Gen-Panels Nierenerkrankungen | LI

Hinweise:

- Gen-Panels mit mehr als 10 Genen dürfen gemäss Analysenliste des BAG nur durch Ärzte mit einem FMH-Titel Medizinische Genetik verordnet werden.
- Die Gen-Panels sind nicht endgültig und können je nach Fragestellung auf Wunsch angepasst werden.
- Die Gen-Panels können abhängig von der aktuellen Datenlage zum Zeitpunkt der Analysen von denjenigen auf dieser Liste abweichen.

Nierenerkrankungen, umfassendes Panel v1 (392 Gene)

ACE, ACTG2, ACTN4, ADAMTS13, ADAMTS9, ADCY10, ADIPOR1, AGT, AGTR1, AGXT, AHI1, ALDH1A2, ALG1, ALG5, ALG8, ALG9, ALMS1, ALPL, AMN, ANKH, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOA2, APOC2, APOC3, APOE, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL6, ARMC9, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, AVIL, AVP, AVPR2, B2M, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, BNC2, BSND, C1GALT1C1, C3, CA2, CACNA1H, CASR, CAV1, CC2D2A, CCDC28B, CCNQ, CD151, CD2AP, CD46, CDC73, CENPF, CEP104, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP418, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CILK1, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CSPP1, CST3, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, CYP3A4, DAAM2, DCDC2, DDX59, DGKE, DHCR7, DICER1, DLC1, DLG5, DMP1, DNAJB11, DSTYK, DYNC2H1, DYNC2I1, DZIP1L, EHHADH, EMP2, ENPP1, EXOC3L2, EYA1, FAH, FAM20A, FAM20C, FAN1, FAT1, FGA, FGF20, FGF23, FLCN, FN1, FOXI1, FRAS1, FREM1, FREM2, FXYD2, G6PC1, GANAB, GAPVD1, GATA3, GATM, GCM2, GLA, GLI3, GLIS2, GNA11, GNAS, GON7, GPC3, GPHN, GREB1L, GRHPR, GRIP1, GSN, HAAO, HGD, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HS2ST1, HSD11B2, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITGB4, ITSN1, JAG1, KANK1, KANK2, KANK4, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KDM6A, KIAA0586, KIAA0753, KIF14, KIF7, KIRREL1, KL, KLHL3, KMT2D, KYNU, LAGE3, LAMA5, LAMB2, LCAT, LIFR, LMX1B, LRIG2, LRP4, LRP5, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MKS1, MMACHC, MOCOS, MOCS1, MT-TF, MT-TI, MT-TL1, MTX2, MUC1, MYH9, MYO1E, MYOCD, NADSYN1, NEIL1, NEK1, NEK8, NHERF1, NIPBL, NLRP3, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NRIP1, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OSGEP, OXGR1, PAX2, PBX1, PDIA6, PDSS2, PHEX, PKD1, PKD2, PKHD1, PLCE1, PLVAP, PMM2, PODXL, PRDM15, PRKCSH, PRPS1, PTPRO, RCAN1, REN, RET, RMND1, ROBO1, ROBO2, ROR2, RPGRIP1L, RRAGD, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC61B, SEC63, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC26A3, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC7A9, SLIT2, SMARCAL1, SPP1, STRA6, STRADA, TBC1D1, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TNS2, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRIM8, TRPC6, TRPM6, TRPV5, TRPV6, TSC1, TSC2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VDR, VHL, VIPAS39, VPS33B, VTN, WBP11, WDPCP, WDR19, WDR35, WDR72, WDR73, WNK1, WNK4, WNT5A, WT1, XDH, XPNPEP3, XPO5, YRDC, ZIC3, ZMYM2, ZNF423

Alport-Syndrom v3 (5 Gene)

CD151, COL4A3, COL4A4, COL4A5, MYH9

Atypisches hämolytisch-urämisches Syndrom (aHUS) v8 (17 Gene)

ADAMTS13, C1GALT1C1, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, INF2, MMACHC, THBD, VTN

Autosomal-dominante tubulointerstitielle Nierenerkrankung (ADTKD) v4 (7 Gene)

DNAJB11, HNF1B, MT-TF, MUC1, REN, SEC61A1, UMOD*

**Die häufigste beschriebene pathogene Variante im MUC1-Gen (Duplikation im VNTR des Gens, NM_001204286.1: c.428dup) kann mittels Hochdurchsatzsequenzierung nicht nachgewiesen werden.*

Bardet-Biedl-Syndrom v7 (28 Gene)

ADIPOR1, AHI1, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP19, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP

Bartter-Syndrom v6 (26 Gene)

ATP1A1, BSND, CACNA1H, CLCN2, CLDN10, CYP11B1, HSD11B2, KCNJ5, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC26A3, CASR, CLCNKA, CLCNKB, FXYD2, HNF1B, KCNJ1, KCNJ10, KCNJ16, MAGED2, MT-TF, MT-TI, SLC12A1, SLC12A3

C3-Glomerulopathie v4 (11 Gene)

C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE

Diabetes insipidus v3 (3 Gene)

AQP2, AVP, AVPR2

Gitelman-Syndrom v7 (26 Gene)

ATP1A1, BSND, CACNA1H, CLCN2, CLDN10, CYP11B1, HSD11B2, KCNJ5, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC26A3, CASR, CLCNKA, CLCNKB, FXYD2, HNF1B, KCNJ1, KCNJ10, KCNJ16, MAGED2, MT-TF, MT-TI, SLC12A1, SLC12A3

Hyperoxalurie v3 (3 Gene)

AGXT, GRHPR, HOGA1

Hypophosphatämische Rachitis v4 (15 Gene)

ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, OCRL, PHEX, SLC34A1, SLC34A3, VDR

Nephrolithiasis, Nephrocalcinose und Urolithiasis v5 (64 Gene)

ADCY10, AGT, AGXT, ALPL, ANKH, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CAV1, CDC73, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, CYP24A1, FAM20A, FOXI1, G6PC, GCM2, GLA, GPHN, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, KCNJ5, KL, MAGED2, MOCOS, MOCS1, OCRL, OXGR1, PHEX, PRPS1, RRAGD, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC7A9, SLC9A3R1, SPP1, STRADA, TRPV5, TRPV6, UMOD, VIPAS39, VPS33B, WDR72, WNK4, XDH

Nephrotisches Syndrom (inkl. FSGS) v7 (87 Gene)

ACTN4, ALDH1A2, ALG1, AMN, ANLN, APOE, APOL1, ARHGAP24, ARHGDIA, AVIL, CD151, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DAAM2, DGKE, DLC1, EMP2, FAN1, FAT1, FN1, GAPVD1, GLA, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, KIRREL1, LAGE3, LAMA5, LAMB2, LCAT, LMX1B, LYZ, MAFB, MAGI2, MEFV, MT-TL1, MYH9, MYO1E, NEIL1, NOS1AP, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PMM2, PODXL, PRDM15, PTPRO, RCAN1, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, WDR73, WT1, XPO5, YRDC

Polyzystische Nierenerkrankung (ARPKD/ADPKD v4 (16 Gene)

ALG5, ALG8, ALG9, ANKS6, DNAJB11, DZIP1L, GANAB, HNF1B, IFT140, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63

Renale Dysplasie / Agenesie, CAKUT v4 (70 Gene)

ACE, ACTG2, AGT, AGTR1, ANOS1, BMP4, BNC2, CCNQ, CENPF, CEP55, CHD7, CHRM3, CHRNA3, CTU2, DHCR7, DSTYK, EXOC3L2, EYA1, FAM58A, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GPC3, GREB1L, GRIP1, HAAO, HNF1B, HOXA13, HPSE2, HS2ST1, ITGA8, JAG1, KDM6A, KMT2D, KYNU, LIFR, LRIG2, LRP4, MYOCD, NADSYN1, NIPBL, NOTCH2, NPHP3, NRIP1, PAX2, PBX1, PLVAP, REN, RET, ROBO1, ROBO2, ROR2, SALL1, SALL4, SIX1, SIX5, SLIT2, STRA6, TBC1D1, TBX18, TFAP2A, TMEM260, TRAP1, WBP11, WNT5A, ZIC3, ZMYM2

Renale tubuläre Acidose v4 (7 Gene)

ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, WDR72

Zystische Nierenerkrankungen v2 (58 Gene)

ALG5, ALG8, ALG9, ANKS6, BICC1, CEP164, CEP290, CEP83, COL4A1, COL4A4, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, EYA1, FLCN, GANAB, GLA, GLIS2, HNF1B, IFT140, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, MT-TF, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC61B, SEC63, SIX5, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, VHL, WDR19, XPNPEP3, ZNF423