Nierenerkrankungen

Panel-ID			Bearbeitungszeit
ID100	**Polyzystische Nierenerkrankung (PKD)**	**Multigen-Panel** (13 Gene) ANKS6, BICC1, DNAJB11, DZIP1L, GANAB, HNF1B, MUC1, NEK8, NPHP3, PKD1, PKD2, PKHD1, UMOD **Basis-Panel I (dominant)** (7 Gene) DNAJB11, GANAB, HNF1B, MUC1, PKD1, PKD2, UMOD **Basis-Panel II (rezessiv)** (5 Gene) ANKS6, DZIP1L, NEK8, NPHP3, PKHD1	3 - 5 Wochen
ID099	**Alport-Syndrom (ATS)**	**Multigen-Panel** (4 Gene) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 Wochen
ID156	**Bartter-Syndrom (BARTS)**	**Multigen-Panel** (8 Gene) BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3	3 - 5 Wochen
ID098	**Nephrotisches Syndrom (NPHS) und Fokal-segmentale Glomerulosklerose (FSGS)**	**Multigen-Panel** (43 Gene) ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, INF2, ITGA3, KANK1, KANK2, KANK4, LAMB2, LMX1B, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PLCE1, PTPRO, SGPL1, SMARCAL1, TPC1D8B, TRPC6, WT1 **Basis-Panel I (NPHS)** (10 Gene) COQ8A, EMP2, LAMB2, NPHS1, NPHS2, NUP93, NUP107, PLCE1, SGPL1, WT1 **Basis-Panel II (FSGS)** (9 Gene) ACTN4, ANLN, ARHGAP24, CRB2, CD2AP, INF2, MYO1E, PAX2, TRPC6 **Basis-Panel III (Alport-Syndrom)** (4 Gene) COL4A3, COL4A4, COL4A5, MYH9	4 - 6 Wochen
ID030	**Nephronophthise (NPHP)**	**Multigen-Panel** (21 Gene) ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 **Basis-Panel** (7 Gene) CEP290, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4	3 - 5 Wochen

Panel-ID			Bearbeitungszeit
ID029	**Senior-Loken-Syndrom (SLSN)**	**Multigen-Panel** (8 Gene) CEP290, NPHP1, NPHP3, IQCB1, NPHP4, SDCCAG8, TRAF3IP1, WDR19 **Basis-Panel** (7 Gene) CEP290, NPHP1, IQCB1, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wochen
ID093	**Bardet-Biedl-Syndrom (BBS)**	**Multigen-Panel** (21 Gene) ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP **Basis-Panel** (16 Gene) ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, IFT27, LZTFL1, MKKS, MKS1, TTC8, WDPCP	3 - 5 Wochen
ID028	**Joubert-Syndrom (JBTS)**	**Multigen-Panel** (35 Gene) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0556, KIAA0586, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423 **Basis-Panel I** (4 Gene) CC2D2A, CEP290, CPLANE1, TMEM67 **Basis-Panel II** (10 Gene) AHI1, B9D1, CC2D2A, INPP5E, KIAA0586, MKS1, RPGRIP1L, SUFU, TMEM138, TMEM216, TMEM67	3 - 5 Wochen
ID032	**Meckel-Syndrom (MKS)**	**Multigen-Panel** (13 Gene) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM231, TMEM216, TMEM67 **Basis-Panel** (9 Gene) CC2D2A, CEP290, MKS1, RPGRIP1L, TCTN2, TMEM107, TMEM231, TMEM216, TMEM67	3 - 5 Wochen
ID251	**Galloway-Mowat-Syndrom (GAMOS)**	**Multigen-Panel** (8 Gene) LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73	3 - 5 Wochen

Panel-ID			Bearbeitungszeit
ID250	**Pseudohypoaldosteronismus (PHA)**	**Multigen-Panel** (8 Gene) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wochen
ID103	**Glomerulonephritis**	**Multigen-Panel** (18 Gene) C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD **Basis-Panel** (15 Gene) C1QA, C1QB, C1QC, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, SOX18, THBD	3 - 5 Wochen
ID163	**Atypisches hämolytisch-urämisches Syndrom (AHUS)**	**Multigen-Panel** (16 Gene) ADAMTS13, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, THBD **Basis-Panel** (13 Gene) C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, DGKE, MMACHC, THBD	3 - 5 Wochen
ID229	**Kongenitale Anomalien der Niere und des Harntraktes (CAKUT)**	**Multigen-Panel** (50 Gene) ACE, AGT, AGTR1, BICC1, BMP4, BNC2, CDC5L, CHD1L, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, GRIP1, HNF1B, HPSE2, ITGA8*, KIF14, LIFR, LRIG2, LRP4, MUC1, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBC1D1, TBX18, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, WNT4 **Basis-Panel I** (11 Gene) CHD1L, DSTYK, HNF1B, NRIP1, PAX2, PBX1, SALL1, SIX2, TBC1D1, TBX18, TRAP1 **Basis-Panel II (Vesikoureteraler Reflux, VUR)** (7 Gene) HPSE2, LRIG2, PAX2, ROBO2, SOX17, TNXB, UPK3A **Basis-Panel III (Renale tubuläre Dysgenesie, RTD)** (4 Gene) ACE, AGT, AGTR1, REN **Basis-Panel IV (Brachiootorenales Syndrom, BOR)** (4 Gene) EYA1, SALL1, SIX1, SIX5 **Basis-Panel V (Fraser-Syndrom, FRASRS)** (3 Gene) FRAS1, FREM2, GRIP1 **Basis-Panel IV (Renale Hypodysplasie/Aplasie)** (10 Gene) DSTYK, FGF20, GATA3, GREB1L, HNF1B, ITGA8, PAX2, RET, UPK3A, WNT4	4 - 6 Wochen

Panel-ID			Bearbeitungszeit
ID231	**Nephrolithiasis und Nephrokalzinose**	**Multigen-Panel** (31 Gene) AGXT, APRT, ATP6V0A4, ATP6V1B1, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, G6PC, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH **Basis-Panel I (Hyperkalziurie)** (11 Gene) CASR, CLCN5, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, SLC12A1, SLC34A1, SLC34A3 **Basis-Panel II (Hyperoxalurie)** (4 Gene) AGXT, GRHPR, HOGA1, SLC26A1 **Basis-Panel III (Hypo-/Hyperurikosurie**) (7 Gene) APRT, G6PC, HPRT1, MOCOS, SLC2A9, SLC22A12, XDH **Basis-Panel IV (Hyperglycinurie)** (3 Gene) SLC6A19, SLC6A20, SLC36A2 **Basis-Panel V (Hypocitraturie)** (4 Gene) ATP6V0A4, ATP6V1B1, SLC4A1, SLC4A2 **Basis-Panel VI (Cystinurie) (**2 Gene) SLC3A1, SLC7A9	3 - 5 Wochen
ID041	**Nierenzellkarzinom**	**Multigen-Panel** (27 Gene) BAP1, DIRC2, DIS3L2, FH, FLCN, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PMS2, PBRM1, PTEN, RNF139, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TSC1, TSC2, VHL, WT1 **Basis-Panel I** (16 Gene) BAP1, DIRC2, FH, FLCN, HNF1A, HNF1B, MET, MITF, PTEN, RNF139, SDHB, SDHC, SDHD, SMARCB1, VHL, WT1 **Basis-Panel II (Lynch-Syndrom)** (4 Gene) MLH1, MSH2, MSH6, PMS2	3 - 5 Wochen

Kosten
Die Kosten werden bei bestehender medizinischer Indikation über einen Überweisungsschein Typ 10 (EBM) abgerechnet. Humangenetische Leistungen sind nicht budgetrelevant.
Es können bis zu 25 Kilobasen (kb) kodierende Sequenzen im Rahmen einer Stufendiagnostik abgerechnet werden (siehe ggf. Basispanels). Mutationssuchen in mehr als 25 kb unterliegen aktuell noch einer Genehmigungspflicht durch die gesetzliche Krankenversicherung.