



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 Clínico:	Secuenciación de las regiones codificantes de más de 6700 genes.
Panel personalizado:	Secuenciación de genes de interés acorde al caso clínico.

PANELES PARA CÁNCER HEREDITARIO

Panel de Cáncer Hereditario Completo (NGS243):	AIP ALK APC ATM BAP1 BLM BMPR1A BRCA1 BRCA2 BRIP1 BUB1B CDC73 CDH1 CDK4 CDKN1C CDKN2A CEBPA CEP57 CHEK2 CYLD DDB2 DICER1 DIS3L2 EGFR EPCAM ERCC2 ERCC3 ERCC4 ERCC5 EXT1 EXT2 EZH2 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM FH FLCN GATA2 GPC3 HNF1A HRAS KIT MAX MEN1 MET MLH1 MSH2 MSH6 MUTYHNB NNF1 NF2 NSD1 PALB2 PHOX2B PMS1 PMS2 PRF1 PRKAR1A PTCH1 PTEN RAD51C RAD51D RB1 RECQL4 RET RHBDF2 RUNX1 SBDS SDHAF2 SDHB SDHC SDHD SLX4 SMAD4 SMARCB1 STK11 SUFU TMEM127 TP53 TSC1 TSC2 VHL WRN WT1 XPA XPC
Panel de Cáncer Hereditario de Mama y Ovario (NGS23):	ATM BARD1 BRCA1 BRCA2 BRIP1 CDH1 CHEK2 MLH1 MSH2 MSH6 NBN NF1 PALB2 PMS2 PTEN RAD51C RAD51D STK11 TP53 DICER1 FANCC FANCM MRE11A MUTYH RAD50 RECQL4 SMARCA4 XRCC2 RINT1
Panel de Cáncer de Mama (BRCA SEC):	BRCA1 BRCA2
Panel de Cáncer Colorrectal Hereditario (NGS265):	APC BMPR1A CHEK2 MLH1 MSH2 MSH6 MUTYH PMS2 PTEN SMAD4 STK11 TP53 MSH3 NTHL1 POLD1 POLE ATM BLM CDH1 GALNT12
Panel de Melanoma familiar (NGS270):	BAP1 BRCA2 CDK4 CDKN2A MITF PTEN RB1 TP53
Panel de Neoplasias Endocrinas Múltiples (NGS271):	MEN1
Panel de Esclerosis Tuberosa (NGS295):	TSC1 TSC2
Panel de Neurofibromatosis (NGS296):	NF1 NF2 LZTR1 SMARCB1 SPRED1
Panel de Retinoblastoma (NGS287):	RB1

PANELES PARA ENFERMEDADES RARAS

Panel de Adrenoleucodistrofia (NGS288):	ABCD1
Panel Amiloidosis Familiar (NGS289):	TTR
Panel de Anemia de Fanconi (NGS239):	BRCA1 BRCA2 BRIP1 ERCC4 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM PALB2 RAD51 RAD51C SLX4
Panel de Anemias Hereditarias (NGS237):	G6PD HBA1 HBA2 HBB SBDS SLC19A2
Panel de Arritmias Cardíacas (NGS240):	ABCC9 AKAP9 ANK2 CACNA1C CACNA2D1 CACNB2 CALM1 CALM2 CALM3 CASQ2 CAV3 DES DSC2 DSG2 DSP GJA5 GPD1L HCN4 JUP KCNA5 KCND3 KCNE1 KCNE2 KCNE3 KCNE5 KCNH2 KCNJ2 KCNJ5 KCNJ8 KCNK3 KCNQ1 LDB3 LMNA MYH6 NKX2-5 NPPA PKP2 PLN PRKAG2 RANGRF RYR2 SCN10A SCN1B SCN2B SCN3B SCN4B SCN5A SLMAP SNTA1 TGFB3 TMEM43 TNNI3 TNNI3K TRDN TRPM4 TTN
Panel de Ataxias Hereditarias (NGS241):	ABCB7 ABHD12 ACO2 AFG3L2 ANGPTL3 ANO10 APOB APTX ATCAY ATM ATP1A3 ATP2B3 ATP8A2 CA8 CACNA1A CACNA1G CACNB4 CASK CCDC88C CLCN2 CLN5 COQ8A CP CWF19L1 CYP27A1 DAB1 DARS2 DNMT1 EEF2 ELOVL4 ELOVL5 FAT2 FGF14 FLVCR1 FXN GOSR2 GRID2 GRM1 ITPR1 KCNA1 KCNC3 KCND3 KCNJ10 KIF1C LAMA1 MARS2 MME MRE11 MTPAP MTPP OPHN1 PCNA PDYN PEX7 PHYH PIK3R5 PLD3 PNKP PNPLA6 POLG POLR3A POLR3B PRKCG RNF216 RUBCN SACS SETX SIL1 SLC1A3 SLC25A46 SLC52A2 SLC9A1 SLC9A6 SNX14 SPTBN2 SQSTM1 STUB1 SYNE1 SYT14 TDP1 TDP2 TGM6 TMEM240 TPP1 TRPC3 TSFM TTBK2 TTPA TUBB4A TWNK TXN2 VAMP1 VLDLR VWA3B WDR73 WDR81 WFS1 WWOX
Panel de Baja Talla (NGS242):	ACAN ANKRD11 ATR ATRIP BLM BMPR1B BRAF BRF1 BTK CBL CCDC8 CDC6 CDKN1C CDT1 CENPJ CEP152 CEP63 CHD7 COL10A1 COL11A1 COL11A2 COL2A1 COL9A1 COL9A2 COL9A3 COMP CREBBP CRIPT CUL7 DHCR7 DNA2 DVL1 EP300 ERCC6 ERCC8 FBN1 FGD1 FGFR3 GDF5 GHR GHRHR HDAC8 HESX1 HRAS IGF1 IGF1R IGF2 IGFALS IHH INSR KDM6A KMT2D KRAS LARP7 LHX3 LIG4 LMNA MATN3 MCM9 NBN NIN NIPBL NPR2 NRAS OBSL1 ORC1 ORC4 ORC6 OTX2 PCNT PDE4D PIK3R1 PITX2 POC1A PRKAR1A PTHLH PTPN11 RAD21 RAF1 RBBP8 RECQL4 RIT1 RNU4ATAC ROR2 SHOC2 SHOX SMARCA1 SMC1A SMC3 SOS1 SOX2 SOX3 SOX9 SRCAP STAT5B TRIM37 WNT5A XRCC4

Panel CADASIL (NGS280):	NOTCH3.
Panel de Coagulopatías Hemorrágicas (NGS290):	F8 F9 FGA GGCX GP6 ITGA2B ITGB3 MPL P2RY12 VWF
Panel de Colestasis Crónica (NGS291):	ATP8B1 ABCB11 ABCB4 CFTR SERPINA1 TJP2
Panel de Craneosinostosis (NGS292):	FGFR1 FGFR2 FGFR3 TWIST1
Panel de Enfermedad de Parkinson (NGS244):	ADH1C ATP13A2 ATP1A3 DNAJC6 EIF4G1 FBXO7 GBA GCH1 GIGYF2 GLUD2 HTRA2 LRRK2 MAPT PARK7 PINK1 PLA2G6 PRKN PRKRA RAB39B SLC6A3 SNCA SPR SYNJ1 TAF1 UCHL1 VPS13A VPS35
Panel de Diabetes Monogénica MODY (NGS245):	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 (NGS293):	APOB LDLRAP1 LDLR PCSK9
Panel de Diskinesia y Distonías (NGS294):	ACTB ADCY5 ANO3 ATP1A3 CACNA1B COL6A3 GCH1 GNAL MR1 NKX2-1 PNKD PRKRA PRRT2 SGCE SLC2A1 SPR TAF1 TH THAP1 TOR1A TUBB4A VPS13A XK
Panel de Distrofia Muscular de Duchenne (NGS274):	DMD
Panel de Distrofias Musculares (NGS246):	ACTA1 ALG13 ANO5 B4GAT1 CAPN3 CAV3 CAVIN1 CHKB COL12A1 COL6A1 COL6A2 COL6A3 CRPPA DAG1 DES DMD DNM2 DPM1 DPM3 DYSF EMD FHL1 FKBP14 FKRP FKTN GAA ITGA7 LAMA2 LARGE1 LMNA MYOT PLEC POMGNT1 POMGNT2 POMT1 POMT2 SELENON SGCA SGCB SGCD SGCG SYNE1 SYNE2 TCAP TMEM43 TRIM32 TTN VCP
Panel de Disturbios de la Función Renal (NGS273):	AQP2 ATP6VOA4 ATP6V1B1 AVPR2 BSND CLCNKA CLCNKB CLDN16 CLDN19 CNNM2 CTNS GLA KCNJ1 SCNN1B SCNN1G SLC12A1 SLC12A3 SLC4A4 TRPM6
Panel de Enfermedad Poliquística Renal (NGS267):	BICC1 HNF1B LRP5 NOTCH2 PKD1 PKD2 PKHD1 PRKCSH SEC63 TSC1 TSC2 UMOD

Panel de Displasias Esqueléticas (NGS248):

ACAN ACP5 ACVR1 ADAMTS10 ADAMTS17 ADAMTSL2
AGA AGPS AKT1 ALPL ALX1 ALX3 ALX4 AMER1 ANKH
ANO5 ANTXR2 ARHGAP31 ARSB ARSL ATP6VOA2
B3GALT6 B4GALT7 BMP1 BMP2 BMPER BMPR1B BPNT2
CA2 CANT1 CASR CC2D2A CCDC8 CCN6 CCNQ CDC6
CDH3 CDKN1C CDT1 CEP290 CHST14 CHST3 CHSY1
CILK1 CKAP2L CLCN5 CLCN7 COG1 COL10A1 COL11A1
COL11A2 COL1A1 COL1A2 COL2A1 COL9A1 COL9A2
COL9A3 COMP CREB3L1 CREBBP CRTAP CTSA CTSK
CUL7 DDR2 DHCR24 DHODH DLL3 DLX3 DLX5 DLX6
DMP1 DOCK6 DVL1 DYM DYNC2H1 EBP EFN1 EFTUD2
EIF2AK3 ENPP1 EOGT EP300 ESCO2 EVC EVC2 EXT1
EXT2 EZH2 FAM111A FAM20C FBLN1 FBN1 FBN2 FBXW4
FERMT3 FGF10 FGF16 FGF23 FGF9 FGFR1 FGFR2 FGFR3
FIG4 FKBP10 FLNA FLNB FMN1 FN1 FUCA1 GALNS
GALNT3 GDF3 GDF5 GDF6 GJA1 GLB1 GLI3 GNAS GNPAT
GNPTAB GNPTG GNS GORAB GPC6 GPX4 GREM1 GUSB
HDAC4 HDAC8 HES7 HGSNAT HOXA11 HOXA13 HOXD13
HPGD HSPG2 IDH1 IDH2 IDS IDUA IFITM5 IFT122 IFT140
IFT172 IFT43 IFT80 IHH IL1RN INPPL1 KAT6B KIF22 KIF7
LBR LEMD3 LFNG LIFR LMBR1 LMNA LMX1B LONP1
LPIN2 LRP4 LRP5 LTBP2 MAFB MAN2B1 MANBA MATN3
MBTPS2 MEGF8 MEOX1 MESP2 MGP MKS1 MMP13 MMP2
MMP9 MNX1 MSX2 MYCN NAGLU NEK1 NEU1 NF1 NFIX
NIPBL NKX3-2 NLRP3 NOG NOTCH2 NPR2 NSD1 NSDHL
OBSL1 OFD1 ORC1 ORC4 ORC6 OSTM1 P3H1 PAM16
PAPSS2 PCNT PCYT1A PDE4D PEX5 PEX7 PHEX PIGV
PIK3CA PITX1 PLEKHM1 PLOD2 PLS3 POLR1C POLR1D
POP1 POR PPIB PRKAR1A PTDSS1 PTH1R PTHLH PTPN11
PYCR1 RAB23 RAB33B RAD21 RASGRP2 RBM8A RBPJ
RECQL4 RMRP ROR2 RPGRIP1L RUNX2 SALL1 SALL4
SBDS SEC24D SERPINF1 SERPINH1 SETD2 SF3B4 SGSH
SH3BP2 SH3PXD2B SHH SHOX SKI SLC17A5 SLC26A2
SLC29A3 SLC34A3 SLC35D1 SLC39A13 SLCO5A1 SMAD3
SMAD4 SMARCAL1 SMC1A SMC3 SNRPB SNX10 SOST
SOX9 SP7 SULF1 SUMF1 TBCE TBX15 TBX3 TBX4 TBX5
TBX6 TBXAS1 TCF12 TCIRG1 TCOF1 TCTN3 TGDS TGFB1
TGFB2 TGFB1R1 TGFB2R1 THPO TMEM38B TMEM67
TNFRSF11A TNFRSF11B TNFSF11 TP63 TRAPPC2 TREM2
TRIP11 TRPS1 TRPV4 TTC21B TWIST1 TYROBP WDR19
WDR34 WDR35 WNT1 WNT10B WNT3 WNT5A WNT7A
XYLT1 XYLT2 ZMPSTE24

<p>Panel de Enfermedades de Neurona Motor y Neuropatías Periféricas (NGS247):</p>	<p>AARS1 AIFM1 ALS2 ANG ANXA11 ARHGEF10 ASAH1 ASCC1 ATL1 ATL3 ATP7A ATXN2 BAG3 BICD2 BSCL2 CCT5 CHCHD10 CHMP2B CTDP1 DCTN1 DNAJB2 DNM2 DNMT1 DST DYNC1H1 EGR2 ELP1 ERBB3 ERBB4 EXOSC8 FBLN5 FBXO38 FGD4 FIG4 FUS GAN GARS1 GDAP1 GJB1 GJB3 GLA GLE1 GRN HARS1 HEXB HK1 HOXD10 HSPB1 HSPB3 HSPB8 IGHMBP2 INF2 KARS1 KIF1A KIF1B KIF5A LITAF LMNA LRSAM1 MAPT MARS1 MATR3 MED25 MFN2 MME MPZ MTMR2 NAGLU NDRG1 NEFH NEFL NEK1 NGF NTRK1 OPTN PFN1 PIP5K1C PLEKHG5 PMP22 PNKP POLG PRPH PRPS1 PRX RAB7A REEP1 RETREG1 SBF2 SCN11A SCN9A SEPTIN9 SETX SH3TC2 SIGMAR1 SLC12A6 SLC25A3 SLC5A7 SOD1 SPG11 SPTLC1 SPTLC2 SQSTM1 SURF1 TARDBP TBK1 TRPV4 TTR UBA1 UBQLN2 VAPB VCP VRK1 WNK1 YARS1</p>
<p>Panel de Enfermedad de Wilson (NGS276):</p>	<p>ATP7B</p>
<p>Panel de Ehlers-Danlos e Cutis Laxa (NGS249):</p>	<p>ADAMTS2 ALDH18A1 ATP6V0A2 ATP7A B3GALT6 B4GALT7 CHST14 COL1A1 COL1A2 COL3A1 COL5A1 COL5A2 EFEMP2 ELN FBLN5 FKBP14 FLNA GORAB LTBP4 PLOD1 RIN2 SLC2A10 SLC39A13 TNXB</p>
<p>Panel de Endocrinopatías Neonatales (NGS268):</p>	<p>ABCC8 CYP11B1 CYP17A1 CYP21A2 DUOXA2 GCK GLIS3 GLUD1 HADH INSR IYD KCNJ11 LHX4 NROB1 PAX8 POU1F1 PROP1 SLC16A1 SLC2A2 SLC5A5 TG THRA THRB TPO TSHB</p>
<p>Panel de Epidermólisis Bullosa (NGS250):</p>	<p>CAST CD151 COL17A1 COL7A1 DSP DST FERMT1 ITGA3 ITGA6 ITGB4 KRT14 KRT5 LAMA3 LAMB3 LAMC2 PLEC TGM5</p>

<p>Panel de Epilepsia (NGS251):</p>	<p>AARS1 ABAT ADAM22 ADGRV1 ADSL ALDH5A1 ALDH7A1 ALG13 AMT AP3B2 ARFGEF2 ARG1 ARHGEF9 ARV1 ARX ASAH1 ATP13A2 BCKDHA BCKDHB BRAT1 BTD CACNA1A CACNA1E CACNA1H CACNB4 CASR CDKL5 CERS1 CHD2 CHRNA2 CHRNA4 CHRN2 CLCN2 CLN3 CLN5 CLN6 CLN8 CNPY3 CPA6 CSTB CTSD CUX2 D2HGDH DBT DENND5A DEPDC5 DLD DNM1 DOCK7 EEF1A2 EFHC1 EPM2A ETHE1 FOLR1 FRRS1L GABBR2 GABRA1 GABRB3 GABRD GABRG2 GAMT GATM GCSH GLDC GLRA1 GLRB GLS GNAO1 GOSR2 GPHN GRIN1 GRIN2A GRIN2B HCN1 HLCS HNRNPU IDH2 ITPA KCNA1 KCNA2 KCNB1 KCNMA1 KCNQ2 KCNQ3 KCNT1 KCTD7 LGI1 MDH2 MFSD8 MOCS1 MOCS2 NECAP1 NEU1 NHLRC1 NTRK2 PANK2 PCDH19 PHACTR1 PHGDH PIGA PLA2G6 PLCB1 PMM2 PNKP PNPO POLG PPT1 PRICKLE1 PRRT2 PSAT1 PSPH RELN RFT1 SCARB2 SCN1A SCN1B SCN2A SCN3A SCN8A SCN9A SLC12A5 SLC13A5 SLC1A2 SLC25A12 SLC25A22 SLC2A1 SLC35A2 SLC6A1 SLC6A5 SLC6A8 SPTAN1 ST3GAL3 STXBP1 SUOX SYNGAP1 SYNJ1 SZT2 TBC1D24 TPP1 TRAK1 WWOX</p>
<p>Panel de Errores Innatos del Metabolismo Tratables (NGS269):</p>	<p>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</p>
<p>Panel de Fibrosis Quística (NGS301):</p>	<p>CFTR</p>
<p>Panel de Hemocromatosis (NGS285):</p>	<p>HFE HFE2 HAMP SLC40A1 TFR2</p>
<p>Panel de Ictiosis (NGS252):</p>	<p>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</p>

**Panel de Inmunodeficiencias
Primarias
(NGS158):**

ACP5 ACTB ADA ADA2 ADAM17 ADAR AICDA AIRE AK2
AP1S3 AP3B1 AP3D1 APOL1 ATM B2M BCL10 BLM BLNK
BTK C1QA C1QB C1QC C1R C1S C2 C3 C4A C4B C5 C6
C7 C8A C8B C9 CARD11 CARD14 CARD9 CASP10 CASP8
CCBE1 CD19 CD247 CD27 CD3D CD3E CD3G CD40
CD40LG CD46 CD55 CD59 CD79A CD79B CD81 CD8A
CEBPE CFB CFD CFH CFHR1 CFHR2 CFHR3 CFHR4
CFHR5 CFI CFP CFTR CHD7 CIITA CLCN7 CLPB CORO1A
CR2 CSF2RA CSF2RB CSF3R CTC1 CTLA4 CTPS1 CTSC
CXCR4 CYBA CYBB DCLRE1B DCLRE1C DKC1 DNMT3B
DOCK8 ELANE EPG5 ERCC6L2 EXTL3 FADD FAS FASLG
FCGR3A FCN3 FERMT3 FOXP1 FOXP3 FPR1 G6PC3
G6PD GATA2 GFI1 HAX1 ICOS IFIH1 IFNAR2 IFNG IFNGR1
IFNGR2 IGKC IGLL1 IKBKB IKZF1 IL10 IL10RA IL10RB
IL12B IL12RB1 IL12RB2 IL17F IL17RA IL1RN IL21 IL21R IL23R
IL2RA IL2RG IL36RN IL6ST IL7R INO80 IRAK1 IRF7 IRF8
ISG15 ITCH ITGB2 ITK JAGN1 JAK1 JAK3 KDM6A KMT2D
KRAS LAMTOR2 LCK LIG1 LIG4 LPIN2 LRBA LRRC8A
LYST MAGT1 MALT1 MASP2 MEFV MOGS MPO MS4A1
MSH6 MTHFD1 MVK MYD88 NBAS NBN NCF1 NCF2 NCF4
NCSTN NFAT5 NFKB1 NFKB2 NFKBIA NHEJ1 NLRC4
NLRP12 NLRP3 NOP10 NRAS ORAI1 OSTM1 PARN PEPD
PGM3 PIK3CD PIK3R1 PIK3R5 PLCG2 PLEKHM1 PMS2
PNP POLA1 POLE2 POMP PRF1 PRKCD PRKDC PSENEN
PSMB8 PSMB9 PSTPIP1 PTEN PTPRC RAB27A RAC2 RAG1
RAG2 RANBP2 RASGRP1 RBCK1 RFX5 RFXANK RFXAP
RHOH RNASEH2A RNASEH2B RNASEH2C RNF168 RORC
RPSA RTEL1 SAMD9 SAMHD1 SBDS SEMA3E SERPING1
SH2D1A SH3BP2 SLC29A3 SLC35C1 SLC37A4 SLC46A1
SMARCAL1 SNX10 SP110 SPINK5 STAT1 STAT2 STAT3
STAT4 STAT5B STIM1 STING1 STK4 STX11 STXBP2 TAP1
TAP2 TAPBP TAZ TBK1 TBX1 TCF3 TCIRG1 TCN2 TERT
TFRC THBD TICAM1 TINF2 TIRAP TLR3 TMC6 TMC8
TNFAIP3 TNFRSF11A TNFRSF13B TNFRSF13C TNFRSF1A
TNFRSF4 TNFSF12 TPP2 TRAF3 TRAF3IP2 TREX1 TRNT1
TTC37 TTC7A TYK2 UNC119 UNC13D UNC93B1 UNG
USB1 VPS13B VPS45 WAS WIPF1 WRAP53 XIAP ZAP70
ZBTB24

**Panel Expandido de
Inmunodeficiencias
autoinflamatorias y
enfermedad inflamatoria
intestinal
(NGS275):**

ABCB1 ACP5 ACTB ADA ADA2 ADAM17 ADAR AICDA
AIRE AK2 AP1S3 AP3B1 AP3D1 APOL1 ATG16L1 ATM B2M
BCL10 BLM BLNK BTK C1QA C1QB C1QC C1R C1S C2 C3
C4A C4B C5 C6 C7 C8A C8B C9 CARD11 CARD14 CARD9
CASP10 CASP8 CCBE1 CD19 CD247 CD27 CD3D CD3E
CD3G CD40 CD40LG CD46 CD55 CD59 CD79A CD79B
CD81 CD8A CEBPE CFB CFD CFH CFHR1 CFHR2 CFHR3
CFHR4 CFHR5 CFI CFP CFTR CHD7 CIITA CLCN7 CLPB
CORO1A CR2 CSF2RA CSF2RB CSF3R CTC1 CTLA4
CTPS1 CTSC CXCR4 CYBA CYBB DCLRE1B DCLRE1C
DDX58 DKC1 DNASE1 DNASE1L3 DNMT3B DOCK8 ELANE
EPG5 ERCC6L2 EXTL3 FADD FAS FASLG FCGR2A
FCGR2B FCGR3A FCN3 FERMT3 FOXD3 FOXP1 FOXP3
FPR1 G6PC3 G6PD GATA2 GFI1 GPNMB HAX1 ICOS IFIH1
IFNAR2 IFNG IFNGR1 IFNGR2 IGKC IGLL1 IKBKB IKZF1
IL10 IL10RA IL10RB IL12B IL12RB1 IL12RB2 IL17F IL17RA
IL1RN IL21 IL21R IL23R IL2RA IL2RG IL31RA IL36RN IL6
IL6ST IL7R INO80 IRAK1 IRF5 IRF7 IRF8 IRGM ISG15
ITCH ITGB2 ITK JAGN1 JAK1 JAK3 KDM6A KMT2D KRAS
LAMTOR2 LCK LIG1 LIG4 LPIN2 LRBA LRRRC8A LYST
MAGT1 MALT1 MASP2 MEFV MOGS MPO MS4A1 MSH6
MTHFD1 MVK MYD88 NBAS NBN NCF1 NCF2 NCF4
NCSTN NFAT5 NFKB1 NFKB2 NFKBIA NHEJ1 NLRC4
NLRP1 NLRP12 NLRP3 NLRP7 NOD2 NOP10 NRAS ORAI1
OSMR OSTM1 PARN PEPD PGM3 PIK3CD PIK3R1 PIK3R5
PLCG2 PLEKHM1 PMS2 PNP POLA1 POLE2 POMP PRF1
PRKCD PRKDC PSEN1 PSENEN PSMB8 PSMB9 PSTPIP1
PTEN PTPN22 PTPRC RAB27A RAC2 RAG1 RAG2
RANBP2 RASGRP1 RBCK1 RFX5 RFXANK RFXAP RHOH
RNASEH2A RNASEH2B RNASEH2C RNF168 RORC RPSA
RTEL1 SAMD9 SAMHD1 SBDS SEMA3E SERPING1 SH2D1A
SH3BP2 SKIV2L SLC29A3 SLC35C1 SLC37A4 SLC46A1
SMARCAL1 SNX10 SP110 SPINK5 STAT1 STAT2 STAT3
STAT4 STAT5B STIM1 STING1 STK4 STX11 STXBP2 TAP1
TAP2 TAPBP TAZ TBK1 TBX1 TCF3 TCIRG1 TCN2 TERT
TFRC THBD TICAM1 TINF2 TIRAP TLR3 TMC6 TMC8
TNFAIP3 TNFRSF11A TNFRSF13B TNFRSF13C TNFRSF1A
TNFRSF4 TNFSF12 TPP2 TRAF3 TRAF3IP2 TREX1 TRNT1
TTC37 TTC7A TTR TYK2 UNC119 UNC13D UNC93B1 UNG
USB1 VPS13B VPS45 WAS WIPF1 WRAP53 XIAP ZAP70
ZBTB24

<p>Panel de Leucodistrofias (NGS284):</p>	<p>AARS2, ABCD1, ACOX1, ADAR, AIMP1, ALDH3A2, APP, ARSA, ASPA, ATP7A, ATP7B, ATPAF2, BCAP31, BCS1L, BEST1, CLCN2, COL4A1, COQ2, COQ8A, COQ9, COX10, COX15, CSF1R, CYP27A1, CYP7B1, D2HGDH, DARS2, DGUOK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ERCC2, ERCC3, ERCC6, ERCC8, ETFDH, FA2H, FAM126A, FUCA1, GALC, GBE1, GFAP, GFM1, GJA1, GJC2, GLA, GLB1, GRN, GTF2H5, HEPACAM, HEXA, HSD17B4, HSPD1, HTRA1, L2HGDH, LMNB1, MAPT, MCOLN1, MLC1, MPLKIP, MRPS16, NDUFAF1, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NOTCH3, NPC1, NPC2, OCLN, OCRL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PHYH, PLP1, POLG, POLG2, POLR1C, POLR3A, POLR3B, PPT1, PRF1, PSAP, PSAT1, PSEN1, PSEN2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RRM2B, SAMHD1, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SLC16A2, SLC17A5, SLC25A12, SLC25A4, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, STX11, STXBP2, SUCLA2, SUMF1, SURF1, TACO1, TREM2, TREX1, TUFM, TWNK, TYMP, TYROBP, UNC13D, ZFYVE26</p>
<p>Panel de Miocardiopatías Hereditarias (NGS253):</p>	<p>ABCC9 ACTA1 ACTA2 ACTC1 ACTN2 ACVR2B ALMS1 ANKRD1 BAG3 BRAF CASQ2 CAV3 CAVIN4 CFC1 CHD7 COX15 CRELD1 CRYAB CSRP3 DES DMD DNAJC19 DOLK DSC2 DSG2 DSP DTNA ELN EMD EYA4 FANCB FHL1 FKRP FKTN FXN GAA GATA4 GATA6 GATAD1 GDF1 GJA1 GLA HADHA HFE HRAS ILK JAG1 JPH2 JUP KLF10 KRAS LAMA2 LAMA4 LAMP2 LDB3 LMNA MAP2K1 MAP2K2 MED13L MGP MIB1 MYBPC3 MYH6 MYH7 MYL2 MYL3 MYLK2 MYO6 MYOZ2 MYPN NEXN NF1 NKX2-5 NKX2-6 NODAL NOTCH1 NOTCH2 NPPA NRAS PDLIM3 PKD1L1 PKP2 PLN PRDM16 PRKAG2 PTEN PTPN11 RAF1 RBM20 SCN5A SDHA SEMA3E SGCB SGCD SGCG SLC25A4 SMAD6 SOS1 TAZ TBX1 TBX20 TBX5 TCAP TFAP2B TLL1 TMPO TNNC1 TNNI2 TNNI3 TNNT2 TPM1 TRIM63 TTN TTR TXNRD2 VCL ZFPM2 ZIC3</p>
<p>Panel de Retinopatías (NGS254):</p>	<p>ABCA4, BBS2, BEST1, C8orf37, CA4, CDHR1, CHM, CLRN1, CNGB1, CRB1, DHDDS, EYS, FAM161A, GUCA1B, HGSNAT, IMPDH1, IMPG2, KLHL7, MAK, MERTK, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PROM1, PRPF31, PRPF6, PRPF8, PRPH2, RBP3, RDH12, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, TOPORS, TTC8, TULP1, ZNF513</p>

Panel de Síndrome de Noonan y Rasopatias (NGS255):	BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1 RIT1 SHOC2 SOS1 SPRED1
Panel de Síndrome Urémico Hemolítico (NGS297):	C3 CD46 CFB CFH CFI THBD
Panel de Síndrome Nefrótico (NGS256):	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 Hipoacusia (NGS258):	ACTG1, ADGRV1, AIFM1, BSND, CABP2, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DIABLO, DIAPH1, EDN3, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NDP, OPA1, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX1, PAX3, PCDH15, PDZD7, PEX7, PHYH, PJVK, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, SERPINB6, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TECTA, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, TYR, USH1C, USH1G, USH2A, WFS1
Panel de Trombofilias (NGS299):	ADAMTS13 CBS F2 F5 PROC PROS1 SERPINC1
Panel de Síndrome de Rett (NGS263):	MECP2
Panel de Síndrome de Marfan (NGS278):	FBN1
Panel para Síndrome de Cornelia de Lange (NGS286):	ANKRD11 HDAC8 NIPBL RAD21 SMC1A SMC3 EP300
Panel para Síndrome de Charge (NGS300):	CDH7

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