

Ciliopathia (Ciliopathy) génpanel v:2022.02.09.

Panel tartalma 247 gén:

ACVR2B, ADAMTS9, AHI1, AIPL1, AK7, ALG8, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC4, ARMC9, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C21orf2, C2CD3, C2orf71, C5orf42, C8orf37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CCNQ, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP300, CFAP410, CFAP418, CFAP53, CFTR, CLUAP1, CPE, CPLANE1, CRB1, CRB2, CRX, CTU2, CSPP1, DCDC2, DDX59, DHCR7, DLG5, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC3L2, EXOC8, FAM149B1, FAM186B, FAM58A, FAN1, FBN3, FGFR1, FGFR2, FGFR3, FOXH1, FOXJ1, GANAB, GAS2L2, GAS8, GDF1, GLI2, GLI3, GLIS2, GUCY2D, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IMPDH1, INPP5E, INTU, INVS, IQCB1, IQCE, KATNIP, KCNJ13, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, LAMA1, LBR, LCA5, LEFTY2, LRAT, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MMP21, MRE11, MUC1, NEK1, NEK8, NKX2-5, NME8, NODAL, NOTCH2, NPHP1, NPHP3, NPHP4, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, PDE6D, PDIA6, PIAS1, PIBF1, PIH1D3, PIK3C2A, PKD1, PKD1L1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, PRKCSH, RABL2A, RBM48, RCOR1, RD3, RDH12, RPE65, RPGR, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SBDS, SCAPER, SCLT1, SDCCAG8, SEC63, SLC30A7, SLC41A1, SMAD2, SPAG1, SPATA7, SPEF2, SUFU, TAPT1, TBC1D32, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TRAPPC3, TRIM32, TTC12, TTC21B, TTC26, TTC8, TULP1, TXNDC15, UMOD, USP9X, VPS13B, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR63, XPNPEP3, ZIC3, ZMYND10, ZNF423, ZSWIM6.

Forrás:

Rare multisystem ciliopathy Super panel (Version 4.122)

<https://panelapp.genomicsengland.co.uk/panels/728/> (Megtekintve: 2022.02.09.)

Ciliopathy Panel

<https://blueprintgenetics.com/tests/panels/nephrology/ciliopathy-panel/>

(Megtekintve: 2022.02.09.)

Ciliopathy Panel

<https://www.preventiongenetics.com/testInfo?val=Ciliopathy+Panel>

(Megtekintve: 2022.02.09.)

Invitae Ciliopathies Panel

<https://www.invitae.com/en/physician/tests/04102/> (Megtekintve: 2022.02.09.)