

# Carrier Screening Invitae Comprehensive Panel

569 genes analizados y relacionados con enfermedades hereditarias.

✓ CFTR	✓ SMN1	
✓ AAAS	✓ ABCA12	✓ ABCA3
✓ ABCA4	✓ ABCB11	✓ ABCB4
✓ ABCC2	✓ ABCC8	✓ ABCD1
✓ ACAD9	✓ ACADM	✓ ACADVL
✓ ACAT1	✓ ACOX1	✓ ACSF3
✓ ADA	✓ ADAMTS2	✓ ADAMTSL4
✓ ADGRG1	✓ ADGRV1	✓ AGA
✓ AGL	✓ AGPS	✓ AGXT
✓ AHI1	✓ AIPL1	✓ AIRE
✓ ALDH3A2	✓ ALDH7A1	✓ ALDOB
✓ ALG1	✓ ALG13	✓ ALG6
✓ ALMS1	✓ ALPL	✓ AMN
✓ AMT	✓ ANO10	✓ AP1S1
✓ AQP2	✓ AR	✓ ARG1
✓ ARL6	✓ ARSA	✓ ARSB
✓ ARSE	✓ ARX	✓ ASL
✓ ASNS	✓ ASPA	✓ ASS1
✓ ATM	✓ ATP6V1B1	✓ ATP7A
✓ ATP7B	✓ ATP8B1	✓ ATRX
✓ AVPR2	✓ BBS1	✓ BBS10

✓ BBS12	✓ BBS2	✓ BBS4
✓ BBS5	✓ BBS7	✓ BBS9
✓ BCKDHA	✓ BCKDHB	✓ BCS1L
✓ BLM	✓ BLOC1S3	✓ BLOC1S6
✓ BMP1	✓ BRIP1	✓ BSND
✓ BTK	✓ CAD	✓ CANT1
✓ CAPN3	✓ CASQ2	✓ CBS
✓ CC2D1A	✓ CC2D2A	✓ CCDC103
✓ CCDC39	✓ CCDC88C	✓ CD3D
✓ CD3E	✓ CD40	✓ CD40LG
✓ CD59	✓ CDH23	✓ CEP152
✓ CEP290	✓ CERKL	✓ CHAT
✓ CHM	✓ CHRNE	✓ CHRNG
✓ CIITA	✓ CLCN1	✓ CLN3
✓ CLN5	✓ CLN6	✓ CLN8
✓ CLRN1	✓ CNGB3	✓ COL11A2
✓ COL17A1	✓ COL27A1	✓ COL4A3
✓ COL4A4	✓ COL4A5	✓ COL7A1
✓ COX15	✓ CPS1	✓ CPT1A
✓ CPT2	✓ CRB1	✓ CRTAP
✓ CTNS	✓ CTSA	✓ CTSC
✓ CTSD	✓ CTSK	✓ CYBA
✓ CYBB	✓ CYP11A1	✓ CYP11B1

✓ CYP11B2	✓ CYP17A1	✓ CYP19A1
✓ CYP1B1	✓ CYP21A2	✓ CYP27A1
✓ CYP27B1	✓ CYP7B1	✓ DBT
✓ DCAF17	✓ DCLRE1C	✓ DDX11
✓ DFNB59	✓ DGAT1	✓ DGUOK
✓ DHCR7	✓ DHDDS	✓ DKC1
✓ DLD	✓ DLL3	✓ DMD
✓ DNAH11	✓ DNAH5	✓ DNAI1
✓ DNAI2	✓ DNMT3B	✓ DOK7
✓ DUOX2	✓ DYNC2H1	✓ DYSF
✓ EDA	✓ EIF2AK3	✓ EIF2B1
✓ EIF2B2	✓ EIF2B3	✓ EIF2B4
✓ EIF2B5	✓ ELP1	✓ EMD
✓ EPG5	✓ ERCC2	✓ ERCC6
✓ ERCC8	✓ ESCO2	✓ ETFA
✓ ETFB	✓ ETFDH	✓ ETHE1
✓ EVC	✓ EVC2	✓ EXOSC3
✓ EYS	✓ F9	✓ FAH
✓ FAM161A	✓ FANCA	✓ FANCB
✓ FANCC	✓ FANCD2	✓ FANCE
✓ FANCG	✓ FANCI	✓ FANCL
✓ FBP1	✓ FBXO7	✓ FH
✓ FHL1	✓ FKBP10	✓ FKRP

✓ FKTN	✓ FMO3	✓ FMR1
✓ FOXN1	✓ FOXRED1	✓ FRAS1
✓ FREM2	✓ FUCA1	✓ G6PC
✓ G6PC3	✓ GAA	✓ GALC
✓ GALE	✓ GALK1	✓ GALNS
✓ GALNT3	✓ GALT	✓ GAMT
✓ GATM	✓ GBA	✓ GBE1
✓ GCDH	✓ GCH1	✓ GDF5
✓ GFM1	✓ GHR	✓ GJB1
✓ GJB2	✓ GLA	✓ GLB1
✓ GLDC	✓ GLE1	✓ GNE
✓ GNPAT	✓ GNPTAB	✓ GNPTG
✓ GNS	✓ GORAB	✓ GRHPR
✓ GRIP1	✓ GSS	✓ GUCY2D
✓ GUSB	✓ HADH	✓ HADHA
✓ HADHB	✓ HAMP	✓ HAX1
✓ HBA1, HBA2	✓ HBB	✓ HCFC1
✓ HEXA	✓ HEXB	✓ HGSNAT
✓ HJV	✓ HLCS	✓ HMGCL
✓ HMOX1	✓ HOGA1	✓ HPD
✓ HPRT1	✓ HPS1	✓ HPS3
✓ HPS4	✓ HPS5	✓ HPS6
✓ HSD17B10	✓ HSD17B3	✓ HSD17B4

✓ HSD3B2	✓ HYAL1	✓ HYLS1
✓ IDS	✓ IDUA	✓ IGHMBP2
✓ IKBKB	✓ IL2RG	✓ IL7R
✓ INVS	✓ ITGA6	✓ ITGB3
✓ ITGB4	✓ IVD	✓ JAK3
✓ KCNJ1	✓ KCNJ11	✓ LTCAM
✓ LAMA2	✓ LAMA3	✓ LAMB3
✓ LAMC2	✓ LARGE1	✓ LCA5
✓ LDLR	✓ LDLRAP1	✓ LHX3
✓ LIFR	✓ LIG4	✓ LIPA
✓ LMBRD1	✓ LOXHD1	✓ LPL
✓ LRAT	✓ LRP2	✓ LRPPRC
✓ LYST	✓ MAK	✓ MAN2B1
✓ MANBA	✓ MCEE	✓ MCOLN1
✓ MCPH1	✓ MECP2	✓ MECR
✓ MED17	✓ MESP2	✓ MFSD8
✓ MID1	✓ MKKS	✓ MKS1
✓ MLC1	✓ MLYCD	✓ MMAA
✓ MMAB	✓ MMACHC	✓ MMADHC
✓ MOCS1	✓ MOCS2	✓ MPI
✓ MPL	✓ MPV17	✓ MRE11
✓ MTHFR	✓ MTM1	✓ MTR
✓ MTRR	✓ MTPP	✓ MUSK

✓ MUT	✓ MVK	✓ MYO15A
✓ MYO7A	✓ NAGA	✓ NAGLU
✓ NAGS	✓ NBN	✓ NCF2
✓ NDRG1	✓ NDUFAF2	✓ NDUFAF5
✓ NDUFS4	✓ NDUFS6	✓ NDUFS7
✓ NDUFV1	✓ NEB	✓ NEU1
✓ NGLY1	✓ NPC1	✓ NPC2
✓ NPHP1	✓ NPHS1	✓ NPHS2
✓ NROB1	✓ NR2E3	✓ NSMCE3
✓ NTRK1	✓ OAT	✓ OCA2
✓ OCRL	✓ OPA3	✓ OSTM1
✓ OTC	✓ OTOA	✓ OTOF
✓ P3H1	✓ PAH	✓ PANK2
✓ PC	✓ PCBD1	✓ PCCA
✓ PCCB	✓ PCDH15	✓ PCNT
✓ PDHA1	✓ PDHB	✓ PEPD
✓ PET100	✓ PEX1	✓ PEX10
✓ PEX12	✓ PEX13	✓ PEX16
✓ PEX2	✓ PEX26	✓ PEX5
✓ PEX6	✓ PEX7	✓ PFKM
✓ PGM3	✓ PHGDH	✓ PHKB
✓ PHKG2	✓ PHYH	✓ PIGN
✓ PKHD1	✓ PLA2G6	✓ PLEKHG5

✓ PLOD1	✓ PLP1	✓ PMM2
✓ PNPO	✓ POLG	✓ POLH
✓ POMGNT1	✓ POMT1	✓ POMT2
✓ POR	✓ POU1F1	✓ PPT1
✓ PRCD	✓ PRDM5	✓ PRF1
✓ PROP1	✓ PRPS1	✓ PSAP
✓ PTPRC	✓ PTS	✓ PUS1
✓ PYGM	✓ QDPR	✓ RAB23
✓ RAG1	✓ RAG2	✓ RAPSN
✓ RARS2	✓ RDH12	✓ RLBP1
✓ RMRP	✓ RNASEH2A	✓ RNASEH2B
✓ RNASEH2C	✓ RP2	✓ RPE65
✓ RPGRIP1L	✓ RS1	✓ RTEL1
✓ RXYLT1	✓ RYR1	✓ SACS
✓ SAMD9	✓ SAMHD1	✓ SCO2
✓ SEC23B	✓ SEPSECS	✓ SGCA
✓ SGCB	✓ SGCD	✓ SGCG
✓ SGSH	✓ SKIV2L	✓ SLC12A1
✓ SLC12A3	✓ SLC12A6	✓ SLC17A5
✓ SLC19A2	✓ SLC19A3	✓ SLC1A4
✓ SLC22A5	✓ SLC25A13	✓ SLC25A15
✓ SLC25A20	✓ SLC26A2	✓ SLC26A3
✓ SLC26A4	✓ SLC27A4	✓ SLC35A3

✓ SLC37A4	✓ SLC38A8	✓ SLC39A4
✓ SLC45A2	✓ SLC4A11	✓ SLC5A5
✓ SLC6A8	✓ SLC7A7	✓ SMARCA1
✓ SMPD1	✓ SNAP29	✓ SPG11
✓ SPR	✓ SRD5A2	✓ ST3GAL5
✓ STAR	✓ STX11	✓ STXBP2
✓ SUMF1	✓ SUOX	✓ SURF1
✓ SYNE4	✓ TANGO2	✓ TAT
✓ TAZ	✓ TBCD	✓ TBCE
✓ TCIRG1	✓ TCN2	✓ TECPR2
✓ TERT	✓ TF	✓ TFR2
✓ TG	✓ TGM1	✓ TH
✓ TK2	✓ TMC1	✓ TMEM216
✓ TMEM67	✓ Tmprss3	✓ TPO
✓ TPP1	✓ TREX1	✓ TRIM32
✓ TRIM37	✓ TRMU	✓ TSEN54
✓ TSFM	✓ TSHB	✓ TSHR
✓ TTC37	✓ TTPA	✓ TULP1
✓ TYMP	✓ TYR	✓ TYRP1
✓ UBR1	✓ UNC13D	✓ USH1C
✓ USH2A	✓ VDR	✓ VLDLR
✓ VPS11	✓ VPS13A	✓ VPS13B

✓ VPS45	✓ VPS53	✓ VRK1
✓ VSX2	✓ WAS	✓ WISP3
✓ WNT10A	✓ WRN	✓ XPA
✓ XPC	✓ ZBTB24	✓ ZFYVE26
✓ ZNF469		

## Genes con presentación variable

✓ BTD	✓ F11	✓ F2	✓ F5
✓ G6PD	✓ GP1BA	✓ GP9	✓ HFE
✓ HGD	✓ MCCC1	✓ MCCC2	✓ MEFV
✓ SERPINA1			