Universitätsspital Basel

Medizinische Genetik
Labor Molekulargenetik

Schönbeinstrasse 40
CH-4031 Basel
Telefon +41 61 265 3620
Telefax +41 61 265 3621

www.unispital-basel.ch/medizinische-genetik

Genpanel Nephropathien

*= MLPA vorhanden

Alport-Syndrom

5 Gene: COL4A1, COL4A3*, COL4A4*, COL4A5*, MYH9*
DD: CFHR5*, COL4A6*, NPHS2,

Referenz: Plevova et al., Medicina, 2017; Clifford E Kashtan, GeneReviews, 21.02.2019 8https://www.ncbi.nlm.nih.gov/books/NBK1207)

Atypisches hämolytisch-urämisches Syndrom

13 Gene: C3, CD46, CFB, CFH, CFHR1, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, THBD, VTN
DD: ADAMTS13, INF2
Hybridallele werden nicht detektiert

Referenz: Genomic England panel app Atypical haemolytic uraemic syndrome (Version 2.15); Kavanagh et al., Seminars in Nephrology, 2013; Noris et al., GeneReviews, 23.09.2021 (https://www.ncbi.nlm.nih.gov/books/NBK1367)

Bartter-Syndrom

8 Gene: SLC12A1, KCNJ1, CLCNKA*, CLCNKB*, BSND, MAGED2, SLC12A3, CASR
DD: KCNJ10, CLDN10, SLC26A3, SCNN1A, SCNN1B, SCNN1G, NR3C2, HSD11B2, CYP11B1, CLCN2, KCNJ5, CACNA1H

Referenz: Fulchiero and Seo-Mayer, Pediatric Clinicl of North America, 2019; GeneReviews (06.09.2022, https://www.ncbi.nlm.nih.gov/books/NBK442019), Konrad et al., 2021 (PMID: 33509356)

Fokal segementale Glomerulosklerose

34 Gene: ACTN4, ADCK, AHRGDIA, ARHGAP24, CD2AP, COL4A3*, COL4A4*, COL4A5*, COQ2, COQ6, EYA1*, IGTB4, INF2, KANK4, LAMB2, LMNA*, LMX1B*, MYH9*, MYO1E, NPHS1, NPHS2, NUP203, NUP95, NXF5, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WDR73, WT1*
DD: ANLN, APOL1, COQ8B, CRB2, FBXW, KANK1, KANK2, LAMA5, NPHP1*, NPHP4, SYNPO, XPO5

Referenz: Rosenberg et Kopp, Clin J Am Soc Nephrol 12: 502–517, 2017. doi: 10.2215/CJN.05960616; Rood et al., Nephrol Dial Transplant (2012) 27: 882–890

Polyzystische Nierenerkrankung V1

7 Gene: ALG5, ALG9, DNAJB11, GANAB, IFT140, PKD1*, PKD2*
DD: ALG8, GANAB, LRP5, PRKCSH, SEC63, SEC61B, COL4A1, COL4A3, COL4A4, COL4A5, FLCN, HNF1B, MUC1, NOTCH2, OFD1, PKHD1, REN, SEC61A1, TSC1, TSC2, UMOD, VHL

Referenz: Harris PC, Torres VE. Polycystic Kidney Disease, Autosomal Dominant. 2002 Jan 10 [Updated 29.09.2022], www.ncbi.nlm.nih.gov/books/NBK1246)

CAKUT

58 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, ROBO2, ROR2, SALL1, SIX5, STRA6, TBC1D1, TBX18, TFAP2A, TMEM260, TRAP1, WBP11, ZIC3, ZMYM2

Referenz: CAKUT (Version 1.172, green) https://panelapp.genomicsengland.co.uk/panels/234/

Nierenerkrankungen (grosses Panel)

281 Gene: ACE, ACTG2, ACTN4, AGT, AGTR1, AGXT, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANOS1, AP2S1, APOA1, APOA2, APOC2, APOE, APRT, AQP2, ARHGDIA, ARL13B, ARL6, ARMC9, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BNC2, BSND, C3, C5orf42, CA2, CASR, CC2D2A, CD151, CD46, CENPF, CEP104, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, DAAM2, DDX59, DGKE, DHCR7, DLC1, DLG5, DNAJB11, DSTYK, DYNC2H1, DZIP1L, EMP2, EYA1, FAH, FAM20A, FAM58A, 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, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITSN1, JAG1, KCNJ1, KCNJ10, KCNJ5, KDM6A, KIAA0586, KIAA0753, KIF7, KLHL3, KMT2D, KYNU, LAGE3, LAMB2, LCAT, LIFR, LMX1B, LRIG2, LRP4, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MKKS, MKS1, MMACHC, MOCOS, MT-TF, MTX2, MUC1, MYH9, MYO1E, MYOCD, NADSYN1, NEK8, NIPBL, 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, REN, RET, ROBO2, ROR2, RPGRIP1L, RRM2B, SALL1, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, 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, TXNDC15, UMOD, VHL, VIPAS39, VPS33B, WBP11, WDPCP, WDR19, WDR35, WDR60, WDR73, WNK1, WNK4, WT1, XDH, XPNPEP3, YRDC, ZIC3, ZMYM2

Referenz: Renal superpanel – broad (Version 2.600, green) https://panelapp.genomicsengland.co.uk/panels/902

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.