PANELES PARA ESTUDIOS GENÉTICOS

PARA BUSCAR EL GEN QUE NECESITA,
PRESIONE CONTROL+F.

Labmedicina
ANÁLISIS CLÍNICOS

Ante cualquier duda o consulta, puede escribirnos a:
estudiosgeneticos@labmedicina.com

Exoma (25.000 genes): Incluye los exones de todos los genes humanos.

Panel personalizado: Secuenciación de genes de interés acorde al caso clínico.

EXÁMENES PARA CÁNCER HEREDITARIO

Panel de Cáncer Hereditario - Principales Genes (37 genes):

APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1

Panel de Cáncer Hereditario Completo (206 genes):

ACD AIP AKT1 ALK APC ASCL1 ATM ATR AXIN2 BAP1 BARD1 BDNF BLM BMPR1A BRCA1 BRCA2 BRIP1BUB1B CASP10 CASP9 CBL CDC73 C DH1 CDH23 CDK4 CDKN1B CDKN1C CDKN2A CEBPA CEP57 CHEK2CREBBP CSF3R CTC1 CTNNB1 CYLD DDB2 DICER1 DIS3L2 DKC1 DNAJ C21 DNMT3B DOCK8 EDN3 EPCAMERCC2 ERCC3 ERCC4 ERCC5 ERCC6 ERCC8 EXT1 EXT2 EZH2 FAN1 FANCA FANCB FANCC FANCD2F ANCE FANCF FANCG FANCI FANCL FANCM FAS FASLG FH FLCN G6PC GATA1 GATA2 GDNF GLMNGNAS GPC3 GREM1 GTF2E2 H19 HNF 1A HOXB13 HRAS IPMK JAG1 JAK2 KIF1B KIT KLLN KRAS LIG4LZTR1 MAD2L2 MAGT1 MAP2K1 MAP2K2 MAP3K1 MAX MEN1 MET MITF MLH1 MLH3 MMP1 MNX1 MRE11AMSH2 MSH3 MSH6 MSR1 MTAP MUTYH NBN NF1 NF2 NHP2 NME1 NOP10 NRAS NSD1 NTHL1 NTRK1PALB2 PA RN PDGFB PDGFRA PDGFRB PHOX2B PIK3CA PMS2 POLD1 POLE POLH POT1 PRF1 PRKAR1APSMC3IP PTCH1 PTCH2 PTEN PTPN11 RAD50 RAD51 RAD51C RAD51D RAD54L RAF1 RASA2 RASAL1 RB1RECQL RECQL4 RET RHBDF2 RNASEL RNF139 RRAS RSPO1 RTEL1 RUNX1 SAM D9 SAMD9L SBDS SDHASDHAF2 SDHB SDHC SDHD SEC23B SERPINA1 SETBP1 SH2D1A SHOC2 SLX4 SMAD4 SMARCA4 SMARCB1SMAR CE1 SOS1 SPRTN STAT3 STK11 SUFU TERC TERT TGFBR2 TINF2 TMC6 TMC8 TMEM127 TP53 TRIP13TSC1 TSC2 UBE2T VHL WAS WIPF1 W NT10A WRAP53 WRN WT1 XIAP XPA XPC XRCC2 ZNF687

Panel de Cáncer Colorrectal Hereditario (37 genes):

APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1

Panel de Cáncer de Mama y Ovario Hereditarios (37 genes):

APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1

Panel de Melanoma (37 genes):

APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1

Panel de Neoplasias Endocrinas Múltiples (37 genes):

APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1

PANELES PARA ENFERMEDADES RARAS

Panel de Adrenoleucodistrofia (1 gen): ABCD1

Panel Amiloidosis Familiar (1 gen): TTR

Panel de Anemia de Fanconi (18 genes):

BRCA1 BRCA2 BRIP1 ERCC4 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM PALB2 RAD51 RAD51C SLX4

Panel de Anemias Hereditárias (6 genes): G6PD HBA1 HBA2 HBB SBDS SLC19A2

Panel de Arritmias Hereditarias (35 genes):

ABCC9 ACTN2 ANK2 CACNA1C CACNB2 CALM1 CALM3 CASQ2 CAV3 DES DSC2 DSG2 DSP EMD GPD1L HCN4 JUP KCNE1 KCNE2 KCNH2 KCNJ2 KCNQ1 LMNA NKX2-5 PKP2 PLN PRKAG2 RBM20 RYR2 SCN5A TGFB3 TMEM43 TNNI3 TNNT2 TRDN

Panel de Ataxias (67 genes):

ABHD12 ACO2 ADCK3 AFG3L2 ANO10 APOB APTX ATCAY ATM ATP8A2 BEAN1 C10ORF2 CACNA1A CACNA1G CACNB4 CCDC88C CLCN2 CLN5 COQ2 CYP27A1 DNMT1 ELOVL4 FGF14 FLVCR1 FXN GOSR2 GRM1 ITPR1 KCNA1 KCNC3 KCND3 KCNJ10 LAMA1 MRE11A MTTP NOP56 PDSS1 PDSS2 PDYN PEX7 PHYH PMPCA PNKP PNPLA6 POLG PRKCG PTF1A RUBCN SACS SCN2A SETX SIL1 SLC1A3 SPTBN2 SYNE1 SYT14 TDP1 TGM6 TPP1 TTBK2 TTPA TXN2 VLDLR VLDLR WFS1 WWOX ZNF592

Panel de Baja Estatura (67 genes):

ACAN ADAMTS10 ANKRD11 ATR BRAF CBL CCDC8 CDC6 CDT1 CENPJ CEP152 CEP63 CHD7 COL10A1 COL2A1 COL9A1 COL9A2 COMP CREBBP CUL7 FBN1 FGF8 FGFR1 FGFR3 GH1 GHR GHRHR GLI2 GLI3 GNAS HESX1 HRAS IGF1 IGF2 IGF1R IGFALS IHH KRAS LHX3

LHX4 MAP2K1 NPPC NPR2 NRAS OBSL1 ORC1 ORC4 ORC6 OTX2 PAPSS2 PCNT PITX2 POU1F1 PRKAR1A PROP1 PTH1R PTPN11 RAF1 RBBP8 SHOC2 SHOX SOS1 SOX3 SOX9 SRCAP STAT5B XRCC4

Panel CADASIL (1 gen): NOTCH3.

Panel de Coagulopatías Hemorrágicas (10 genes):

F8 F9 FGA GGCX GP6 ITGA2B ITGB3 MPL P2RY12 VWF

Panel de Colestasis Crónica (6 genes): ATP8B1 ABCB11 ABCB4 CFTR SERPINA1 TJP2

Panel de Craneosinostosis (4 genes): FGFR1 FGFR2 FGFR3 TWIST1

Panel de Demencias y Parkinson (62 genes):

ABCA7 ABCD1 APP ARSA ATP13A2 ATP1A3 ATP7B CHMP2B CSF1R CYP27A1 DCTN1 DNAJC6 EIF4G1 FBXO7 FUS GALC GBA GCH1 GFAP GIGYF2 GLA GRN HEXA HTRA2 ITM2B LMNB1 LRRK2 MAPT NOTCH3 NPC1 NPC2 PANK2 PARK2 PARK7 PINK1 PLA2G6 PNKD POLG PPT1 PRKRA PRNP PRRT2 PSAP PSEN1 PSEN2 SGCE SLC2A1 SLC6A3 SNCA SPG11 SPR SQSTM1 TARDBP TH THAP1 TOR1A TREM2 TYROBP UBQLN2 UCHL1 VCP VPS35

Panel de Diabetes Monogénica MODY (24 genes):

ABCC8 BLK CEL EIF2AK3 GATA6 GCK HNF1A HNF1B HNF4A INS KCNJ11 KLF11 NEUROD1 NEUROG3 PAX4 PDX1 PLAGL1 PTF1A RFX6 SH2B1 SLC19A2 SLC2A2 WFS1 ZFP57

Panel de Dislipidemias (4 genes): APOB LDLRAP1 LDLR PCSK9 2/4

Panel de Distonías (37 genes):

ADCY5 ANO3 ARSA ATM ATP1A3 ATP7B CACNA1B CIZ1 COL6A3 CP DRD2 GCDH GCH1 GNAL KCNMA1 KCTD17 MR1 MRE11A PANK2 PARK2 PCNA PLA2G6 PNKD PRKRA PRRT2 RELN SGCE SLC2A1 SLC6A3 SPR TAF1 TH THAP1 TIMM8A TOR1A TUBB4A WDR45

Panel de Distrofia Muscular de Duchenne (1 gen): DMD

Panel de Distrofias Musculares, Miopatías y Miastenia (72 genes):

ACTA1 AGRN ANO5 B4GAT1 BAG3 BIN1 CAPN3 CAV3 CFL2 CHAT CHKB CHRNA1 CHRNB1 CHRND CHRNE CNTN1 COL6A1 COL6A2 COL6A3 COLQ CRYAB DAG1 DES DMD DNAJB6 DNM2 DOK7 DPAGT1 DPM1 DPM3 DYSF EMD FHL1 FKRP FKTN FLNC GAA GFPT1 GNE IGHMBP2 ISPD ITGA7 KBTBD13 LAMA2 LARGE LDB3 LMNA MATR3 MTM1 MUSK MYH7 MYL2 MYOT NEB PLEC PNPLA2 POMGNT1 POMT1 POMT2 RAPSN RYR1 SEPN1 SGCA SGCB SGCD SGCG TCAP TNNT1 TPM3 TRIM32 TTN VCP

Panel de Disturbios de la Función Renal (19 genes):

AQP2 ATP6V0A4 ATP6V1B1 AVPR2 BSND CLCNKA CLCNKB CLDN16 CLDN19 CNNM2 CTNS GLA KCNJ1 SCNN1B SCNN1G SLC12A1 SLC12A3 SLC4A4 TRPM6

Panel de Enfermedad Poliquística Renal (6 genes):

DZIP1L GANAB PKD1 PKD2 PKHD1 NOTCH2

Panel de Enfermedades Esqueléticas (150 genes):

ACAN ACP5 ADAMTSL2 AGA AGPS ALPL ALX4 AMER1 ANKH ARSB ARSE ATP6V0A2 B4GALT7 BMP1 BMPER BMPR1B CA2 CANT1 CASR CDC6 CDT1 CICN5 CLCN7 COG1 COL10A1 COL11A1 COL11A2 COL1A1 COL1A2 COL2A1 COL9A1 COL9A2 COL9A3 COMP CRTAP CTSA CTSK CUL7 DDR2 DHCR24 DLL3 DLX3 DMP1 DYM DYNC2H1 EBP EIF2AK3 ENPP1 EVC EVC2 FAM20C FBN1 FERMT3 FGF23 FGFR2 FGFR3 FIG4 FKBP10 GALNS GDF5 GDF6 GJA1 GLB1 GNPTAB GNPTG GNS GORAB GPC6 GUSB HES7 HGSNAT HPGD HSPG2 IDS IDUA IFITM5 IFT122 IFT140 IFT80 IHH IKBKG KIF22 LBR LEMD3 LFNG LIFR LRP5 MAN2B1 MATN3 MESP2 MMP13 MMP9 MNX1 MSX2 NAGLU NEK1 NEU1 NKX3-2 NPR2 NSDHL OBSL1 ORC1 ORC4 ORC6 OSTM1 P3H1 PCNT PDE4D PEX7 PHEX PLEKHM1 PLOD2 PPIB PRKAR1A PTH1R PYCR1 RASGRP2 RBBP8 ROR2 RUNX2 SBDS SERPINH1 SGSH SHOX SLC17A5 SLC34A3 SLC35D1 SLC39A13 SMARCAL1 SOST SOX9 SP7 SULF1 SUMF1 TBCE TBXAS1 TCIRG1 TGFB1 TNFRSF11A TNFRSF11B TNFSF11 TRAPPC2 TRIP11 TRPS1 TRPV4 TTC21B WDR19 WDR35 XRCC4 XYLT1

Panel de Enfermedades de Neurona Motor y Neuropatías Periféricas (82 genes):

AARS AIFM1 ALS2 ANG AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 ATL1 ATXN2 BSCL2 C12ORF65 CHMP2B CYP7B1 DNM2 DYNC1H1 EGR2 ERBB4 ERLIN2 FA2H FGD4 FIG4 FUS GARS GDAP1 GJB1 GJC2 HSPB1 HSPB8 HSPD1 IGHMBP2 KIAA0196 KIF1A KIF5A L1CAM LITAF LMNA LRSAM1 MATR3 MED25 MFN2 MPZ MTMR2 NDRG1 NEK1 NIPA1 OPTN PARK7 PFN1 PLP1 PMP22 PNPLA6 PRPS1 PRX RAB7A REEP1 RTN2 SACS SBF2 SCN10A SCN9A SETX SH3TC2 SIGMAR1 SLC33A1 SOD1 SPAST SPG11 SPG20 SPG21 SPG7 SQSTM1 TARDBP TBK1 TRPV4 TTR UBQLN2 VAPB VCP YARS ZFYVE26

Panel de Enfermedad de Wilson (1 gen): ATP7B

Panel de Ehlers-Danlos e Cutis Laxa (24 genes):

ADAMTS2 ALDH18A1 ATP6V0A2 ATP7A B3GALT6 B4GALT7 CHST14 COL1A1 COL1A2 COL3A1 COL5A1 COL5A2 EFEMP2 ELN FBLN5 FKBP14 FLNA GORAB LTBP4 PLOD1 RIN2 SLC2A10 SLC39A13 TNXB

Panel de Endocrinopatías Neonatales (25 genes):

ABCC8 CYP11B1 CYP17A1 CYP21A2 DUOXA2 GCK GLIS3 GLUD1 HADH INSR IYD KCNJ11 LHX4 NR0B1 PAX8 POU1F1 PROP1 SLC16A1 SLC2A2 SLC5A5 TG THRA THRB TPO TSHB

Panel de Epidermólisis Bullosa (17 genes):

CAST CD151 COL17A1 COL7A1 DSP DST FERMT1 ITGA3 ITGA6 ITGB4 KRT14 KRT5 LAMA3 LAMB3 LAMC2 PLEC TGM5

Panel de Epilepsia (96 genes):

ADSL ALDH7A1 ALG13 AMT ARHGEF9 ARX ATP13A2 ATP1A2 BRAF BRAT1 CASK CDKL5

CES1 CHD2 CHRNA2 CHRNA4 CHRNB2 CLN3 CLN5 CLN6 CLN8 CNTNAP2 CSTB CTSD DNAJC5 DNM1 DYNC1H1 EFHC1 EPM2A FOLR1 FOXG1 GABRA1 GABRB3 GABRG2 GAMT GATM GBA GLDC GOSR2 GRIN1 GRIN2A GRIN2B GRN HCN1 HNRNPU KANSL1 KCNA1 KCNJ10 KCNQ2 KCNQ3 KCNT1 KCTD7 LGI1 LIAS MBD5 MECP2 MEF2C MFSD8 MOCS1 MOCS2 NHLRC1 NRXN1 PCDH19 PLCB1 PNKP PNPO POLG PPT1 PRICKLE1 PRRT2 RBFOX1 ROGDI SCARB2 SCN1A SCN1B SCN2A SCN8A SCN9A SLC25A22 SLC2A1 SLC6A8 SLC9A6 SMS SPTAN1 STXBP1 SUOX SYN1 SYNGAP1 TBC1D24 TCF4 TPP1 TSC1 TSC2 UBE3A WWOX ZEB2

Panel de Errores Innatos do Metabolismo Tratables (124 genes):

ABCC8 ABCD1 ACADM ACADVL ACAT1 AGL ALDH7A1 ALDOB ARG1 ARSA ARSB ASAH1 ASL ASS1 ATP7A ATP7B AUH BCKDHA BCKDHB BCKDK BTD CBS COQ2 COQ9 CPS1 CPT1A CPT2 CTNS CYB5A CYB5R3 CYP11B1 CYP17A1 CYP21A2 CYP27A1 DBT DLD ETFA ETFB ETFDH ETHE1 FAH FBP1 FOLR1 G6PC G6PD GAA GALE GALK1 GALT GAMT GATM GBA GBE1 GCDH GCK GLA GLB1 GLUD1 GUSB GYS2 HADH HADHA HADHB HLCS HMGCL HMGCS2 HPD IDS IDUA INSR IVD KCNJ11 LIPA LMBRD1 MMAA MMAB MMACHC MMADHC MOCS1 MPI MTHFR MTR MTRR MUT NAGLU NAGS NPC1 NPC2 OTC OXCT1 PAH PCBD1 PCCA PCCB PDSS1 PDSS2 PGM1 PHGDH PHKA2 PSAT1 PSPH PTS PYGL QDPR SGSH SI SLC16A1 SLC19A2 SLC19A3 SLC22A5 SLC25A13 SLC25A15 SLC25A20 SLC2A1 SLC2A2 SLC37A4 SLC46A1 SLC52A2 SLC52A3 SLC7A9 SMPD1 TAT TCN2 TPP1

Panel de Esclerosis Tuberosa (2 genes): TSC1 TSC2

Panel de Fibrosis Quística (1 gen): CFTR

Panel de Hemocromatosis (5 genes): HFE HFE2 HAMP SLC40A1 TFR2

Panel de Ictiosis (28 genes):

ABCA12 ABHD5 ALDH3A2 ALOX12B ALOXE3 ALOXE3 AP1S1 CLDN1 CSTA CYP4F22 ELOVL4 ERCC2 FLG GJB2 KRT1 KRT10 KRT2 LIPN LOR NIPAL4 PNPLA1 PNPLA1 POMP SLC27A4 SNAP29 ST14 STS TGM1

Panel de Inmunodeficiencias Primarias (60 genes):

ADA AICDA BLNK BTK CD247 CD3D CD3E CD3G CD40 CD40LG CD79A CD79B CIITA CYBA CYBB DCLRE1C ELANE FOXN1 FOXP3 G6PC3 GATA2 GFI1 HAX1 IFNGR1 IFNGR2 IGLL1 IL12RB1 IL2RG IL7R JAK3 LRRC8A MAGT1 MPO MYD88 NCF1 NCF2 NCF4 NHEJ1 ORAI1 PNP PRF1 PTPRC RAC2 RAG1 RAG2 RFX5 RFXANK RFXAP SH2D1A STAT1 STX11 STXBP2 TAP1 TAP2 TAPBP UNC13D UNG WAS WIPF1 XIAP

Panel Expandido de Inmunodeficiencias, autoinflamatorias y enfermedad inflamatoria intestinal (438 genes):

A2ML1, ABCD4, ACD, ACP5, ADA, ADAM17, ADNP, AGA, AICDA, AIRE, AK2, ALG1, ALG12, AP1S3, AP3B1, ARMC4, ARPC1B, ATG16L1, ATM, B2M, BCL10, BCL11B, BLM, BLNK, BRCA1,

BRCA2, BRIP1, BTK, BUB1B, C11orf70, C1QA, C1QB, C1QC, C1R, C1S, C2, C21orf59, C3, C5, C6, C7, C8A, C8B, C8orf37, CARD11, CARD14, CARD9, CASP10, CASP8, CCBE1, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDCA7, CDSN, CDX1, CEBPE, CECR1, CFB, CFD, CFH, CFI, CFP, CHAMP1, CHD1, CHD7, CIITA, CLEC7A, CLPB, COG6, COG7, CORO1A, CR2, CREBBP, CRIPT, CSF3R, CTC1, CTLA4, CTPS1, CXCR4, CYBA, CYBB, DCLRE1C, DEAF1, DEFB1, DHFR, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE1L3, DNMT3B, DOCK2, DOCK8, DRC1, DSG1, DYX1C1, ECR1, EDA, EFL1, EGFR, ELANE, EPG5, ERCC2, ERCC4, ERCC6L2ETV6, EXTL3, F12, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FBXL4, FCGR2C, FCGR3A, FCN3, FERMT3, FMO3, FOXN1, FOXP3, FUT2, G6PC3, GALNS, GAS8, GATA1, GATA2, GATA3, GFI1, GSS, GTF2H5, GUCY2C, HAX1, HELLS, HGSNAT, HTR1A, HYDIN, ICOS, IFIH1, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKAP, IKBKB, IKBKG, IKZF1, IKZF2, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL15, IL15RA, IL17A, IL17F, IL17RA, IL17RC, IL1RL1, IL1RN, IL21, IL21R, IL23A, IL23R, IL2RA, IL2RG, IL33, IL36RN, IL7R, IRAK1, IRAK4, IRF2BP2, IRF7, IRF8, IRGM, ISG15, ITCH, ITGB2, ITK, IVD, JAGN1, JAK3, KMT2D, KRAS, LAMTOR2, LAT, LCK, LEP, LIG4, LPIN2, LRBA, LRRC6, LRRC8A, LYST, MAD2L2, MAGT1, MALT1, MAN2B1, MANBA, MC2R, MCM4, MEFV, MGP, MMAA, MMAB, MMACHC, MOGS, MPL, MPO, MS4A1, MSN, MTHFD1, MUT, MVK, MYD88, MYO5B, NBN, NCF1, NCF2, NCF4, NCSTN, NFASC, NFE2L2, NFKB1, NFKB2, NFKBIA, NGF, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NLRP7, NME8, NOD2, NOP10, NRAS, NSMCE3, ORAI1, OXCT1, PALB2, PARN, PCCA, PCCB, PEPD, PGM3, PIH1D3, PIK3CD, PIK3R1, PLCG2, PMM2, PN1, PNP, POLA1, POLE, PPP1R21, PRDM1, PRF1, PRKCD, PRKDC, PSENEN, PSMB8, PSTPIP1, PTEN, PTPRC, PTRF, RAB27A, RAC2, RAD50, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RBCK1, RBM8A, RELB, RFWD3, RFX5, RFXANK, RFXAP, RMRP, RNF113A, RNF168, RNF186, RNF31, RORC, RPL11, RPL15, RPL18, RPL26, RPL35A, RPL5, RPS10, RPS17,RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SCNN1B, SCNN1G, SDCCAG8, SEMA3E, SERAC1, SERPING1, SGPL1, SH2D1A, SH3BP2, SKIV2L, SLC29A3, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4 , SLC39A8, SLC46A1, SLK, ,SLX4, SMARCAL1, SMARCD2, SNAI2, SP110, SPAG1, SPATA5, SPINK5, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STN1, STX11, STXBP2, TALDO1, TAP1, TAP2, TAPBP, TAZ, TBCE, TBX1, TBXAS1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, TGFB2, TGFB3, TINF2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TPI1, TRAC, TRAF3IP2, TRNT1, TRPS1, TSR2, TTC25, TTC37, TTC7A, TYK2, UBE2T, UMPS, UNC119, UNC13D, UNG, USB1, VIPAS39, VPS13B, VPS33B, VPS45, WAS, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZMYND10

Panel de Leucodistrofias (136 genes):

AARS2 ABCD1 ACOX1 ADAR ADCK3 AIMP1 ALDH3A2 ARSA ASPA ATP7A ATP7B ATPAF2 BCAP31 BCS1L C10ORF2 CLCN2 COL4A1 COQ2 COQ9 COX10 COX15 CSF1R CYP27A1 CYP2U1 CYP7B1 D2HGDH DARS DARS2 DGUOK EARS2 EIF2B1 EIF2B2 EIF2B3 EIF2B4 EIF2B5 ERCC2 ERCC3 ERCC6 ERCC8 ETFDH FA2H FAM126A FUCA1 GALC GBE1 GFAP GFM1 GJA1 GJC2 GLA GLB1 GTF2H5 HEPACAM HEXA HSD17B4 HSPD1 HSPD1 HTRA1 L2HGDH LMNB1 MCOLN1 MLC1 MPLKIP MRPS16 NDUFAF1 NDUFS1 NDUFS2 NDUFS4 NDUFS7

NDUFS8 NDUFV1 NOTCH3 NPC1 NPC2 OCLN OCRL PEX1 PEX5 PEX2 PEX3 PEX6 PEX7 PEX10 PEX11B PEX12 PEX13 PEX14 PEX16 PEX19 PEX26 PHGDH PHYH PLP1 POLG POLG2 POLR3A POLR3B PPT1 PRF1 PSAP PSAT1 RNASEH2A RNASEH2B RNASEH2C RNASET2 RRM2B SAMHD1 SCO1 SCP2 SDHA SDHAF1 SDHB SLC16A2 SLC17A5 SLC25A1 SLC25A12 SLC25A4 SOX10 SPAST SPG11 SPG20 SPG21 SPG7 STX11 STXBP2 SUCLA2 SUMF1 SURF1 TACO1 TREX1 TUBB4A TUFM TYMP TYROBP UNC13D ZFYVE26

Panel de Miocardiopatías Hereditarias (49 genes):

ABCC9 ACTC1 ACTN2 ALMS1 BAG3 CAV3 CRYAB CSRP3 DES DMD DSC2 DSG2 DSP ELAC2 EMD EYA4 FHL1 FKRP FKTN GLA HCN4 JUP LAMP2 LMNA MTO1 MYBPC3 MYH7 MYL2 MYL3 PKP2 PLN PRKAG2 RAF1 RBM20 RYR2 SCN5A SDHA SGCD TAZ TCAP TGFB3 TMEM43 TNNC1 TNNI3 TNNT2 TPM1 TTN TTR VCL

Panel de Neurofibromatosis (3 genes): NF1 NF2 SPRED1

Panel de Retinopatías (224 genes):

ABCA4 ABCC6 ABCD1 ABHD12 ACO2 ADAM9 ADGRV1 AHI1 AIPL1 ALMS1 AMACR ARL13B ARL6 ATF6 B9D1 B9D2 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 BEST1 C12ORF65 C1QTNF5 C2ORF71 C5ORF42 C8ORF37 CA4 CABP4 CACNA1F CACNA2D4 CASK CC2D2A CDH23 CDH3 CDHR1 CEP290 CEP41 CERKL CFH CHM CIB2 CISD2 CLN3 CLN5 CLN6 CLN8 CLRN1 CNGA1 CNGB1 CNGB3 CNNM4 CRB1 CRX CTSD CYP4V2 DFNB31 DHDDS DNAJC5 EFEMP1 ELOVL4 EYS FAM161A FLVCR1 FRMD7 FSCN2 FZD4 GDF6 GJB2 GJB6 GNAT1 GNAT2 GNPTG GPR143 GPR179 GRK1 GRM6 GRN GUCA1A GUCA1B GUCY2D HARS HGSNAT HK1 HMCN1 HMX1 IDH3B IFT140 IMPDH1 IMPG2 IQCB1 ITM2B KCNJ13 KCNV2 KCTD7 KIF7 KLHL7 LAMA1 LCA5 LRAT LRP5 LZTFL1 MAK MERTK MFN2 MFRP MFSD8 MKKS MKS1 MMACHC MVK MYO7A NDP NEUROD1 NMNAT1 NPHP1 NPHP3 NPHP4 NR2F1 NRL NYX OAT OFD1 OPA1 OPA3 OPN1LW OPN1MW OTX2 PAX6 PCDH15 PDE6A PDE6B PDE6C PDE6G PDE6H PDZD7 PEX1 PEX10 PEX11B PEX12 PEX13 PEX14 PEX16 PEX19 PEX2 PEX26 PEX3 PEX5 PEX6 PEX7 PHYH PITPNM3 PNPLA6 PPT1 PRCD PROM1 PRPF3 PRPF31 PRPF6 PRPF8 PRPH2 PRPS1 RAB28 RAX2 RBP3 RBP4 RD3 RDH12 RDH5 RGR RGS9 RGS9BP RHO RIMS1 RLBP1 ROM1 RP1 RP1L1 RP2 RP9 RPE65 RPGR RPGRIP1 RPGRIP1L RS1 SAG SDCCAG8 SEMA4A SLC24A1 SNRNP200 SPATA7 TCTN1 TCTN2 TEAD1 TIMM8A TIMP3 TMEM126A TMEM138 TMEM216 TMEM237 TMEM67 TOPORS TPP1 TRIM32 TRPM1 TSPAN12 TTC21B TTC8 TUBGCP4 TUBGCP6 TULP1 TYR UNC119 USH1C USH1G USH2A VPS13B WDPCP WDR19 WFS1 ZNF513

Panel de Síndrome de Noonan y Rasopatias (15 genes):

BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1 RIT1 SHOC2 SOS1 SPRED1

Panel de Síndrome Urémico Hemolítico (6 genes): C3 CD46 CFB CFH CFI THBD

Panel de Síndrome Nefrótico (29 genes):

ACTN4 APOL1 ARHGAP24 CD2AP COL4A3 COL4A4 COL4A5 COQ2 COQ6 FN1 IFIH1

INF2 ITGA3 LAMB2 LMX1B MYH9 MYO1E NPHS1 NPHS2 PAX2 PDSS2 PLCE1 PMM2 PTPRO SCARB2 SLC17A5 SMARCAL1 TRPC6 WT1

Panel de Síndromes Clínicamente Reconocibles (133 genes):

ALMS1 ANKRD11 ARHGAP31 ARID1B ARL6 ATR BANF1 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 BLM BRAF CBL CDC6 CDT1 CENPJ CEP152 CEP290 CEP63 CREBBP DHCR7 DOCK6 EDN3 EDNRB ELP4 EP300 ERCC8 EYA1 EZH2 FBN2 FGD1 FOXL2 GATA3 GCM2 GLE1 GNAS GPC3 GPR143 HDAC8 HRAS IRF6 JAG1 KDM6A KIF1BP KMT2A KMT2D KRAS LMNA LMX1B LZTFL1 MAP2K1 MAP2K2 MID1 MITF MKKS MKS1 MYBPC1 MYH3 MYH8 NF1 NFIX NIPBL NOTCH1 NOTCH2 NRAS NRXN1 NSD1 OCA2 OFD1 ORC1 ORC4 ORC6 PAX3 PAX6 PHF6 PIEZO2 POLR1C POLR1D PTH PTPN11 RAD21 RAF1 RASA1 RBBP8 RECQL4 RIT1 ROR2 RPS6KA3 SALL1 SDCCAG8 SHOC2 SIX5 SKI SLC45A2 SMAD3 SMC1A SMC3 SNAI2 SOS1 SOX10 SPECC1L SPRED1 SRCAP STX16 TBCE TBX5 TCF4 TCOF1 TFAP2A TGFB2 TGFB3 TGFBR1 TGFBR2 TMEM67 TNNI2 TNNT3 TP63 TRIM32 TTC8 TYR TYRP1 VIPAS39 VPS13B VPS33B WDPCP WNT5A WRN ZEB2

Panel de Sordera - Principales Genes (18 genes):

ATP6V0A4 ATP6V1B1 CLDN14 DFNB59 GJB2 GJB3 GJB6 HGF ILDR1 LHFPL5 MARVELD2 MYO15A OTOF SLC26A4 TIMM8A TMIE TMPRSS3 TRIOBP

Panel de Sordera Expandido (114 genes):

ACTB ACTG1 ADGRV1 ATP6V1B1 BCS1L BSND CATSPER2 CCDC50 CDH23 CEACAM16 CEMIP CLDN14 CLRN1 COCH COL11A2 COL9A2 COL9A3 CRYM DFNA5 DFNB31 DFNB59 DIAPH1 DSPP ECE1 EDNRA EDNRB ERCC2 ERCC3 ESPN ESRRB EYA4 FAS FGF3 FGFR3 FOXI1 GATA3 GIPC3 GJA1 GJB1 GJB2 GJB3 GJB4 GJB6 GPSM2 GRHL2 GRXCR1 GSTP1 HAL HGF ILDR1 JAG1 KCNE1 KCNJ10 4/4 KCNQ1 KCNQ4 LHFPL5 LHX3 LOXHD1 LRTOMT MARVELD2 MITF MSRB3 MTAP MYH14 MYH9 MYO15A MYO1A MYO1C MYO1F MYO3A MYO6 MYO7A NDP NR2F1 OTOA OTOF OTOR PAX3 PCDH15 PDZD7 PMP22 POU3F4 POU4F3 PRPS1 PTPRQ RDX SERPINB6 SIX1 SIX5 SLC17A8 SLC26A4 SLC26A5 SLC4A11 SMPX SNAI2 SOX2 SPINK5 STRC TBL1X TCF21 TECTA TFCP2 TIMM8A TJP2 TMC1 TMIE TMPRSS3 TMPRSS5 TPRN TRIOBP USH1C USH1G USH2A WFS1

Panel de Trombofilias (7 genes): ADAMTS13 CBS F2 F5 PROC PROS1 SERPINC1

Ante cualquier duda o consulta, puede escribirnos a:

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