



Catálogo de pruebas genómicas  
por especialidad

Nefrología y  
endocrinología

## 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, CFHRI, 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, AH11, 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, DHCRR7, 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, FEZ1, 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, HSD1B2, HSD3B7, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IHHL, 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, LMFI, LMX1B, LRAT, LRIG2, LRP4, LRP5, LZTFL1, MAFB, MAGI2, MAPKBPI, 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, NKK2-5, NKK3-2, NME8, NMNAT1, NODAL, NOG, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR0B2, NR1H4, NR3C2, NSDHL, NSMF, NUP107, NUP93, OBL1, OCRL, OFDI, OSGE, 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, PROPI, PRPH2, PTH1R, PTHLH, PTPRO, RBBP8, RD3, RDH12, RDH5, REN, RET, RHO, RLRP1, 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, SLC10A1, SLC10A3, SLC10A5, SLC10A6, SLC10A7, 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## 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, FBPI, 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, LASIL, 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, NEURODI, NEUROG3, NGLY1, NHLRC1, NKX2-2, NPHP1, NPHP3, NR0B2, NSD1, NTRK2, OFDI, 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, RLBPI, RPGRIPI1, RPS6KA3, SDCCAG8, SETD2, SH2B1, SIM1, SLC16A1, SLC19A2, SLC29A3, SLC2A2, SLC35A1, SLC35A2, SLC35C1, SNRPN, SPG11, SRD5A3, SSR4, STAT1, STAT3, TBX3, THOC2, THRA, TMEM165, TMEM67, TRAF3IP1, TRAPP9, TRIM32, TRMT10A, TTC21B, TTC8, TUSC3, UCP2, UCP3, VPS13B, WFS1, XRCC4, XYL1, ZBTB20, ZFP57, ZMPSTE24, ZNF711