

Vesebetegségek (Renal disorders) génpanel v:2023.10.12.

Panel tartalma 471 gén:

ABCG2, ACE, ACTA2, ACTG2, ACTN4, ADAMTS13, ADAMTS9, ADCY10, AGK, AGT, AGTR1, AGXT, AHI1, ALDH1A2, ALG1, ALG5, ALG8, ALG9, ALMS1, ALPL, AMN, ANKFY1, ANKS6, ANLN, ANOS1, AP2S1, APOE, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL3, ARL6, ARMC9, ATP1A1, ATP6V0A4, ATP6V1B1, ATXN10, AVP, AVPR2, B9D1, B9D2, BAP1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BMP7, BMPR2, BNC2, BSND, C1QA, C1QB, C1QC, C2CD3, C3, C5orf42, C8orf37, CA2, CACNA1H, CASR, CAV1, CC2D2A, CCDC28B, CD151, CD2AP, CD46, CDC5L, CDK20, CDKN2B, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD1L, CHD7, CHRM3, CHRNA3, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CLU, CNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ7, COQ8B, COQ9, COX10, CPLANE1, CPS1, CRB2, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27B1, CYP2R1, CYS1, CSPP1, DAAM2, DACH1, DACT1, DCDC2, DDX59, DGKE, DHCR7, DHFR, DKC1, DLC1, DLG3, DLG5, DMP1, DNAJB11, DSTYK, DYNC2H1, DZIP1L, E2F3, EGF, EHHADH, EMP2, ENPP1, ETFA, EXOC3L2, EXOC8, EYA1, FAH, FAM149B1, FAM20A, FAM58A, FAN1, FAT1, FBXW7, FGF20, FGF23, FH, FIBP, FLCN, FN1, FOXC1, FOXC2, FOXD2, FOXI1, FRAS1, FREM1, FREM2, FXYD2, GANAB, GAPVD1, GATA2, GATA3, GATM, GDNF, GIF, GLA, GLI3, GLIS2, GNA11, GNAS, GON7, GPC3, GREB1L, GREM1, GRHRP, GRIP1, HAAO, HCN3, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HS2ST1, HSD11B2, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, IL1RAP, INF2, INPP5E, INVS, IQCB1, ISL1, ITGA3, ITGA8, ITGB4, ITS1, ITS2, JAG1, KANK1, KANK2, KANK4, KATNIP, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KCNK3, KDM6A, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, KIRREL1, KIT, KL, KLHL3, KMT2D, KYNU, LAGE3, LAMA5, LAMB2, LCAT, LIFR, LMNA, LMX1B, LRIG2, LRP4, LRP5, LZTFL1, MAFB, MAGED2, MAGI2, MAPKB1, MED28, MEFV, MET, MITF, MKKS, MKS1, MLH1, MMACHC, MMADHC, MMUT, MOCOS, MSH2, MSH6, MT-TF, MTR, MTRR, MTX2, MUC1, MYH11, MYH9, MYO1E, MYOCD, NADSYN1, NEIL1, NEK1, NEK8, NEU1, NIPBL, NLRP3, NOP10, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NRIP1, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OSGEP, OSR1, PAX2, PBX1, PDE6D, PDIA6, PDSS2, PHEX, PIBF1, PIGA, PKD1, PKD2, PKHD1, PLCE1, PLG, PLVAP, PMM2, PMS2, PNMT, POC1B, PODXL, PRDM15, PRKCSH, PTEN, PTGIS, PTPRO, PXDN, RCAN1, REN, RET, ROBO2, ROR2, RPGRIP1L, RRAGD, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SDHB, SDHC, SDHD, SEC61A1, SEC63, SGPL1, SHH, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC19A2, SLC19A3, SLC1A1, SLC22A12, SLC26A1, SLC26A6, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC9A3, SLC9A3R1, SLIT2, SMAD9, SMARCA4, SMARCA1, SOX17, SPRY1, SRGAP1, STRA6, STRADA, SUFU, SYNPO, TBC1D1, TBC1D8B, TBX18,

TBX6, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM107, TMEM127, TMEM138, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TNS2, TNXB, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRAPPC3, TRIM32, TRIM8, TRPC6, TRPM6, TSC1, TSC2, TSHZ3, TTC21B, TTC8, TXNDC15, UMOD, UPK2, UPK3A, VDR, VHL, VIPAS39, VPS33B, VTN, WBP11, WDPCP, WDR19, WDR35, WDR60, WDR72, WDR73, WNK1, WNK4, WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, XPR1, YRDC, ZIC3, ZMPSTE24, ZMYM2, ZNF365, ZNF423

Forrás:

Kidney Diseases

<https://cegat.com/diagnostics/rare-diseases/kidney-diseases/> (Megtekintve: 2023.07.19.)

Polycystic Kidney Disease Panel

<https://blueprintgenetics.com/tests/panels/nephrology/polycystic-kidney-disease-panel/>
(Megtekintve: 2023.05.18.)

Atypical haemolytic uraemic syndrome (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/139/> (Megtekintve: 2023.05.18.)

CAKUT (Version 1.173)

<https://panelapp.genomicsengland.co.uk/panels/234/> (Megtekintve: 2023.05.18.)

Cystic kidney disease (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/283/> (Megtekintve: 2023.05.18.)

Extreme early-onset hypertension (Version 1.21)

<https://panelapp.genomicsengland.co.uk/panels/314/> (Megtekintve: 2023.05.18.)

Haematuria (Version 2.11)

<https://panelapp.genomicsengland.co.uk/panels/99/> (Megtekintve: 2023.05.18.)

Membranoproliferative glomerulonephritis including C3 glomerulopathy (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/83/> (Megtekintve: 2023.05.18.)

Nephrocalcinosis or nephrolithiasis (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/149/> (Megtekintve: 2023.05.18.)

Proteinuric renal disease (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/106/> (Megtekintve: 2023.05.18.)

Renal and urinary tract disorders (Version 1.22)

<https://panelapp.genomicsengland.co.uk/panels/493/> (Megtekintve: 2023.05.18.)

Renal tubulopathies (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/292/> (Megtekintve: 2023.05.18.)

Unexplained kidney failure in young people (Version 1.117)

<https://panelapp.genomicsengland.co.uk/panels/156/> (Megtekintve: 2023.05.18.)

Tubulointerstitial kidney disease (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/548/> (Megtekintve: 2023.05.18.)

Renal ciliopathies (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/725/> (Megtekintve: 2023.05.18.)

Unexplained young onset end-stage renal disease (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/678/> (Megtekintve: 2023.05.18.)

Inherited renal cancer (Version 1.24)

<https://panelapp.genomicsengland.co.uk/panels/521/> (Megtekintve: 2023.05.18.)