Genpanel Nephropathien

*= MLPA vorhanden

Alport-Syndrom inkl Haematurie

3 Gene: COL4A1, COL4A3*, COL4A4*, COL4A5*, MYH9*

Referenz: "Haematuria" v2.15, green, Genomics England PanelApp

Atypisches hämolytisch-urämisches Syndrom

9 Gene: C3, CD46, CFB, CFH, CFHR1, CFHR3, CFI, DGKE, MMACHC

Referenz: "Atypical haemolyic uraemic syndrome" v.3.6, green, Genomics England PanelApp

Zystische Nierenerkrankung

36 Gene: ALG5, ALG8, ALG9, ANKS6, CEP164, CEP83, CLCN5, COL4A1, DNAJB11, DZIP1L, FLCN, GANAB, GLA, HNF1B, IFT140, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, PAX2, PKD1, PKD2, PKHD1, PRKCSH, SEC63, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, VHL, WDR19, XPNPEP3

Referenz: "Cystic kidney disease", v7.10, green, Genomics England PanelApp

Renale Ciliopathien

75 Gene: AHI1, ALMS1, ANKS6, ARL13B, ARL6, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C5orf42, CC2D2A, CENPF, CEP104, CEP164, CEP290, CEP41, CEP83, CRB2, CSPP1, DDX59, DHCR7, DLG5, DYNC2H1, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, INPP5E, INVS, IQCB1, KIAA0586, KIAA0753, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKD1, PKD2, PKHD1, PMM2, RPGRIP1L, SDCCAG8, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, WDR35, WDR60, XPNPEP3

Referenz: "Renal Ciliopathies", v3.15, green, Genomics England PanelApp

CAKUT

59 Gene: ACE, ACTG2, AGT, AGTR1, ANOS1, BNC2, CEP55, CHD7, CHRM3, CHRNA3, CTU2, DHCR7, DSTYK, EYA1, FAM58A, FRAS1, FREM1, FREM2, GATA3, GLI3, GPC3, GREB1L, GRIP1, HAAO, HNF1B, HOXA13, HPSE2, ITGA8, JAG1, KDM6A, KMT2D, KYNU, LIFR, LRIG2, LRP4, MYOCD, NADSYN1, NIPBL, NOTCH2, NPHP3, PAX2, PBX1, PLVAP, REN, RET, ROBO1, ROBO2, ROR2, SALL1, SIX5, STRA6, TBC1D1, TBX18, TFAP2A, TMEM260, TRAP1, WBP11, ZIC3, ZMYM2

Referenz: «CAKUT» (Version 1.178, green), Genomics England PanelApp

Membranoproliferative Glomerulonephritis

8 Gene: C3, CFB, CFH, CFHR1, CFHR2, CFHR5, CFI, DGKE

Referenz: «Membranoproliferative glomerulonephritis including C3 glomerulopathy» (Version 3.6, green), Genomics England PanelApp

Nephrokalzinose, Nephrolithiasis

35 Gene: AGXT, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, OCRL, PHEX, RRAGD, SLC12A1, SLC22A12, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, STRADA, VIPAS39, VPS33B, WDR72, XDH

Referenz: «Nephrocalcinosis or nephrolithiasis» (Version 4.19, green), Genomics England PanelApp

Nierenerkrankungen (grosses Panel)

290 Gene: ACE, ACTG2, ACTN4, AGT, AGTR1, AGXT, AHI1, ALG5, ALG8, ALG9, ALMS1, AMN, ANKS6, ANOS1, AP2S1, APOA1, APOA2, APOC2, APOE, APRT, AQP2, ARHGDIA, ARL13B, ARL6, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BNC2, BSND, C3, CA2, CASR, CC2D2A, CCNQ, CD151, CD46, CENPF, CEP104, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CILK1, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, DAAM2, DDX59, DGKE, DHCR7, DLC1, DLG5, DNAJB11, DSTYK, DYNC2H1, DYNC2I1, DZIP1L, EYA1, FAH, FAM20A, FAN1, FAT1, FGA, FLCN, FN1, FRAS1, FREM1, FREM2, GANAB, GATA3, GATM, GLA, GLI3, GNA11, GON7, GPC3, GREB1L, GRHPR, GRIP1, GSN, HAAO, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HSD11B2, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITSN1, JAG1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KDM6A, KIAA0586, KIAA0753, KIF7, KLHL3, KMT2D, KYNU, LAGE3, LAMB2, LCAT, LIFR, LMX1B, LRIG2, LRP4, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MKKS, MKS1, MMACHC, MOCOS, MTX2, MUC1, MYH9, MYO1E, MYOCD, NADSYN1, NEK8, NIPBL, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NUP107, NUP133, NUP85, NUP93, OCRL, OFD1, OSGEP, PAX2, PBX1, PDSS2, PHEX, PKD1, PKD2, PKHD1, PLCE1, PLVAP, PMM2, PODXL, PRDM15, PRKCSH, REN, RET, RMND1, ROBO1, ROBO2, ROR2, RPGRIP1L, RRAGD, RRM2B, SALL1, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC7A9, SMARCAL1, STRA6, STRADA, TBC1D1, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TNS2, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM8, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VHL, VIPAS39, VPS33B, WBP11, WDPCP, WDR19, WDR35, WDR72, WDR73, WNK1, WNK4, WT1, XDH, XPNPEP3, YRDC, ZIC3, ZMYM2

Referenz: Renal superpanel – broad (Version 23.33, green), Genomics England PanelApp

Die aufgeführten Gene entsprechen den der jeweiligen Referenz zugrundeliegenden Empfehlungen. Die gemäss Genomics England PanelApp aufgeführten Gene entsprechen den als «diagnostic-grade», also diagnostisch gewerteten Genen (s.a. https://panelapp.genomicsengland.co.uk/#!Guidelines).

Die Auswertung weiterer Gene ist nach Rücksprache mit unserem Labor möglich.