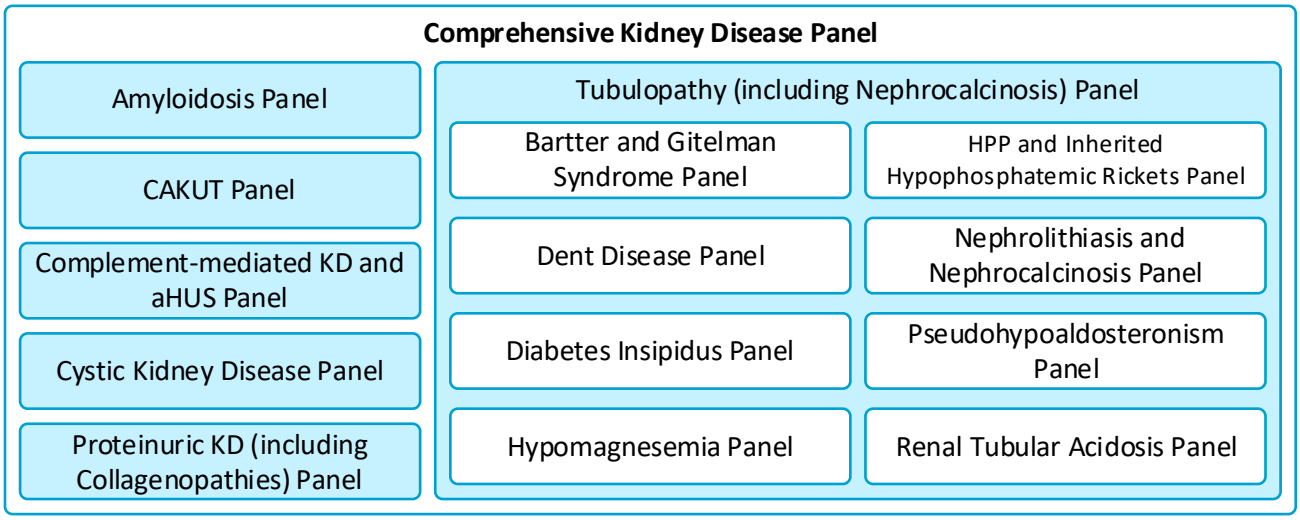
Chronic Kidney Disease Multigene Panels

The Chronic Kidney Disease Multigene Panels (Figure 1) were curated to include relevant genes associated with chronic kidney disease of genetic etiology. When choosing the most appropriate test for a patient and/or family, the test should be sufficiently comprehensive to encompass the genes associated with the differential diagnoses under consideration. The panels' structure and design should allow flexibility to the ordering clinician to select a more targeted or broad panel depending on the diagnostic certainty. The choice of panels should be driven by clinical judgement, informed by the patient phenotype, and dictated by disease certainty. For more information in guiding test selection and establishing eligibility see the **Chronic Kidney Disease Genetic Testing Recommendations.**

Figure 1. Chronic Kidney Disease Multigene Panels



aHUS, atypical hemolytic uremic syndrome; **CAKUT**, congenital anomalies of the kidney and urinary tract; **HPP**, hypophosphatemia; **KD**, kidney disease.

This figure illustrates the organization of gene panels for chronic kidney disease. The **Comprehensive Kidney Disease Panel** is the largest panel and encompasses several sub-panels, each shown in blue and outlined with rectangular boxes. A subset of these are grouped under the **Tubulopathy (including Nephrocalcinosis) Panel**, shown in blue outline with white boxes, which itself contains additional, more targeted sub-panels that clinicians can order based on specific clinical indications.

The panels should capture the coding regions and flanking intron/exon boundaries and identify relevant copy number variants (CNVs) of all genes under analysis. Select relevant intronic variants should be included for the genes listed in the panel.

Note that although *MUC1* is included on the Comprehensive Kidney Disease Panel, the most common pathogenic variants are not detectable using standard sequencing methods. If autosomal dominant tubulointerstitial kidney disease (ADTKD)-*MUC1* is clinically suspected and the results from the Comprehensive Kidney Disease Panel are negative or inconclusive, clinicians are advised to seek input from a kidney genetics expert regarding *MUC1* testing (e.g., via eConsult or referral to a local genetics clinic).

In addition, *APOL1* is included in both the Proteinuric Kidney Disease (including Collagenopathies) Panel and Comprehensive Kidney Disease Panel which can be offered to all patients, regardless of race or ethnicity. This approach accounts for inaccuracies of self-reported race and the limitations of using social constructs, such as race and ethnicity.

The Renal Genetics Expert Group followed an evidence-based framework for each panel to achieve consensus on which genes to include on the genetic kidney disease panels. ClinGen, Genomics England PanelApp, and/or PanelApp Australia curations may not be available for all the disease entities included in the molecular panels.

Table 1. Gene Contents for Kidney Disease Genetic Testing Panels

Panel (Number of genes)	Genes on Panel
Comprehensive Kidney Disease Panel (398)	Includes all genes currently on the disease-specific panels, plus the following genes associated with ciliopathies, and tubulointerstitial kidney disease: *ACE, ACTG2, ACTN4, AFF3, AGT, AGTR1, AGXT, AHI1, AIRE, ALG1, ALG5, ALG8, ALG9, ALMS1, ALPL, AMMECR1, AMN, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOA2, APOA4, APOC2, APOE, APOL1, APRT, AQP2, ARHGDIA, ARL13B, ARL6, ARMC9, ATP1A1, ATP6V0A4, ATP6V1B1, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BMP4, BNC2, BSND, C1GALT1C1, C2CD3, C3, C5, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CC2D2A, CCNQ, CD151, CD2AP, CD46, CDC73, CDKN1B, CDX2, CELSR3, CENPF, CEP104, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CILK1, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CPT2, CRB2, CSPP1, CST3, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27B1, CYP2R1, DAAM2, DCDC2, DDX59, DGKE, DHCR7, DLC1, DLG5, DMP1, DNAJB11, DSTYK, DYNC2H1, DYNC2I1, DYRK1A, DZIP1L, EHHADH, ENPP1, EXOC3L2, EYA1, FAH, FAM111A, FAM20A, FAM20C, FAN1, FAT1, FGA, FGF23, FLCN, FN1, FOXC1, FOXI1, FOXP1, FRAS1, FREM1, FREM2, FXYD2, GALNT3, GANAB, GATA3, GATM, GCM2, GDF6, GFRA1, GLA, GLI3, GLIS2, GNA11, GNAS, GON7, GPC3, GPNMB, GREB1L, GRHPR, GRIP1, GSN, HAAO, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HS2ST1, HSD11B2, HSD3B2, HSPA9, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT54, IFT74, INF2, INVS, IQCB1, ITGA3, ITGA8, ITSN1, ITSN2, JAG1, KANK2, KAT6B, KATNIP, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KDM2B, KDM6A, KIAA0586, KIAA0753, KIF14, KIF7, KLHL3, KMT2D, KYNU, LAGE3, LAMA5, LAMB2, LCAT, LIFR, LMX1B, LRIG2, LRP2, LRP4, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MAPKBP1, MEN1, MKKS, MKS1, MMACHC, MMUT, MOCOS, MT-TF, MTX2, MUC1[a], MYH9,*

[a] Although *MUC1* is included on the comprehensive kidney disease panel, the most common pathogenic variant is not detectable using standard sequencing methods. Rare *MUC1* variants may be detected and reported. If autosomal dominant tubulointerstitial kidney disease (ADTKD)-*MUC1* is clinically suspected and the results from the Comprehensive Kidney Disease Panel are negative or inconclusive, clinicians are advised to seek input from a kidney genetics expert regarding *MUC1* testing (e.g., via eConsult or referral to a local genetics clinic).

Panel (Number of genes)	Genes on Panel
	MYO1E, MYOCD, NADSYN1, NDUFAF6, NEK1, NEK8, NFIA, NIPBL, NLRP3, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPNT, NPR1, NR3C1, NR3C2, NR6A1, NRIP1, NUP107, NUP133, NUP160, NUP85, NUP93, OCRL, OFD1, OSGEP, OSMR, P3H2, PAN2, PAX2, PBX1, PCBD1, PDE3A, PDIA6, PDSS2, PHEX, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PLVAP, PMM2, PMPCA, PODXL, PRDM15, PRKCSH, PSKH1, PTH, PTH1R, PTPRO, REN, RET, RMND1, ROBO1, ROBO2, ROR2, RPGRIP1L, RRAGD, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SHROOM4, SLC12A1, SLC12A3, SLC20A1, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC7A7, SLC7A9, SMARCAL1, SON, STRA6, STRADA, STX16, TBC1D1, TBC1D8B, TBCE, TBX18, TBX6, TCTN1, TCTN2, TCTN3, TFAP2A, TMEM107, TMEM138, TMEM17, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TNS2, TOP2B, TP53RK, TPRKB, TRAP1, TRIM8, TRPC6, TRPM6, TRPM7, TSC1, TSC2, TSEN2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VDR, VIPAS39, VPS33B, WBP11, WDPCP, WDR19, WDR35, WDR44, WDR72, WDR73, WLS, WNK1, WNK4, WNT5A, WT1, XDH, XPNPEP3, YRDC, ZIC3, ZMYM2, ZNF423
Amyloidosis Panel (13)	*APOA1, APOA2, APOA4, APOC2, B2M, CST3, FGA, GPNMB, GSN, LYZ, NLRP3, OSMR, TTR*
CAKUT Panel (99)	*ACE, ACTG2, AFF3, AGT, AGTR1, ALMS1, ANOS1, BMP4, BNC2, CCNQ, CDX2, CELSR3, CENPF, CEP55, CHD7, CHRM3, CHRNA3, COL4A1, CTU2, DDX59, DHCR7, DLG5, DSTYK, DYRK1A, EXOC3L2, EYA1, FAT1, FLCN, FOXC1, FOXP1, FRAS1, FREM1, FREM2, GATA3, GDF6, GFRA1, GLI3, GPC3, GREB1L, GRIP1, HAAO, HNF1B, HOXA13, HPSE2, HS2ST1, HSPA9, ITGA8, JAG1, KAT6B, KDM2B, KDM6A, KIF14, KMT2D, KYNU, LIFR, LRIG2, LRP4, MYOCD, NADSYN1, NEK8, NFIA, NIPBL, NOTCH2, NPHP3, NPNT, NR6A1, NRIP1, OFD1, PAN2, PAX2, PBX1, PLVAP, REN, RET, RMND1, ROBO1, ROBO2, ROR2, SALL1, SALL4, SHROOM4, SLC20A1, SON, STRA6, TBC1D1, TBX18, TBX6, TFAP2A, TMEM260, TOP2B, TRAP1, TXNDC15, WBP11, WDR44, WLS, WNT5A, WT1, ZIC3, ZMYM2*
Complement Mediated Kidney Disease and Atypical Hemolytic Uremic Syndrome (aHUS) Panel (14)	*C1GALT1C1, C3, C5*[b]*, CD46, CFB, CFH*[c]*, CFHR1*[c]*, CFHR2*[c]*, CFHR3*[c]*, CFHR5*[c]*, CFI, DGKE, MMACHC, TSEN2*
Cystic Kidney Disease Panel (44)	*ALG5, ALG8, ALG9, ANKS6, C2CD3, CEP104, CEP164, CEP83, CLCN5, COL4A1, CYP24A1, DCDC2, DNAJB11, DZIP1L, FLCN, GANAB, GLA, GLIS2, HNF1B, IFT140, INVS, IQCB1, MAPKBP1, MKS1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, SEC63, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, WDR19, XPNPEP3*

[b] *C5* gene testing is not indicated for diagnosis but for treatment considerations testing can be considered as it may be associated with treatment resistance.

[c] Genomic rearrangement can occur in the *CFH* and *CFHR1-3, and CFHR5* in patients with aHUS. If complex rearrangements are suspected, additional assays are required including Multiplex Ligation-dependent Probe Amplification (MLPA).

Panel (Number of genes)	Genes on Panel
Proteinuric Kidney Disease (Including Collagenopathies) Panel (80)	*ACTN4, ALMS1, AMN, ANLN, APOA1, APOA2, APOA4, APOC2, APOE, APOL1, ARHGDIA, CD151, CD2AP, CLCN5, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CTNS, CUBN, DAAM2, DGKE, DLC1, FAT1, FN1, GLA, GON7, INF2, ITGA3, ITSN1, ITSN2, KANK2, LAGE3, LAMA5, LAMB2, LCAT, LMX1B, LRP2, MAFB, MAGI2, MTX2, MYH9, MYO1E, NOS1AP, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP85, NUP93, OCRL, OSGEP, P3H2, PAX2, PBX1, PDSS2, PLCE1, PODXL, PRDM15, PTPRO, REN, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, UMOD, WDR73, WT1, YRDC*
Tubulopathies (including Nephrocalcinosis) Panel (122)	*AGXT, AIRE, ALPL, AMMECR1, AP2S1, APRT, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BCS1L, BSND, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CDC73, CDKN1B, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, CPT2, CTNS, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27B1, CYP2R1, DMP1, EHHADH, ENPP1, FAH, FAM111A, FAM20A, FAM20C, FGF23, FOXI1, FXYD2, GALNT3, GATA3, GATM, GCM2, GLA, GNA11, GNAS, GRHPR, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, HSD3B2, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KLHL3, LCAT, LRP2, MAGED2, MEN1, MMUT, MOCOS, NDUFAF6, NR3C1, NR3C2, OCRL, PCBD1, PDE3A, PHEX, PTH, PTH1R, REN, RET, RMND1, RRAGD, RRM2B, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC7A7, SLC7A9, STRADA, STX16, TBCE, TRPM6, TRPM7, UMOD, VDR, VIPAS39, VPS33B, WDR72, WNK1, WNK4, XDH*
Dent Disease Panel (2)	*CLCN5, OCRL*
Diabetes Insipidus Panel (4)	*AQP2, AVP, AVPR2, STRADA*
Pseudohypoaldosteronism Panel (11)	*CUL3, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4*
Bartter and Gitelman Syndrome Panel (12)	*BSND, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, GNA11, HSD11B2, KCNJ1, MAGED2, SLC12A1, SLC12A3*
Renal Tubular Acidosis Panel (11)	*ATP6V0A4, ATP6V1B1, CA2, FOXI1, GATM, HNF4A, RMND1, SLC4A1, SLC4A4, VIPAS39, WDR72*
Hypomagnesemia Panel (17)	*ATP1A1, BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, FAM111A, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SARS2, SLC12A3, TRPM6, TRPM7*
Hypophosphatemia and Inherited Hypophosphatemic Rickets Panel (15)	*ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, OCRL, PHEX, SLC34A1, SLC34A3, VDR*

Panel (Number of genes)	Genes on Panel
Nephrolithiasis and Nephrocalcinosis Panel (41)	*AGXT, ALPL, AMMECR1, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CTNS, CYP24A1, FAM20A, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, OCRL, PHEX, PTH1R, RRAGD, SLC12A1, SLC22A12, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, STRADA, VDR, VIPAS39, VPS33B, WDR72, XDH*

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