardiopatía hipertrófica familiar tipo 11 ACTC1
Miocardiopatía hipertrófica familiar tipo 12 CSRP3
Miocardiopatía hipertrófica familiar tipo 16 MYOZ2

Gen med

Laboratorio de diagnóstico clínico y genómico

Cardiología

Miocardiopatía hipertrófica familiar tipo 17 JPH2
Miocardiopatía hipertrófica familiar tipo 19 CALR3
Miocardiopatía hipertrófica familiar tipo 2 TNNT2
Miocardiopatía hipertrófica familiar tipo 3 TPM1
Miocardiopatía hipertrófica familiar tipo 4 MYBPC3
Miocardiopatía hipertrófica familiar tipo 6 PRKAG2
Miocardiopatía hipertrófica familiar tipo 7 TNNI3
Miocardiopatía hipertrófica familiar tipo 8 MYL3
Miocardiopatía hipertrófica familiar tipo 9 TTN
Miocardiopatía familiar restrictiva tipo 1 TNNI3
Miocardiopatía, fatal, relacionada con MT-TI MT-TI
Miocardiopatía hipertrófica medioventricular digénica MYLK2
Miocardiopatía hipertrófica relacionada con MT-TG
Miocardiopatía hipertrófica tipo 18 PLN
Miocardiopatía, dilatada idiopática, mitocondrial, relacionada con MT-TH L-J
Miocardiopatía hipertrófica infantil relacionada con MT-ATP8
Miocardiopatía, no compactación del ventrículo izquierdo, relacionada con MYH7B
Síndrome de hipoventilación central con o sin enfermedad de Hirschsprung PHOX2B
Síndrome de hipoventilación central, congénito ASCL1
Defectos cardíacos congénitos y displasia ectodérmica PRKD1
Múltiples tipos de defectos cardíacos congénitos GATA5
Múltiples tipos de defectos cardíacos congénitos TAB2
Enfermedad coronaria, susceptibilidad a, tipo 6 MMP3
Deficiencia de CR1
Dnfermedad de danon LAMP2
Hernia diafragmática tipo 3 ZFPM2
Miocardiopatía dilatada con pelo lanoso, queratodermia y agenesia dental DSP
Deficiencia de dopamina beta-hidroxilasa (DBH) DAP
Fibrilación auricular familiar tipo 13 SCN1B
Bloqueo cardíaco, progresivo, familiar, tipo 1A SCN5A
Síndrome corazón-mano, tipo esloveno LMNA
Síndrome de Jervell y Lange-Nielsen tipo 1 KCNQ1
Síndrome de Jervell y Lange-Nielsen tipo 2 KCNE1
No compactación del ventrículo izquierdo 1, con o sin cardiopatías congénitas DTNA
No compactación del ventrículo izquierdo 7 MIB1
Enfermedad del legionario, susceptibilidad a TLR5
Enfermedad del legionario, susceptibilidad a TLR5
Síndrome de QT largo tipo 1 KCNQ1
Síndrome de QT largo tipo 10 SCN4B
Síndrome de QT largo tipo 11 AKAP9
Síndrome de QT largo tipo 12 SNTA1
Síndrome de QT largo tipo 13 KCNJ5
Síndrome de QT largo tipo 15 CALM2
Síndrome de QT largo tipo 2 KCNH2
Síndrome de QT largo tipo 3 SCN5A
Síndrome de QT largo tipo 4 ANK2
Síndrome de QT largo tipo 5 KCNE1
Síndrome de QT largo tipo 6 KCNE2
Síndrome de QT largo tipo 8 CACNA1C
Síndrome de QT largo tipo 9 CAV3
síndrome de Marfan FBN1
síndrome MASS FBN1
Síndrome de McKusick-Kaufman MKKS
síndrome MELAS MT-TC
síndrome MELAS MT-TF
Síndrome MELAS, relacionado con MT-TL1
Síndrome de superposición MERRF/MELAS, relacionado con MT-TS1
Síndrome de superposición MERRF/MELAS, relacionado con MT-TS2
Miopatía mitocondrial y anemia sideroblástica tipo 1 PUS1
Miopatía mitocondrial, infantil, transitoria, relacionada con MT-TE
Miopatía mitocondrial, aislada MT-TD
Miopatía mitocondrial, relacionada con MT-TA
Miopatía mitocondrial, relacionada con MT-TM
Deficiencia de portador de fosfato mitocondrial SLC25A3
Enfermedad de moyamoya tipo 5 ACTA2
Síndrome de disfunción multisistémica del músculo liso ACTA2
Miopatía relacionada con MT-TQ MT-TQ
Agenesia pancreática y cardiopatías congénitas GATA6
Bloqueo cardíaco familiar progresivo TRPM4
Fibrosis pulmonar, idiopática SFTPA1
Hipertensión pulmonar del recién nacido CRHR1
Enfermedad venooclusiva pulmonar tipo 2 EIF2AK4
Síndrome de Senger AGK
Síndrome de QT corto tipo 1 KCNH2
Síndrome de QT corto tipo 2 KCNQ1
Síndrome de QT corto tipo 3 KCNJ2
Síndrome del seno enfermo tipo 1 SCN5A
Síndrome del seno enfermo tipo 3 MYH6
Disfunción del nódulo sinoauricular y sordera CACNA1D
Síndrome de muerte súbita del lactante, susceptibilidad a SCN5A
Síndrome de muerte súbita del lactante con disgenesia de los testículos TSPYL1
Anomalías testiculares con o sin cardiopatía congénita GATA4
Tetralogía de Fallot ALDH1A2
Tetralogía de Fallot GATA4
Tetralogía de Fallot GATA6
Tetralogía de Fallot ZFPM2
Disección de aneurisma de aorta torácica SMAD2
Transposición de las grandes arterias, dextro-loop 1 MED13L
Fibrilación ventricular paroxística familiar tipo 1 SCN5A
Defecto del tabique ventricular tipo 1 GATA4
Defecto del tabique ventricular tipo 2 CITED2
Taquicardia ventricular catecolaminérgica polimórfica tipo 1 RYR2
Taquicardia ventricular catecolaminérgica polimórfica tipo 2 CASQ2
Taquicardia ventricular catecolaminérgica polimórfica tipo 4 CALMA1
Taquicardia ventricular catecolaminérgica polimórfica tipo 5 TRDN
Síndrome de Wolff-Parkinson-White PRKAG2

Gen med

Laboratorio de diagnóstico clínico y genómico

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, 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, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, 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, CHM, CHRNA1, CHRNB1, 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, 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, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, 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, 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, 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, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, 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, 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,

Gen med

Laboratorio de diagnóstico clínico y genómico

Neonatología

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, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, 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

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

Panel de Infertilidad

Nuestro panel de infertilidad se recomienda para pacientes que intentan concebir durante un año o más, con problemas de fertilidad conocidos, que han experimentado más de un aborto espontáneo, con menstruación irregular o ausente, con bajo recuento, forma o movimiento de espermatozoides, o con ausencia de desarrollo. de rasgos sexuales secundarios. Nuestro panel incluye los genes más importantes relacionados con la infertilidad en hombres y mujeres. Conocer la causa exacta de la infertilidad permite tomar mejores decisiones de diagnóstico y posibilita un mejor asesoramiento para las parejas.

Incluye 270 genes: AKR1C4, AMH, AMHR2, ANOS1, AR, ARL13B, ARL6, ARX, ATP6V0A4, ATRX, AURKC, B3GLCT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BMP15, BMP4, BNC2, CATSPER1, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CDKN1C, CEP164, CEP290, CEP41, CFAP298, CFAP300, CFAP418, CFTR, CHD4, CHD7, CREBBP, CUL4B, CUL7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DAZ2, DHCR24, DHCR7, DHH, DMRT1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DUSP6, DYNC2H1, EFNB1, ENPP1, EPG5, ERAL1, ESCO2, EVC, EVC2, FAT4, FBXL4, FEZF1, FGF10, FGF17, FGF8, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FLRT3, FMR1, FOXJ1, FOXL2, FRAS1, FREM2, FSHB, FSHR, GAS8, GATA4, GLI3, GNRH1, GNRHR, GPC3, GRIP1, HBA1, HCCS, HESX1, HEXA, HFE, HNF1B, HOXA13, HS6ST1, HSD17B3, HSD17B4, HSD3B2, HUWE1, IFT172, IFT27, IL17RD, INPP5E, INSL3, IRF6, KHDC3L, KIF7, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LMNA, LRRC56, LZTFL1, MAMLD1, MAP3K1, MCM9, MED12, MID1, MKKS, MKS1, 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, MYO7A, MYRF, NEK1, NEK10, NME8, NPHP1, NPHP3, NR0B1, NR0B2, NR3C1, NR5A1, NSMF, ODAD4, OFD1, OPHN1, PANX1, PATL2, PAX6, PCNT, PCSK1, PDE4D, PEX1, PHF6, PITX2, PLCZ1, PNPLA6, POLR3B, POMC, POR, POU1F1, PPARG, PROK2, PROKR2, PROM1, PROP1, PRPH2, PSMC3IP, PTDSS1, PTPN11, RBBP8, RDH5, RHO, RIPK4, RLBP1, RNF216, ROR2, RPGRIP1L, RPL10, RSPO1, SALL1, SAMD9, SDCCAG8, SEMA3A, SETBP1, SGPL1, SOS1, SOX10, SOX2, SOX3, SOX9, SPAG1, SPECC1L, SPRY4, SRY, STAR, TAC3, TACR3, TBX15, TEX11, TEX15, TLE6, TMEM67, TMEM70, TOE1, TP63, TRAF3IP1, TRAIP, TRIM32, TSPYL1, TTC12, TTC21B, TTC8, TUBB8, TWIST2, UBR1, USP9Y, WDPCP, WDR11, WDR35, WEE2, WNT4, ZMYND10, ZP1, ZP2, ZP3

Genes de expansión : AR, FMR1

CentoScreen

Panel de detección integral de 330 trastornos autosómicos, recesivos y ligados al cromosoma X. Análisis adicionales para síndrome X frágil, atrofia muscular espinal e hiperplasia suprarrenal congénita (genes FMR1, SMN1 y CYP21A2 respectivamente). Brinda la oportunidad de tomar decisiones informadas y revisar la gama de opciones disponibles para guiar el embarazo y la planificación familiar. Incluye evaluación de panel completo con análisis de CNV de 34 genes para cada socio.

Existen 3 tipos

-Solamente 1 paciente (Padre o Madre)

-Pareja que quiere concebir (Padres)

-2 parejas (abuelos y padres) [asociación]

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

Prueba prenatal (invasiva y no invasiva)

Diseñado para analizar las aneuploidías cromosómicas del feto después de las 10 semanas de gestación. Se reportan sobrerepresentaciones de los cromosomas 21, 18 y 13, así como las aneuploidías de los cromosomas sexuales XO, XXX, XXY y XYY. Esta prueba de detección no detecta aneuploidías en otros cromosomas no mencionados anteriormente y, por lo tanto, no puede excluir anomalías en estos.

Incluye: AAAS, AARS1, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAN, ACAT1, ACE, ACO2, ACOX1, ACP5, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTL6B, ACTN2, ACVR2B, ACVRL1, ACY1, ADA, ADAM17, ADAM22, ADAMTS19, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRHL2, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGR2, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, AKT1, AKT2, AKT3, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALPL, ALS2, ALX3, ALX4, AMACR, AMER1, AMN, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKRD11, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARL6IP1, ARMC4, ARMC9, ARNT2, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD3A, ATCAY, ATIC, ATL1, ATM, ATOH7, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B3, ATP5MD, ATP6AP2, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT7, B4GAT1, B9D1, B9D2, BAG3, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BGN, BHLHA9, BICD2, BIN1, BLM, BLNK, BMP1, BMP2, BMP4, BMPER, BMPR1B, BOLA3, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRIP1, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C12orf65, C15orf41, C19orf12, C1QBP, C2CD3, C8orf37, CA2, CA5A, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CAD, CAMK2A, CAMK2B, CAMTA1, CANT1, CARD11, CARS2, CASK, CASP10, CASR, CAV1, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC114, CCDC115, CCDC151, CCDC22, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCND2, CCNK, CCNO, CCNQ, CCT5, CD19, CD247, CD2AP, CD3D, CD3E, CD3G, CD40, CD40LG, CD79A, CD79B, CD96, CDC45, CDH1, CDH2, CDH23, CDH3, CDK10, CDK13, CDK19, CDK19, CDK5RAP2, CDKL5, CDKN1C, CDON, CDSN, CDT1, CENPF, CENPJ, CEP104, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CEP83, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CILK1, CISD2, CIT, CKAP2L, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COROA1, COX10, COX15, COX20, COX6B1, COX7B, CPLANE1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREB3L1, CREBBP, CRELD1, CRIPT, CRKL, CRLF1, CRPPA, CRTAP, CRX, CRYAA, CRYAB, CSF1R, CSF2RB, CSF3R, CSNK1E, CSNK2A1, CSNK2B, CSPP1, CSTA, CSTB, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTNS, CTPS1, CTSA, CTSD, CTSK, CTU2, CUL3, CUL4B, CUL7, CWC27, CWF19L1, CXCR4, Cxorf56, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCC, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DDX6, DEAF1, DENND5A, DEPDC5, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHTKD1, DHX16, DHX30, DHX37, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DLL3, DLL4, DLX5, DMD, DMP1, DMPK, DMXL2, DNA2, DNAAF3, DNAAF4, DNAAF5, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC3, DNAJC5, DNM1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPF2, DPM1, DPM2, DPYD, DRC1, DSE, DSG1, DSP, DST, DSTYK, DTNA, DUOX2, DUOXA2, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, EDA, EDAR, EDN3, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFNB1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELAC2, ELANE, ELMO2, ELN, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EMX2, ENG, ENPP1, ENTPD1, EOGT, EP300, EPB42, EPCAM, EPG5, EPHB4, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERLIN1,

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC8, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, F10, F13A1, F2, F7, F8, F9, FA2H, FADD, FAH, FAM111A, FAM126A, FAM149B1, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FAS, FASLG, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXW11, FBXW4, FCSK, FEZF1, FGA, FGB, FGD1, FGD4, FGF10, FGF12, FGF3, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGG, FH, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FN1, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXL2, FOXN1, FOXP1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FTCD, FTL, FTSJ1, FUCA1, FUT8, FXN, G6PC, G6PC3, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GAS8, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GDAP1, GDF1, GDF3, GDF5, GDF6, GDI1, GEMIN4, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJA8, GJB2, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNA11, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPT2, GPX4, GREB1L, GRHL2, GRHL3, GRIA2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GSS, GTF2H5, GTPBP1, GTPBP3, GUCY2C, GUF1, GUSB, GYS1, GZF1, H1-4, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HAX1, HBA1, HBA2, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HINT1, HIVEP2, HK1, HLCS, HMGCL, HMGCS2, HMX1, HNF1B, HNF4A, HNRNPH2, HNRNPK, HNRNPU, HOXA1, HOXA13, HOXC13, HOXD13, HPD, HPDL, HPGD, HPRT1, HPS1, HPSE2, HRAS, HSD11B2, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HUWE1, HYAL1, HYDIN, HYLS1, IARS1, IARS2, IBA57, ICOS, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFNGR2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IGF1, IGF1R, IGF2, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKB, IKBKG, IL11RA, IL12RB1, IL1RAPL1, IL21R, IL2RA, IL2RG, IL7R, IMPAD1, INPP5E, INPP5K, INPPL1, INS, INSR, INVS, IPO8, IQCB1, IQSEC1, IQSEC2, IRAK4, IRF6, IRF8, IRX5, ISCA2, ITFG2, ITGA3, ITGA6, ITGA7, ITGA8, ITGB4, ITK, ITPA, ITPR1, IVD, JAG1, JAGN1, JAK3, JAM2, JAM3, KANK1, KANSL1, KARS, KARS1, KAT5, KAT6A, KAT6B, KAT8, KATNB1, KBTBD13, KCNA2, KCNB1, KCNC1, KCNC3, KCNE1, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ6, KCNMA1, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD1, KCTD7, KDM5C, KDM6A, KIAA0586, KIAA1109, KIDINS220, KIF11, KIF1A, KIF1C, KIF22, KIF2A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KLF1, KLHL15, KLHL40, KLHL41, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT10, KRT8, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LAS1L, LBR, LDB3, LEMD3, LETM1, LFNG, LGI4, LHX3, LHX4, LIAS, LIFR, LIG4, LINS1, LIPA, LIPT1, LMBRD1, LMNA, LMOD3, LMX1B, LONP1, LPIN2, LRBA, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LRRC6, LTBP2, LTBP3, LYRM7, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAG, MAGEL2, MAGI2, MAGT1, MALT1, MAMLD1, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K1, MAP3K7, MAPK8IP3, MAPRE2, MARS1, MASP1, MAT1A, MATN3, MBD5, MBOAT7, MBTPS2, MC2R, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MDH2, MECOM, MECP2, MECR, MED12, MED13, MED13L, MED17, MED23, MED25, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESP2, METTL5, MFN2, MFRP, MFSD2A, MFSD8, MGAT2, MGME1, MGP, MICU1, MID1, MIPEP, MIR17HG, MITF, MKKS, MKS1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP13, MMP21, MMUT, MN1, MNX1, MOCS1, MOCS2, MOGS, MORC2, MORC2, MPDU1, MPDZ, MPI, MPLKIP, MPV17, MPZ, MRE11, MRPL3, MRPL44, MRPS2, MRPS22, MRPS34, MSL3, MSMO1, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTO1, MTOR, MTR, MTRR, MTTP, MUSK, MUTYH, MVK, MYBPC1, MYBPC3, MYCN, MYD88, MYH3, MYH6, MYH7, MYH8, MYH9, MYL3, MYO18B, MYO5A, MYO5B, MYO7A, MYOD1, MYPN, MYT1L, NAA10, NAA15, NACC1, NADSYN1, NAGA, NAGLU, NAGS, NALCN, NANS, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NBN, NCAPD3, NCKAP1L, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEMF, NEU1, NEUROG3, NEXMIF, NF1, NFASC, NFIA, NFIB, NFIX, NFKBIA, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPAL4, NIPBL, NKX2-1, NKX2-5, NKX2-6, NKX3-2, NKX6-2, NLRC4, NLRP3, NMNAT1, NNT, NODAL, NOG, NONO, NOTCH1, NOTCH2, NOTCH3, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR1H4, NR2F1, NR2F2, NR3C2, NR5A1, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, NT5C2, NT5C3A, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP107, NUP188, NYX, OBSL1, OCLN, OCRL, ODAPH, OFD1, OGDH, OLA1, OPA1, OPA3, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTC, OTOGL, OTUD5, OTUD6B, OTULIN, OTX2, OXCT1, OXR1, P3H1, P4HB, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PALB2, PAM16, PANK2, PAPSS2, PARN, PARS2, PAX2, PAX3, PAX6, PAX8, PC, PCARE, PCBD1, PCCA, PCCB, PCDH15, PCDH19, PCGF2, PCK1, PCNT, PCYT1A, PCYT2, PDCD10, PDE10A, PDE4D, PDE6D, PDE6G, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PERCC1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHACTR1, PHEX, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHKG2, PHOX2B, PIEZO1, PIEZO2, PIGA, PIGG, PIGK, PIGL, PIGN, PIGO, PIGQ, PIGS, PIGT, PIGV, PIGY, PIH1D3, PIK3CA, PIK3CD, PIK3R1, PIK3R2, PISD, PITX1,

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

PITX2, PITX3, PKD1L1, PKHD1, PKLR, PLA2G6, PLAA, PLCB1, PLCB4, PLCE1, PLCG2, PLEC, PLEKHG5, PLG, PLK4, PLOD1, PLOD2, PLOD3, PLP1, PLPBP, PLS3, PMM2, PMP22, PMPCB, PMS2, PNKP, PNP, PNPLA1, PNPLA8, PNPO, PNPT1, POC1A, POC1B, POGZ, POLA1, POLD1, POLE, POLG, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMP, POMT1, POMT2, POR, PORCN, POU1F1, PPA2, PPIB, PPM1D, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM12, PRDM16, PREPL, PRF1, PRICKLE1, PRKAG2, PRKAR1A, PRKCD, PRKD1, PRMT7, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PRRX1, PRSS12, PRSS56, PRUNE1, PRX, PSAP, PSAT1, PSMB8, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTH1R, PTHLH, PTPN11, PTPN14, PTPN23, PTPRC, PTRH2, PTS, PUF60, PURA, PUS1, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB18, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAC2, RAD21, RAD50, RAD51C, RAF1, RAG1, RAG2, RAI1, RALGAPA1, RAPSN, RARB, RARS1, RARS2, RASA1, RAX, RB1, RBBP8, RBCK1, RBM10, RBM8A, RBPJ, RDH11, RELN, REN, RERE, RET, RETREG1, RFT1, RFX5, RFX6, RHOA, RHOBTB2, RIC1, RIMS2, RIN2, RIPK4, RIT1, RLIM, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, ROBO3, ROGDI, ROR2, RORA, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL26, RPL5, RPS19, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RTEL1, RTN4IP1, RTTN, RUNX2, RXYLT1, RYR1, SACS, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1, SARS2, SASS6, SATB2, SBDS, SBF1, SC5D, SCAMP5, SCAPER, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCNN1A, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINF1, SERPINH1, SET, SETBP1, SETD1A, SETD2, SETD5, SETX, SF3B4, SFTPB, SFTPC, SFXN4, SGCA, SGCE, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SHANK1, SHANK2, SHH, SHOC2, SHOX, SHROOM4, SIK1, SIL1, SIM1, SIN3A, SIX1, SIX3, SIX5, SKI, SKIV2L, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC26A2, SLC26A3, SLC26A7, SLC27A4, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMCHD1, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNAP29, SNORD118, SNRPB, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX17, SOX2, SOX3, SOX5, SOX6, SOX9, SP110, SP7, SPAG1, SPART, SPAST, SPATA5, SPECC1L, SPEG, SPG11, SPR, SPRED1, SPTAN1, SPTBN2, SPTBN4, SPTLC2, SQSTM1, SRCAP, SRD5A3, SRP54, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STAR, STAT2, STAT5B, STIL, STIM1, STRA6, STRADA, STS, STX11, STX1B, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SUZ12, SVBP, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAB2, TAC3, TACR3, TAF1, TAF13, TAF2, TAF6, TANGO2, TAOK1, TASP1, TAT, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCF12, TCF20, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TECPR2, TEK, TELO2, TERT, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, TH, THAP1, THOC2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, ROBO3, ROGDI, ROR2, RORA, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL26, RPL5, RPS19, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RTEL1, RTN4IP1, RTTN, RUNX2, RXYLT1, RYR1, SACS, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1, SARS2, SASS6, SATB2, SBDS, SBF1, SC5D, SCAMP5, SCAPER, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCNN1A, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINF1, SERPINH1, SET, SETBP1, SETD1A, SETD2, SETD5, SETX, SF3B4, SFTPB, SFTPC, SFXN4, SGCA, SGCE, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SHANK1, SHANK2, SHH, SHOC2, SHOX, SHROOM4, SIK1, SIL1, SIM1, SIN3A, SIX1, SIX3, SIX5, SKI, SKIV2L, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC26A2, SLC26A3, SLC26A7, SLC27A4, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMCHD1, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNAP29, SNORD118, SNRPB, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX17, SOX2, SOX3, SOX5, SOX6, SOX9, SP110, SP7, SPAG1, SPART, SPAST, SPATA5, SPECC1L, SPEG, SPG11, SPR, SPRED1, SPTAN1, SPTBN2, SPTBN4, SPTLC2, SQSTM1, SRCAP, SRD5A3, SRP54, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STAR, STAT2, STAT5B, STIL, STIM1, STRA6, STRADA, STS, STX11, STX1B, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SUZ12, SVBP, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAB2, TAC3, TACR3, TAF1, TAF13, TAF2, TAF6, TANGO2, TAOK1, TASP1, TAT, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCF12, TCF20, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TECPR2, TEK, TELO2, TERT, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1,

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

TH, THAP1, THOC2, THOC6, THRA, TIMM8A, TINF2, TJP2, TK2, TKT, TLK2, TMCO1, TMEM107, TMEM126B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMEM94, TMPRSS6, TMTC3, TMX2, TNFRSF11A, TNFRSF11B, TNFRSF13B, TNFSF11, TNNT1, TOE1, TOP3A, TP53, TP63, TPI1, TPK1, TPM2, TPO, TPP1, TRAPPC11, TRAPPC2, TRAPPC4, TRAPPC9, TREX1, TRIM2, TRIM32, TRIM37, TRIO, TRIP11, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMU, TRNT1, TRPM1, TRPM6, TRPS1, TRPV3, TRPV4, TRPV6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPYL1, TTC19, TTC21B, TTC26, TTC37, TTC7A, TTC8, TTN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TWNK, TXNL4A, TYMP, TYR, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3A, UBE3B, UBR1, UBTF, UCHL1, UFC1, UFM1, UGDH, UGP2, UGT1A1, UMPS, UNC13D, UNC80, UPB1, UPF3B, UQCC2, UQCRC2, UROC1, UROS, USB1, USH1C, USH1G, USH2A, USP18, USP53, USP9X, UVSSA, VAMP1, VAMP2, VANGL1, VARS2, VDR, VIPAS39, VLDLR, VPS13B, VPS33B, VPS37A, VPS41, VPS45, VPS53, VRK1, VSX2, WAC, WAS, WASF1, WASHC5, WDFY3, WDPCP, WDR11, WDR19, WDR26, WDR34, WDR35, WDR37, WDR45, WDR45B, WDR60, WDR62, WDR73, WDR81, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WT1, WWOX, XIAP, XPA, XPC, XRCC2, XRCC4, XYLT1, XYLT2, YAP1, YARS2, YIF1B, YWHAG, YY1, ZAP70, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFP57, ZFPM2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMIZ1, ZMPSTE24, ZMYM6, ZMYND10, ZMYND11, ZNF335, ZNF423, ZNF462, ZNF711, ZNHIT3

Estudios de un solo gen

46,XX inversión sexual tipo 1 SRY
46,XY inversión sexual tipo 8, modificador de AKR1C4
Deficiencia de aromatasa CYP19A1
Azoospermia inducida por microdeleciones del cromosoma Y AZF
Ausencia congénita bilateral de conductos deferentes CFTR
Criptorquidia RXFP2
Sordera e infertilidad masculina STRC
Sordera e infertilidad masculina, relacionadas con CATSPER2
Trastornos del desarrollo sexual con paladar hendido FOXF2
Deficiencia de hormona foliculoestimulante, aislada FSHB
síndrome de gutmacher HOXA13
Síndrome mano-pie-útero HOXA13
Mola hidatidiforme NLRP7
Mola hidatiforme, recurrente, tipo 2 KHDC3L
Hipogonadismo hipogonadotrópico KISS1
Hipogonadismo hipogonadotrópico NSMF
Hipogonadismo hipogonadotrópico tipo 6 FGF8
Hipogonadismo hipogonadtrópico tipo 14 WDR11
Hipospadias tipo 1, ligada al cromosoma X Arkansas
Hipospadias tipo 2, ligada al cromosoma X MAMLD1
Hipoplasia de células de Leydig tipo 1 LHCGR
Oligo-asteno-teratozoospermia NANOS1
Defecto de maduración de ovocitos ZP1
disfunción de la ovogénesis SOHLH1
Disgenesia ovárica tipo 1 FSHR
Disgenesia ovárica tipo 2 BMP15
Síndrome persistente del conducto de Müller tipo 1 AMH
Síndrome persistente del conducto de Müller tipo 2 AMHR2
Preeclampsia/eclampsia tipo 5 CORIN
Pérdida de embarazo, recurrente, relacionada con C4BPA
Pseudohermafroditismo con ginecomastia HSD17B3
SPGF4 SYCP3
SPGF5 AURKC
SPGF6 ESPATA16
SPGF7 CATSPER1
SPGF8 NR5A1
SPGF9 DPY19L2
Anomalías testiculares con o sin cardiopatía congénita GATA4

Se recomienda la prueba de un solo gen en pacientes que tienen:

- Características clínicas distintivas
- Antecedentes familiares de un trastorno específico.
- Trastornos de un solo gen
- Posible trastorno epigenético
- Posibles trastornos de repetición triple
- Confirmación de prueba familiar

Gen med

Laboratorio de diagnóstico clínico y genómico

Oncología

CentoBreast

CentoBreast detecta mutaciones en los genes BRCA1 y BRCA2, que son las causas hereditarias más comunes del cáncer de mama. Además, nuestro panel incluye otros genes como ABRAXAS1, ATM, BARD1, BRIP1, CDH1, CHEK2, DICER1, EPCAM, FANCC, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53, XRCC2, que también se han asociado con un mayor riesgo de cáncer.

CentoCancer

Cada gen en CentoCancer ha sido cuidadosamente seleccionado en función de su potencial de riesgo en el desarrollo de uno o más de los siguientes cánceres: mama, ovario, colorrectal, gástrico, tiroides, endometrio, páncreas, melanoma, renal y próstata. Este panel es apropiado para pacientes con antecedentes personales positivos de cáncer de aparición temprana, cáncer raro, cáncer bilateral o cánceres primarios múltiples.

Incluye 70 genes: ABRAXAS1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, DIS3L2, EPCAM, FANCC, FH, FLCN, GALNT12, HNF1B, HOXB13, KIT, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS1, PMS2, POLD1, POLE, POT1, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TGFBR2, TP53, TSC1, TSC2, VHL, WT1, XRCC2, XRCC3

Panel Completo de Cáncer

CentoCancer complete es nuestro panel de cáncer más extenso y cubre una gran cantidad de genes asociados con el cáncer. Cada gen de este panel ha sido cuidadosamente seleccionado en función de su potencial de riesgo en el desarrollo de uno o más de los siguientes cánceres: mama, ovario, colorrectal, gástrico, tiroides, endometrio, páncreas, melanoma, renal y próstata, entre otros.

Incluye 110 genes: ABRAXAS1, ACVRL1, AKT1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, DDX41, DICER1, DIS3L2, EGFR, EPCAM, ETV6, EXT1, EXT2, FANCC, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, REST, RET, RNF43, RPS20, RUNX1, SAMD9L, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERT, TGFBR2, TMEM127, TP53, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XRCC2, XRCC3

CentoColon

CentoColon detecta genes que están asociados con el cáncer de colon, páncreas y gástrico.

Incluye: APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, EPCAM, FLCN, GALNT12, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PRSS1, PTEN, RNF43, SMAD4, STK11, TGFBR2, TP53, VHL.

Gen med

Laboratorio de diagnóstico clínico y genómico

Oncología

Estudios de un solo gen

Se recomienda la prueba de un solo gen en pacientes que tienen:

- Características clínicas distintivas
- Antecedentes familiares de un trastorno específico.
- Trastornos de un solo gen
- Posible trastorno epigenético
- Posibles trastornos de repetición triple
- Confirmación de prueba familiar

TOME EN CUENTA QUE LOS GENES PUEDEN ESTAR ALTERADOS EN OTRAS NEOPLASIAS, EN LA LISTA SE DESCRIBE LA MÁS COMUNMENTE ASOCIADA

Oncogen ABL ABL1
Prolactinoma y adenomas pituitarios AIP
Oncogen AKT1
Marcador endotelial tumoral ANTXR1
Gen supresor de tumores APC
Síndrome de predisposición tumoral BAP1
Gen supresor de tumores BRCA1
Síndromes mieloproliferativos CALR
Protoncogen CBL
Neoplasia endócrina múltiple CDKN1B
Asociado a melanoma cutáneo CDKN2A
Asociado a cancer gástrico CTNNA1
Tumorogénesis en cabeza y cuello CYLD
Factor de crecimiento EGFR
Neoplasias hematológicas EZH2
Tumorogenesis pulmonar IPO8
Carcinoma renal papilar hereditario MET
Síndrome de Lynch MLH1
Cáncer endometrial MLH3
Síndrome de Lynch MSH2
Síndrome de Lynch MSH6
Supresor de tumores MSR1
Poliposis MUTYH
Neurofibromatosis NF1
Neurofibromatosis NF2
Síndrome tumoral NTHL1
Oncogen PIK3CA
Cáncer endometrial PPP2R1A
Carcinomas fibrolamelares PRKCA
Síndrome de carcinoma nevoide de células basales PTCH2
Cáncer pancreático PTEN
Síndromes de Noonan y LEOPARD PTPN11
Cáncer intestinal RAD50
Cáncer de mama y ovario RAD51C
Cánceres epiteliales REST
Cáncer prostático hereditario RNASEL
Mielodisplasias SAMD9L
Paraganglioma-feocromocitoma tipo 2 hereditario SDHAF2
Carcinoma de células renales SDHB
Tumores neuroendócrinos SDHC
Cánceres hematológicos SF3B1
Metástasis de Cáncer de mama SMARCE1
Supresor de tumores STAG1
Cáncer pulmonar STK11
Cáncer pulmonar TAF15
Supresor de tumores TP53
Cáncer gástrico ATP6V1A
Cáncer de pulmón de células no pequeñas CACNA1E
Factor de necrosis tumoral CD70
Cáncer renal y cáncer colorrectal CNPY3
Fenotipo maligno de cáncer gástrico CPLX1
Supresor de tumores DENND5A
Cáncer de pulmón ERBB4
Cáncer ovárico PPM1D
Retinoblastoma RB1
Meduloblastoma SUFU
Cáncer de células renales AP3B2
Asociado a riesgo de cáncer de pulmón CHRNA3
Sarcoma de células claras de tejido blando EWSR1
Carcinoma de endometrio FGFR2
Cáncer cervicouterino FYB1
Cáncer de colon familiar GABRA2
Linfoma difuso de células B grandes GNB1
Cáncer pancreatico GOT2
Cáncer colorrectal RAPIGDS1
Pronóstico en cáncer de mama SASH1

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,

Gen med

Laboratorio de diagnóstico clínico y genómico

Desordenes metabólicos

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

Gen med

Laboratorio de diagnóstico clínico y genómico

Desordenes metabólicos

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,

Gen med

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

Gen med

Laboratorio de diagnóstico clínico y genómico

Oftalmología

CentoVisión

Diseñado para encontrar la base genética de las enfermedades oculares, incluidas aquellas que son las principales causas de ceguera entre los bebés (amaurosis congénita de Leber), los niños (retinosis pigmentaria de aparición temprana) y los adultos (distrofia de patrón). Nuestro panel incluye las enfermedades oftalmológicas más comunes, como el glaucoma congénito, la retinosis pigmentaria, la enfermedad de Stargardt, el síndrome de Stickler, la acromatopsia y el síndrome de Usher, entre otras. También detecta diferentes tipos de albinismo (oculocutáneo y ocular), así como el síndrome de Hermasky-Pudlak.

Incluye 446 genes: ABCA4, ABCB6, ABHD12, ACO2, ACVR1, ADAM9, ADAMTS18, ADAMTSL4, ADGRV1, AFG3L2, AGBL5, AGK, AHI1, AIPL1, ALDH18A1, ALDH1A3, AP3B1, APTX, ARHGEF18, ARL13B, ARL2BP, ARL6, ARSG, ASB10, ATF6, ATOH7, AUH, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, C1QTNF5, C2CD3, CA4, CABP4, CACNA1F, CACNA2D4, CANT1, CAPN5, CC2D2A, CCDC28B, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP290, CEP41, CERKL, CFAP418, CHD7, CHM, CHMP4B, CIB2, CISD2, CLCN7, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COX7B, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CSPP1, CTC1, CTDP1, CTNNA1, CTNNB1, CTSD, CWC27, CYP1B1, CYP27A1, CYP4V2, DGUOK, DHDDS, DHX38, DKC1, DNA2, DNAJC19, DNM1L, DRAM2, DTNBP1, EDN3, EDNRB, EFEMP1, ELOVL4, ENPP1, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, EYS, FAM126A, FAM161A, FDXR, FLVCR1, FOXC1, FOXL2, FRAS1, FREM1, FREM2, FSCN2, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GBA, GCNT2, GDF3, GDF6, GJA1, GJA3, GJA8, GNAT2, GNPTG, GPR143, GRIP1, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HESX1, HEXA, HGSNAT, HK1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, HTRA2, IDH3B, IFT140, IFT172, IFT27, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA0586, KIF11, KIF7, KIT, KLHL7, LCA5, LEMD2, LEP, LEPR, LIM2, LMX1B, LOXL1, LRAT, LRMDA, LRP2, LRP5, LSS, LTBP2, LYST, LZTFL1, MAB21L2, MAF, MAK, MC1R, MERTK, MFN2, MFRP, MFSD8, MIP, MITF, MKKS, MKS1, MLPH, 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, MTRFR, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NF2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR0B2, NR2F1, NRL, NTF4, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPTN, OSTM1, OTX2, P3H2, PAX2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PHF6, PHYH, PIK3R5, PITPNM3, PITX2, PITX3, PLA2G5, PNKP, PNPLA6, POC1B, POLG, POLG2, POMC, POMGNT1, PPARG, PPT1, PQBP1, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PXDN, RAB18, RAB27A, RAB28, RAB3GAP1, RAB3GAP2, RARB, RAX, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRM2B, RS1, RTN4IP1, SAG, SBF2, SDCCAG8, SEMA4A, SERAC1, SETX, SHH, SIL1, SIX3, SIX6, SLC16A12, SLC24A5, SLC25A4, SLC25A46, SLC33A1, SLC38A8, SLC45A2, SLC52A2, SLC7A14, SLC9A6, SMCHD1, SMOC1, SNAI2, SNRNP200, SNX10, SOX10, SOX2, SPATA7, SPG7, STRA6, TBC1D20, TBK1, TCIRG1, TCTN1, TCTN2, TCTN3, TDRD7, TEK, TENM3, TFAP2A, TIMM50, TIMM8A, TIMP3, TK2, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TNFRSF11A, TNFSF11, TOPORS, TPP1, TRAF3IP1, TRIM32, TRNT1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TULP1, TWNK, TYMP, TYR, TYRP1, USH1C, USH1G, USH2A, VCAN, VIM, VPS13B, VSX2, WDPCP, WDR19, WDR36, WFS1, WHRN, WRN, ZIC2, ZNF408, ZNF423, ZNF513

Gen med

Laboratorio de diagnóstico clínico y genómico

Otorrinolaringología

CentoHear

La pérdida de audición es una condición común en los niños, que afecta a 1 de cada 100 nacidos vivos. En más del 50% de los casos existe una causa genética para este trastorno, de los cuales el 70% son hipoacusias no sindrómicas. Genes asociados con la pérdida auditiva sindrómica y no sindrómica, casos autosómicos recesivos y dominantes. Además, síndromes como Alport, Pendred, Waardenburg, Usher y branchio-oto-renal entre otros.

Incluye 196 genes: ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GAB1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MGP, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PJVK, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TWNK, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN

Gen med

Laboratorio de diagnóstico clínico y genómico

Inmunología

CentoInmuno

Solución para la inmunodeficiencia y los trastornos de inmunodeficiencia combinada grave (SCID). Nuestro panel incluye genes dirigidos a la inmunodeficiencia combinada grave, la neutropenia congénita, la deficiencia de anticuerpos primarios, la inmunodeficiencia variable común, la enfermedad granulomatosa crónica, la linfoproliferación autoinmune y la agammaglobulinemia.

Incluye 326 genes: ACD, ACP5, ACTB, ADA, ADA2, ADAR, AICDA, AIRE, AK2, AP1S3, AP3B1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CEBPE, CFAP298, CFB, CFD, CFH, CFHR1, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COPA, CORO1A, CR2, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE1L3, DNMT3B, DOCK2, DOCK8, DRC1, DTNBP1, ELANE, EPG5, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCN3, FERMT3, FGA, FGB, FGG, FOXN1, FOXP3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GUCY2C, HAX1, HELLS, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYDIN, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF3, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, KRAS, LAMTOR2, LAT, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MASP2, MBL2, MCM4, MEFV, MOGS, MS4A1, MSN, MTHFD1, MVK, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCSTN, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE3, OFD1, OSTM1, OTULIN, PARN, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PMM2, PMS2, PNP, POLA1, POLE, PRF1, PRKCD, PSENEN, PSMB4, PSMB8, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RSPH1, RSPH4A, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEMA3E, SERPING1, SH2D1A, SKIV2L, SLC29A3, SLC35C1, SLC7A7, SMARCAL1, SMARCD2, SP110, SPAG1, SPINK5, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STING1, STK4, STX11, STXBP2, TAFAZZIN, TAP1, TAP2, TAPBP, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERT, TFRC, THBD, TICAM1, TINF2, TLR3, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF11, TPP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10

Gen med

Laboratorio de diagnóstico clínico y genómico

Dismorfología

CentoDismorfia

Diseñado para ayudar a los médicos a diagnosticar a los pacientes que padecen un síndrome dismórfico. El panel incluye craneosinostosis, trastornos craneofaciales, paladar hendido/labio, holoprosencefalia, síndrome de Waardenburg, enfermedad de Hirschsprung, lisencefalia y trastornos de malformación cerebral, entre otros.

Incluye 770 genes: A2ML1, ABCA12, ABCB6, ABCC6, ABL1, ACP5, ACTA1, ACTA2, ACTB, ACTG1, ACVR2B, ADAMTS18, ADAMTS2, ADAMTSL2, ADGRG1, ADGRG6, AEBP1, AFF4, AGPS, AGRN, AHDC1, AHI1, AKR1C4, AKT3, ALDH18A1, ALDH1A3, ALG2, ALMS1, ALPL, ALX1, ALX4, AMELX, AMER1, AMH, AMHR2, AMPD2, ANKH, ANKLE2, ANKRD11, ANKS6, ANO5, ANOS1, AP4M1, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARSL, ARX, ASPM, ASXL1, ASXL3, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1E1, ATP7A, ATR, ATRX, B3GALNT2, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCL11A, BCOR, BGN, BIN1, BMP1, BMP2, BMP4, BMPR1B, BNC2, BRAF, C1R, C1S, CANT1, CASK, CASR, CBL, CBS, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2, CCN6, CCNO, CDC45, CDH1, CDK13, CDK5RAP2, CDKN1C, CDON, CENPF, CENPJ, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP418, CFAP53, CFL2, CHAT, CHD4, CHD7, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CILK1, CIT, CLCN5, CLP1, CNOT1, CNTNAP1, COASY, COG5, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COX7B, CRB2, CREB3L1, CREBBP, CRELD1, CRIPT, CRPPA, CRTAP, CSGALNACT1, CSPP1, CTNNA2, CTNND1, CUL4B, CUL7, CYB5A, CYP19A1, CYP1B1, CYP26B1, DAG1, DCC, DCHS1, DCX, DDR2, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB13, DNAL1, DNM2, DOCK6, DOK7, DPAGT1, DPF2, DRC1, DSE, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2LI1, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFNB1, EFTUD2, EGR2, EIF2AK3, EIF2S3, ELN, ENPP1, EOGT, EPG5, ERCC1, ERCC2, ERCC5, ERCC6, ERF, ESCO2, EVC, EVC2, EXOSC3, EXOSC9, EXT1, EXT2, EYA1, FAM20C, FAS, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL4, FBXW11, FGD1, FGF10, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLCN, FLNA, FLNB, FOXC1, FOXE1, FOXL2, FRAS1, FREM1, FREM2, FZD6, GAS8, GATA4, GBA, GBE1, GDF1, GDF3, GDF5, GDF6, GDNF, GFPT1, GGCX, GJA1, GLE1, GLI2, GLI3, GMPPB, GNAS, GNPAT, GORAB, GPC3, GPC6, GRHL3, GRIP1, HBA1, HCCS, HDAC8, HES7, HESX1, HEXA, HMX1, HOXA13, HOXD13, HRAS, HSPG2, HUWE1, HYDIN, HYLS1, IER3IP1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IGF1R, IHH, IL11RA, INPP5E, INPPL1, INVS, IPO8, IRF6, ITGB4, JAG1, KAT6B, KATNB1, KBTBD13, KDM5C, KDM6A, KIAA0586, KIF11, KIF14, KIF22, KIF2A, KIF5C, KIF7, KIFBP, KIT, KLHL40, KLHL41, KMT2A, KMT2D, KNL1, KRAS, KRIT1, L1CAM, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LARGE1, LBR, LFNG, LIFR, LMNA, LMOD3, LMX1B, LOX, LRP2, LRP4, LRP5, LTBP3, LZTFL1, LZTR1, MAB21L2, MACF1, MAFB, MAGEL2, MAP2K1, MAP2K2, MASP1, MATN3, MBTPS2, MCIDAS, MCPH1, MED12, MED13L, MED17, MEGF8, MEIS2, MEOX1, MESP2, MFAP5, MFRP, MFSD2A, MGP, MID1, MITF, MKKS, MKS1, MMP13, MMP15, MMP21, MMP9, MN1, MRAS, MSMO1, MSX1, MSX2, 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, MUSK, MYBPC1, MYCN, MYH11, MYH2, MYH3, MYH8, MYLK, MYO18B, MYO7A, MYO9A, NAA10, NALCN, NBAS, NCAPD3, NDE1, NDP, NEB, NECTIN1, NEK1, NEK8, NEK9, NEPRO, NF1, NF2, NHEJ1, NIPBL, NKX2-5, NKX3-2, NME8, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPR2, NR2F2, NRAS, NRG1, NSD1, NSDHL, NSUN2, NT5E, OBSL1, OCRL, ODAD4, OFD1, OPHN1, ORC1, OTX2, P3H1, P4HB, PAFAH1B1, PAPSS2, PAX2, PAX3, PAX6, PAX7, PAX9, PCNT, PDCD10, PDE4D, PDE6D, PEX7, PGM1, PHEX, PHF6, PHF8, PHYH, PIBF1, PIEZO2, PIGA, PIGV, PIP5K1C, PITX2, PKD1L1, PKD2, PKHD1, PLEKHA7, PLK4, PLOD1, PLOD2, PLP1, PMM2, PNKP, PNPLA6, POLA1, POLR1C, POLR1D, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PORCN, PPARG, PPIB, PPP1CB, PQBP1, PRDM5, PREPL, PRKAR1A, PRKG1, PRPH2, PRSS56, PTCH1, PTH1R, PTHLH, PTPN11, PXDN, PYCR1, PYCR2, QARS1, RAB18, RAB23, RAB3GAP1, RAB3GAP2, RAD21, RAF1, RAPSN, RARB, RARS2, RASA1, RASA2, RAX, RBBP8, RBP4, RBPJ, RELN, RET, RHO, RIN2, RIPK4, RIPPLY2, RIT1, ROBO3, ROR2, RPGRIP1L, RPL10, RPS6KA3, RSPH1, RSPH3, RSPH4A, RSPH9, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SASS6, SATB2, SBDS, SCN4A, SDCCAG8, SEC24D, SELENON, SEMA3E, SEPSECS, SERPINF1, SERPINH1, SF3B4, SH3PXD2B, SHH, SHOC2, SHROOM4, SIX3, SIX6, SKI, SLC18A3, SLC25A19, SLC26A2, SLC2A10, SLC34A3, SLC35D1, SLC38A8, SLC39A13, SLC5A7, SLC9A6, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMCHD1, SMOC1, SMS, SNAI2, SNAP29, SNTG1, SOS1, SOS2, SOX10, SOX11, SOX2, SOX9, SP7, SPAG1, SPARC, SPECC1L, SPRED1, SPRY4, STAMBP, STIL, STRA6, SUFU, SYT2, TAF6, TBC1D20, TBC1D23, TBC1D24, TBX1, TBX15, TBX2, TBX22, TBX3, TBX5, TBX6, TCF12, TCF4, TCOF1,

Gen med

Laboratorio de diagnóstico clínico y genómico

Dismorfología

TCTN1, TCTN2, TCTN3, TENM3, TENT5A, TFAP2A, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TK2, TMCO1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMTC3, TNFRSF11B, TNNI2, TNNT1, TNNT3, TNXB, TOE1, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRIM32, TRIP11, TRMT10A, TRPS1, TRPV4, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TTC21B, TTC8, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TXNL4A, TYR, UBA1, VAMP1, VCAN, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, WDR11, WDR19, WDR35, WDR62, WNT1, WNT5A, WNT7A, XYLT1, YWHAE, ZBTB24, ZC4H2, ZEB2, ZFPM2, ZIC1, ZIC2, ZIC3, ZMYND10, ZNF335, ZNF423, ZNF469, ZNF699, ZSWIM6

Panel de tejido conectivo y trastornos relacionados

Nuestro panel de tejido conectivo y trastornos relacionados proporciona una evaluación profunda en un solo paso de varios genes para detectar diferentes trastornos con fenotipos similares, como el síndrome de Marfan, Loeys-Dietz, cutis laxa, Ehlers-Danlos, síndrome de Stickler y aneurisma aórtico torácico familiar y disección.

Incluye 76 genes: ABCC6, ACTA2, ADAMTS2, ADAMTSL2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GAT3, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CREB3L1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, GORAB, ITGB4, LAMA3, LAMB3, LAMC2, LOX, LRP2, LTBP3, MBTPS2, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, SPARC, TENT5A, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, VCAN, WNT1, ZNF469

Gen med

Laboratorio de diagnóstico clínico y genómico

Dermatología

CentoSkin

Solución para pacientes que presentan trastornos de la piel. Nuestro panel incluye genes para hipotricosis, epidermólisis ampollosa e ictiosis congénita, entre otros. Para melanoma, consulte nuestra sección de Oncología.

Incluye 152 genes: ABCA12, ABHD5, ACD, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AP3B1, AP3D1, APCDD1, ARHGAP31, ARSL, ATP6V0A2, ATP7A, BLOC1S3, BLOC1S6, CASP14, CDSN, CERS3, CHST8, CLDN1, COL17A1, COL7A1, CPOX, CSTA, CTC1, CYP4F22, DKC1, DLL4, DOCK6, DSG1, DSG4, DSP, DST, DTNBP1, EBP, EDA, EDAR, EDARADD, EDN3, EDNRB, EFEMP2, ELN, ELOVL4, EOGT, EPG5, ERCC2, ERCC3, EXPH5, FBLN5, FECH, FERMT1, FLG, FLG2, GJB2, GJB3, GJB4, GJB6, GPR143, GTF2H5, HMBS, HPS1, HPS3, HPS4, HPS5, HPS6, HR, ITGA3, ITGA6, ITGB4, JUP, KCTD1, KDSR, KITLG, KRT1, KRT10, KRT14, KRT2, KRT5, KRT74, KRT85, KRT9, LAMA3, LAMB3, LAMC2, LIPH, LIPN, LORICRIN, LPAR6, LRMDA, LYST, MBTPS2, MC1R, MITF, MLPH, MMP1, MPLKIP, MYH9, MYO5A, NECTIN1, NHP2, NIPAL4, NOP10, NOTCH1, NSDHL, OCA2, PARN, PAX3, PEX7, PHGDH, PHYH, PKP1, PLEC, PNPLA1, POMP, PPOX, PSAT1, PYCR1, RAB27A, RBPJ, RPL21, RTEL1, SDR9C7, SERPINB8, SLC24A5, SLC27A4, SLC38A8, SLC45A2, SNAI2, SNAP29, SNRPE, SOX10, SPINK5, ST14, STS, SUMF1, TERT, TGM1, TGM5, TINF2, TYR, TYRP1, UROD, UROS, USB1, VPS33B, WNT10A, WRAP53, ZMPSTE24

Gen med

Laboratorio de diagnóstico clínico y genómico

Tejido óseo

Panel de mineralización anormal

Nuestro panel de mineralización anormal incluye osteogénesis imperfecta, osteopetrosis, trastornos de alta y baja densidad ósea y genes de diagnóstico diferencial necesarios para discriminar la causa genética real. Las enfermedades procesables, como la hipofosfatasia, también se incluyen en nuestro panel.

Incluye 94 genes: ABCC6, ALPL, AMER1, ANKH, ANO5, AP2S1, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CASR, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTSK, CYP24A1, CYP27B1, CYP2R1, DMP1, DSPP, ENPP1, FAH, FAM20C, FERMT3, FGF23, FGFR1, FGFR3, FKBP10, GALNT3, GJA1, GNA11, GNAS, GORAB, GPAA1, HPGD, HRAS, IFITM5, KRAS, LEMD3, LRP4, LRP5, MBTPS2, MESD, MTAP, NBAS, NOTCH2, NRAS, OCRL, OSTM1, P3H1, P4HB, PHEX, PLEKHM1, PLOD2, PLS3, PPIB, PTDSS1, PTH1R, SEC24D, SERPINF1, SERPINH1, SGMS2, SH3PXD2B, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC9A3R1, SLCO2A1, SNX10, SOST, SOX9, SP7, SPARC, SQSTM1, TAPT1, TBXAS1, TCIRG1, TGFB1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, TYROBP, VDR, WNT1, XYLT2, ZBTB20, TENT5A

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

Gen med

Laboratorio de diagnóstico clínico y genómico

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,

Gen med

Laboratorio de diagnóstico clínico y genómico

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.

Gen med

Laboratorio de diagnóstico clínico y genómico

Hematología

Panel de coagulación

Contiene genes para el diagnóstico molecular de trombofilia, trombocitopenia, telangiectasia hemorrágica hereditaria, síndrome ARC, síndrome de Hermasky-Pudlak, trastornos del factor de coagulación y trastornos relacionados con las plaquetas.

Incluye 112 genes: ABCG5, ABCG8, ACTN1, ACVRL1, ADAMTS13, ADAMTS2, ANKRD26, ANO6, AP3B1, ARPC1B, BLOC1S3, BLOC1S6, C1R, CCM2, CD36, CD40LG, CDC42, CHST14, COL1A2, COL3A1, COL4A1, COLGALT1, CTC1, CYCS, DIAPH1, DTNBP1, EFEMP2, EFL1, ENG, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FANCA, FCGR2C, FERMT3, FGA, FGB, FGG, FLI1, FLNA, FYB1, GATA1, GFI1B, GGCX, GNE, GP1BA, GP1BB, GP6, GP9, GUCY1A1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, IGFBP7, IL2RG, ITGA2B, ITGB3, JAM3, KDSR, KRIT1, LMAN1, LYST, LYZ, MCFD2, MECOM, MPL, MYH9, NBEAL2, P2RY12, PDCD10, PLA2G4A, PLAT, PLAU, PROC, PROS1, PTPN11, RBM8A, RUNX1, SBDS, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SLC35A1, SLC7A7, SLFN14, SMAD4, SRC, STIM1, STN1, TBXA2R, THBD, THPO, TUBB1, VIPAS39, VKORC1, VPS33B, VWF, WAS, WIPF1

Panel de anemia/insuficiencia de médula ósea

Está destinado a pacientes con anomalías en más de 2 tipos de células sanguíneas (glóbulos rojos, glóbulos blancos y plaquetas) que presentan síntomas de letargo, infecciones recurrentes, sangrado excesivo, pigmentación anormal, agrandamiento del bazo, y malignidades. Algunos trastornos específicos detectados con este panel son linfohistiocitosis hemofagocítica, síndrome de Seckel, trombocitopenia, anemia de Fanconi, disqueratosis congénita, síndrome de Shwachman Diamond así como otros tipos de anemias, como talasemia alfa y beta, enfermedad de células falciformes, esferocitosis, anemia megaloblástica, anemia congénita anemia sideroblástica y diseritropoyética.

Incluye 211 genes: ABCB6, ABCB7, ABCG5, ABCG8, ACD, ACTN1, ADA, ADA2, ADAMTS13, AK1, AK2, ALAS2, ALDOA, AMMECR1, AMN, ANK1, ANKRD26, AP3B1, ATM, ATRX, BLM, BLOC1S3, BRCA1, BRCA2, BRIP1, CASP10, CBL, CBLIF, CD36, CD40LG, CD59, CDAN1, CDC42, CENPJ, CEP152, CHEK2, CLCN7, CLPB, COL4A1, CSF3R, CTC1, CTLA4, CUBN, CXCR4, CYB5R3, CYCS, DHFR, DIAPH1, DKC1, DNAJC21, DTNBP1, EFL1, ELANE, EPB41, EPB42, ERCC4, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCGR2C, FLI1, G6PC3, G6PD, GATA1, GCLC, GFI1, GFI1B, GLRX5, GNE, GP1BA, GP1BB, GP9, GPI, GPX1, GSR, GSS, GYPC, HAX1, HBA1, HBA2, HBB, HBD, HFE, HK1, HMOX1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HSPA9, IKZF1, IL2RG, ITGA2B, ITGB3, ITK, JAGN1, KCNN4, KDM1A, KDSR, KIT, KLF1, KRAS, LIG4, LPIN2, LYST, MECOM, MLH1, MPIG6B, MPL, MRE11, MSH2, MSH6, MTR, MTRR, MYH9, NBEAL2, NBN, NF1, NFKB1, NHP2, NOP10, NRAS, NT5C3A, PALB2, PARN, PC, PDHA1, PDHX, PFKM, PGK1, PIEZO1, PKLR, PLAU, PMS2, PRF1, PTPN11, PUS1, RAB27A, RAC2, RAD51, RAD51C, RBBP8, RBM8A, REN, RHAG, RIT1, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SBDS, SEC23B, SH2D1A, SLC19A2, SLC19A3, SLC25A19, SLC25A38, SLC2A1, SLC35C1, SLC4A1, SLFN14, SLX4, SPTA1, SPTB, SRP72, STAT3, STIM1, STX11, STXBP2, TCN2, TERT, THPO, TINF2, TMPRSS6, TP53, TPI1, TPK1, TRNT1, UBE2T, UNC13D, VPS13B, VPS45, WAS, WNT4, WRAP53, XIAP, XK, XRCC2, YARS2

Gen med

Laboratorio de diagnóstico clínico y genómico

Hepatología y Nefrología

Panel de síndrome urémico hemolítico atípico

Contiene genes para el diagnóstico molecular de este síndrome.

Incluye 25 genes: ADAMTS13, C3, CD46, CD59, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CR1, CR2, DGKE, F12, G6PD, INF2, MMACHC, MMUT, PIGA, PLG, PRDX1, THBD, VWF

MLPA: CFHR1, CFHR2, CFHR3, CFHR5, CFH

CentoNefro

Aproximadamente el 10% de la población mundial se ve afectada por enfermedades renales crónicas. CentoNefro ofrece una herramienta integral para detectar los trastornos renales hereditarios más destacados, incluida la enfermedad renal poliquística, el síndrome de Alport, el panel de acidosis tubular renal, el panel de glomerulonefrosis focal y la hiperoxaluria primaria, entre otros. El análisis de PKD1 no está incluido en este panel.

Para incluir PKD1 solicite CentoNefro Plus.

Incluye 495 genes: ABCB11, ABCB4, ABCC2, ACE, ACP5, ACTG2, ACTN4, ACVR2B, AGPS, AGT, AGTR1, AHI1, AIPL1, AKR1D1, ALDOB, ALG8, ALG9, ALPL, AMER1, ANKH, ANKS6, ANLN, ANO5, ANOS1, AP2S1, ARHGAP31, ARHGDIA, ARL13B, ARL3, ARL6, ARMC5, ARMC9, ARSL, ATP6V0A4, ATP6V1B1, ATP8B1, ATR, AVPR2, B9D1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP1, BMP4, BMPR1B, BNC2, BSND, C2CD3, CA2, CABP4, CANT1, CASP10, CASR, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CCDC65, CCN6, CCNO, CD2AP, CDKN1C, CENPF, CENPJ, CEP120, CEP152, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP418, CFAP53, CFTR, CHD1L, CHD7, CHRNA3, CHST3, CHSY1, CILK1, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, COL10A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A3, COMP, COQ2, COQ6, COQ8B, COQ9, CRB1, CRB2, CRELD1, CRTAP, CRX, CSPP1, CTNS, CTU2, CUBN, CUL3, CWC27, CYP7B1, DCDC2, DDR2, DDX59, DGKE, DGUOK, DHCR7, DICER1, DLL3, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB11, DNAL1, DRC1, DSTYK, DUSP6, DYM, DYNC2H1, DYNC2LI1, DZIP1L, EBP, EIF2AK3, EMP2, ENPP1, ESCO2, EVC, EVC2, EXT1, EXT2, EYA1, FAH, FAM20C, FAN1, FAS, FASLG, FAT4, FEZF1, FGF17, FGF23, FGF8, FGFR1, FGFR2, FKBP10, FLNB, FLRT3, FN1, FOXP1, FRAS1, FREM1, FREM2, FSHB, FXYD2, GANAB, GATA3, GDF1, GDF5, GFM1, GHR, GLA, GLI2, GLI3, GLIS2, GLIS3, GNA11, GNAS, GNPAT, GNRH1, GNRHR, GPC3, GPC6, GREB1L, GRIP1, GUCY2D, HAAO, HAMP, HESX1, HEXA, HFE, HNF1B, HNF4A, HOXA13, HOXD13, HPSE2, HS6ST1, HSD11B2, HSD3B7, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IHH, IL17RD, IMPDH1, INF2, INPP5E, INPPL1, INVS, IQCB1, ITGA3, ITGA8, JAG1, KANK2, KCNJ1, KCNJ10, KCNJ13, KCNJ5, KDM6A, KIAA0586, KIF14, KIF22, KIF7, KISS1, KISS1R, KLHL3, KMT2D, KYNU, LAGE3, LAMB2, LBR, LCA5, LCAT, LCT, LEP, LEPR, LHB, LHX3, LHX4, LIFR, LMF1, LMX1B, LRAT, LRIG2, LRP4, LRP5, LZTFL1, MAFB, MAGI2, MAPKBP1, MATN3, MCEE, MERTK, MESP2, MGP, MKKS, MKS1, MMAA, MMAB, MMADHC, MMP13, MMP21, MMP9, MMUT, MPV17, MUC1, MYH9, MYO1E, MYO5B, MYO7A, MYOCD, NADSYN1, NBAS, NEK1, NEK8, NEUROG3, NIPBL, NKX2-5, NKX3-2, NME8, NMNAT1, NODAL, NOG, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR0B2, NR1H4, NR3C2, NSDHL, NSMF, NUP107, NUP93, OBSL1, OCRL, OFD1, OSGEP, OTX2, P3H1, PAPSS2, PAX2, PBX1, PCSK1, PDE4D, PDE6D, PDSS2, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, PEX7, PHEX, PHF6, PIBF1, PKD1L1, PKD2, PKHD1, PLCE1, PLOD2, PMM2, PNPLA6, POLG, POLR3B, POMC, POU1F1, PPARG, PPIB, PRKAR1A, PRKCSH, PROK2, PROKR2, PROM1, PROP1, PRPH2, PTH1R, PTHLH, PTPRO, RBBP8, RD3, RDH12, RDH5, REN, RET, RHO, RLBP1, RMND1, RNF216, ROBO1, ROBO2, ROR2, RPE65, RPGRIP1, RPGRIP1L, RRM2B, RSPH1, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3A, SERPINA1, SERPINF1, SERPINH1, SGPL1, SH3PXD2B, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC4A1, SLC4A4, SLCO1B1, SLCO1B3, SLIT2, SMARCAL1, SMPD1, SOX10, SOX11, SOX17, SOX2, SOX3, SOX9, SPAG1, SPATA7, SPINT2, SPRY4, STRA6, SUFU, TAC3, TACR3, TBC1D1, TBX15, TBX18, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TFR2, TJP2, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRIP11, TRMU, TRPC6, TRPS1, TRPV4, TSC1, TSC2, TTC21B, TTC37, TTC8, TULP1, TXNDC15, UGT1A1, UMOD, UPK3A, VHL, VIPAS39, VPS33B, WDR11, WDR19, WDR35, WDR4, WDR73, WNK1, WNK4, WNT4, WNT5A, WNT7A, WT1, XPNPEP3, XYLT1, ZIC3, ZMYND10, ZNF423

Gen med

Laboratorio de diagnóstico clínico y genómico

Endocrinología

Panel de hiperplasia suprarrenal congénita

Diseñado para pacientes con sospecha de este diagnóstico. Incluye el análisis del gen CYP21A2, que codifica para la enzima 21-hidroxilasa. Más del 90% de los casos son causados por una deficiencia de esta enzima.

Incluye 12 genes: AARMC5, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, PDE11A, PDE8B, POR, PRKAR1A, STAR

MLPA: CYP21A2

Panel de pancreatitis

Incluye genes asociados con la pancreatitis crónica y, para el diagnóstico diferencial, incluye genes asociados con el cáncer de páncreas.

Incluye 29 genes: ABCB4, APC, APOA5, APOC2, ATM, BMPR1A, BRCA1, BRCA2, CASR, CDKN2A, CFTR, CPA1, CTRC, EPCAM, GPIHBP1, LPL, MEN1, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53, UBR1, VHL.

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

Gen med

Laboratorio de diagnóstico clínico y genómico

Bioquímica

CentoLSD

Análisis completo del panel de enzimas para enfermedades relacionadas. Enzimas: lipasa ácida, alfa-glucosidasa, alfa-fucosidasa, alfa-galactosidasa, alfa-L-iduronidasa, alfa-manosidasa, alfa-N-acetilgalactosaminidasa, arilsulfatasa B, beta-galactosidasa, beta-glucocerebrosidasa, beta-glucuronidasa, beta-hexosaminidasa, Beta-manosidasa, Hexosaminidasa AB, Iduronato-2-sulfatasa, N-acetil-alfa-glucosaminidasa, N-acetilgalatosamina-6-sulfato-sulfatasa, Palmitoil-proteína tioesterasa, Tripeptidil peptidasa

CentoMPS

Análisis completo del panel de enzimas para enfermedades relacionadas Enzima: Alfa-L-iduronidasa, Iduronato-2-sulfatasa, N-acetil-alfa-glucosaminidasa, N-acetilgalatosamina-6-sulfato-sulfatasa, Beta-galactosidasa, Arilsulfatasa B, Beta-glucuronidasa, Alfa-manosidasa.

CentoNCL

Análisis completo del panel de enzimas para enfermedades relacionadas Enzimas: palmitoil-proteína tioesterasa, tripeptidil peptidasa.

CentoEsfingo

Análisis completo del panel de enzimas para enfermedades relacionadas Enzimas: beta-glucocerebrosidasa, alfa-galactosidasa, alfa-glucosidasa, beta-hexosaminidasa, hexosaminidasa AB, alfa-N-acetilgalactosaminidasa, lipasa ácida, alfa-manosidasa, beta-manosidasa, alfa-fucosidasa.

Gen med

Laboratorio de diagnóstico clínico y genómico

Biomarcadores y enzimas

DDC - DOPA descarboxilasa
ARSB - Arisulfatasa B
CHIT1 - Quitotriosidasa
FUCA1 - Alfa-L-fucosidasa 1
GALNS - N-acetilgalactosamina-6-sulfatasa
GBA - Glucocerebrosidasa
GLA - Alfa-galactosidasa A
GLB1 - Beta-galactosidasa
GUSB - Beta-glucoronidasa
HEXA - Beta-hexosaminidasa A
HEXB - Beta-hexosaminidasa B
IDUA - Alfa-L-iduronidasa
MAN2B1 - Alfa-manosidasa.
NAGA - Alfa-N-acetilgalactosaminidasa
NPC1 - Transportador intracelular de colesterol 1
NPC2 - Transportador intracelular de colesterol 2
PPT1 - Palmitoil-proteína tioesterasa 1
SMPD1 - Esfingomielinasa
GAA - Alfa glucosidasa ácida
IDS - Iduronato-2-sulfato sulfatasa
LIPA - Lipasa ácida lisosomal
MANBA - Beta-manosidasa
NAGLU - Alfa-N-acetilglucosaminidasa
SERPING1 - Inhibidor de C1
TPP1 - Tripeptidil peptidasa 1