



Testing is performed to evaluate for the presence of variants in coding regions and extending to +/-10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis may cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria. To verify if a specific region/exon/variant is covered by this assay or to confirm transcript version used, contact a laboratory genetic counselor at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ABCC6	NM_001171.5	-	CNV analysis in exons 1–9, 30–31 is not performed
ACE	NM_000789.4	-	-
ACTN4	NM_004924.6	chr:39138352C>T (c.-34C>T)	-
ADAMTS13	NM_139025.4	-	-
ADCY10	NM_018417.6	-	-
AGTR1	NM_031850.3	-	CNV analysis in exon 3 is not performed
AGXT	NM_000030.3	-	CNV analysis in exon 1 is not performed
AHI1	NM_001134831.2	-	-
ALG1	NM_019109.5	-	-
ALG8	NM_024079.5	-	-
ALG9	NM_024740.2 [GRCh38(hg38)]	-	-
ALMS1	ENST00000264448.6	-	-
ALPL	NM_000478.6	chr1:21896768_21896787del (c.793-30_793-11del)	-
ANKS6	NM_173551.5	-	CNV analysis in exon 1 is not performed
ANLN	NM_018685.5	-	-
ANOS1	NM_000216.4	-	-
AP2S1	NM_004069.5	-	-
APOA1	NM_000039.2	-	-
APOE	NM_000041.4	-	-
APOL1	NM_003661.4	APOL1 gene risk haplotype: G1 and G2 alleles	Analyses for sequence variants and CNV is not performed for the gene except for the specified risk variants.
APRT	NM_000485.3	-	-
AQP2	NM_000486.5	-	-
ARHGAP24	NM_001025616.3	-	CNV analysis in exon 10 is not performed
ARHGDIA	NM_001185077.3	-	-
ARL6	NM_032146.5	-	-
ARL13B	NM_182896.3	-	-
ATP6V0A4	NM_020632.3	-	-
ATP6V1B1	NM_001692.4	-	-
ATP7B	NM_000053.4	chr13:52586924-52585484 covers 5'UTR region	-
AVP	NM_000490.5	-	-

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>AVPR2</i>	NM_000054.6	-	-
<i>B9D1</i>	NM_015681.5	-	-
<i>B9D2</i>	NM_030578.4	-	-
<i>BBIP1</i>	NM_001195306.1	-	-
<i>BBS1</i>	NM_024649.5	-	-
<i>BBS10</i>	NM_024685.4	-	-
<i>BBS12</i>	NM_152618.3	-	-
<i>BBS2</i>	NM_031885.4	-	-
<i>BBS4</i>	NM_033028.5	-	-
<i>BBS5</i>	NM_152384.3	-	-
<i>BBS7</i>	NM_176824.3	-	-
<i>BBS9</i>	NM_198428.3	-	CNV analysis in exon 4 is not performed
<i>BICC1</i>	NM_001080512.3	-	-
<i>BSND</i>	NM_057176.3	-	-
<i>C2</i>	NM_000063.6	-	-
<i>C2CD3</i>	NM_001286577.1	-	-
<i>C3</i>	NM_000064.4	-	-
<i>C5</i>	NM_001735.3	Ecuzimab resistance-associated variants: c.2654G>A (p.Arg885His) c.2653C>T (p.Arg885Cys)	Analyses for sequence variants and CNV is not performed for the gene except for the specified variants.
<i>C8A</i>	NM_000562.3	-	-
<i>C8orf37</i>	NM_177965.4	-	-
<i>CA2</i>	NM_000067.3	-	-
<i>CACNA1D</i>	NM_000720.4	-	-
<i>CACNA1H</i>	NM_021098.3	-	-
<i>CASR</i>	NM_000388.4	-	-
<i>CC2D2A</i>	NM_001080522.2	-	-
<i>CD151</i>	NM_004357.5	-	-
<i>CD2AP</i>	NM_012120.3	-	CNV analysis in exon 17 is not performed
<i>CD46</i>	NM_002389.4	<i>CD46</i> (MCPggaac) risk haplotype	-
<i>CEP41</i>	NM_018718.3	-	CNV analysis in exon 4 is not performed
<i>CEP83</i>	NM_016122.3	-	-
<i>CEP104</i>	NM_014704.4	-	-
<i>CEP120</i>	NM_153223.3	-	-
<i>CEP164</i>	NM_014956.5	-	-
<i>CEP290</i>	NM_025114.4	chr12:88462434A>T (c.6012-12T>A)	CNV analysis in exons 5, 13, and 53 is not performed
<i>CFB</i>	NM_001710.6	-	-
<i>CFH</i>	NM_000186.4	<i>CFH</i> -H3 risk haplotype	Exon 20 CNV and <i>CFH</i> - <i>CFHR</i> hybrid alleles may not be detected
<i>CFHR1</i>	NM_002113.2	-	CNV analysis in exon 1 and 4 is not performed
<i>CFHR2</i>	NM_005666.4	-	CNV analysis in exon 1 is not performed
<i>CFHR3</i>	NM_021023.5	-	CNV analysis in exon 5 is not performed

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CFHR4</i>	NM_001201551.2	-	CNV analysis in exon 9 is not performed
<i>CFHR5</i>	NM_030787.4	-	-
<i>CFI</i>	NM_000204.4	-	CNV analysis in exon 7 is not performed
<i>CHD7</i>	NM_017780.4	chr8:61763035G>A (c.5405-17G>A)	-
<i>CLCN5</i>	NM_000084.5	-	-
<i>CLCNKA</i>	NM_004070.4	-	-
<i>CLCNKB</i>	NM_000085.5	-	CNV analysis in exon 20 is not performed
<i>CLDN16</i>	NM_006580.3	-	-
<i>CLDN19</i>	NM_148960.3	-	-
<i>CNNM2</i>	NM_017649.5	-	-
<i>COL4A1</i>	NM_001845.6	-	CNV analysis in exon 16 is not performed
<i>COL4A3</i>	NM_000091.5	-	-
<i>COL4A4</i>	NM_000092.5	-	-
<i>COL4A5</i>	NM_000495.5	-	-
<i>COL4A6</i>	NM_001847.4	-	CNV analysis in exon 1 is not performed
<i>COQ2</i>	NM_015697.8	-	-
<i>COQ6</i>	NM_182476.3	-	-
<i>COQ8B</i>	NM_024876.4	-	-
<i>CPLANE1</i>	NM_023073.3	-	CNV analysis in exon 44 is not performed
<i>CRB2</i>	NM_173689.7	-	-
<i>CREBBP</i>	NM_004380.3	-	-
<i>CSPP1</i>	NM_024790.6	-	CNV analysis in exons 4 and 26 is not performed
<i>CTNS</i>	NM_004937.3	-	-
<i>CUBN</i>	NM_001081.4	-	-
<i>CUL3</i>	NM_003590.5	-	-
<i>CYP11B1</i>	NM_000497.3	-	-
<i>CYP11B2</i>	NM_000498.3	-	CNV analysis in exon 9 is not performed
<i>CYP24A1</i>	NM_000782.5	-	CNV analysis in exons 6 and 9 is not performed
<i>CYP27B1</i>	NM_000785.4	-	-
<i>CYP2R1</i>	NM_024514.4	-	-
<i>DCDC2</i>	NM_016356.5	-	-
<i>DDX59</i>	NM_001031725.6	-	-
<i>DGKE</i>	NM_003647.3	chr17: g.54925466A>G (c.888+40A>G)	-
<i>DHCR7</i>	NM_001360.2	-	-
<i>DMP1</i>	NM_004407.4	-	CNV analysis in exon 3 is not performed
<i>DNAJB11</i>	NM_016306.5	-	-
<i>DYNC2H1</i>	NM_001080463.2	-	CNV analysis in exons 27 and 64 is not performed
<i>DZIP1L</i>	NM_173543.3	-	-
<i>EGF</i>	NM_001963.6	-	-
<i>EMP2</i>	NM_001424.6	-	-

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ENPP1</i>	NM_006208.3	-	CNV analysis in exons 10 and 22 is not performed
<i>EYA1</i>	NM_000503.6	-	-
<i>FAH</i>	NM_000137.3	-	-
<i>FAM20A</i>	NM_017565.4	-	-
<i>FAN1</i>	NM_014967.5	-	-
<i>FAT1</i>	NM_005245.4	-	-
<i>FGA</i>	NM_000508.5	-	-
<i>FGF20</i>	NM_019851.3	-	-
<i>FGF23</i>	NM_020638.3	-	-
<i>FGFR1</i>	NM_023110.3	-	-
<i>FGFR2</i>	NM_000141.4	-	-
<i>FN1</i>	NM_212482.3	-	-
<i>FOXI1</i>	NM_012188.5	-	-
<i>FOXP1</i>	NM_032682.5	-	CNV analysis in exon 9 is not performed
<i>FRAS1</i>	NM_025074.7	-	-
<i>FREM1</i>	NM_144966.5	-	-
<i>FREM2</i>	NM_207361.6	-	-
<i>FXYP2</i>	NM_001680.5	-	-
<i>FXYP2</i>	NM_021603.4	-	-
<i>GALNT3</i>	NM_004482.4	-	-
<i>GANAB</i>	NM_198335.4	-	-
<i>GATA3</i>	NM_001002295.2	-	-
<i>GLA</i>	NM_000169.2	chrX:100654735C>T (c.640-801G>A)	-
<i>GLI3</i>	NM_000168.6	-	-
<i>GLIS2</i>	NM_032575.2	-	-
<i>GNAI1</i>	NM_002067.5	-	-
<i>GPC3</i>	NM_004484.4	-	-
<i>GREB1L</i>	NM_001142966.2	-	CNV analysis in exons 3–33 is not performed
<i>GRHPR</i>	NM_012203.2	-	-
<i>GRIP1</i>	NM_021150.4	-	CNV analysis in exon 6 is not performed
<i>GSN</i>	NM_000177.5	-	CNV analysis in exon 16 is not performed
<i>HNF1B</i>	NM_000458.4	-	-
<i>HNF4A</i>	NM_175914.4	-	-
<i>HOGA1</i>	NM_138413.4	-	-
<i>HPRT1</i>	NM_000194.3	chrX:133594415C>T (c.27+47C>T)	CNV analysis in exon 5 is not performed
<i>HPSE2</i>	NM_021828.4	-	-
<i>HSD11B2</i>	NM_000196.4	-	-
<i>IFT27</i>	NM_006860.5	-	-
<i>IFT43</i>	NM_052873.3	-	-
<i>IFT80</i>	NM_020800.3	-	CNV analysis in exon 20 is not performed
<i>IFT81</i>	NM_014055.4	-	CNV analysis in exon 9 is not performed

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>IFT122</i>	NM_052985.4	-	-
<i>IFT140</i>	NM_014714.4	-	-
<i>IFT172</i>	NM_015662.3	-	-
<i>INF2</i>	NM_022489.4	-	-
<i>INPP5E</i>	NM_019892.6		CNV analysis in exons 4–5 is not performed
<i>INVS</i>	NM_014425.5	-	-
<i>IQCB1</i>	NM_001023570.4	-	CNV analysis in exon 7 is not performed
<i>ITGA3</i>	NM_002204.4	-	-
<i>ITGA8</i>	NM_003638.3	-	CNV analysis in exon 23 is not performed
<i>ITGB4</i>	NM_000213.5	-	-
<i>JAG1</i>	NM_000214.3	-	-
<i>KANK2</i>	NM_001136191.3	-	-
<i>KCNA1</i>	NM_000217.3	-	-
<i>KCNJ1</i>	NM_000220.5	-	-
<i>KCNJ5</i>	NM_000890.5	-	-
<i>KCNJ10</i>	NM_002241.5	-	-
<i>KIAA0556</i> (<i>KATNIP</i>)	NM_015202.4	-	-
<i>KIAA0586</i>	NM_001244189.2	-	CNV analysis in exons 6 and 33 is not performed
<i>KIAA0753</i>	NM_014804.3	-	-
<i>KIF7</i>	NM_198525.3	-	CNV analysis in exon 5 is not performed
<i>KIF12</i>	NM_138424.1	-	-
<i>KIF14</i>	NM_014875.3	-	CNV analysis in exon 7 is not performed
<i>KL</i>	NM_004795.4	-	-
<i>KLHL3</i>	NM_017415.3	-	-
<i>LAMA5</i>	NM_005560.6	-	-
<i>LAMB2</i>	NM_002292.4	-	-
<i>LMNA</i>	NM_170707.4	-	-
<i>LMX1B</i>	NM_002316.4	-	-
<i>LRIG2</i>	NM_014813.3	-	-
<i>LRP2</i>	NM_004525.3	-	-
<i>LRP5</i>	NM_002335.4	-	-
<i>LYZ</i>	NM_000239.3	-	-
<i>LZTFL1</i>	NM_020347.4	-	-
<i>MAGED2</i>	NM_177433.3	-	-
<i>MAGI2</i>	NM_012301.4	-	CNV analysis in exon 11 is not performed
<i>MAPKBP1</i>	NM_001128608.2	-	-
<i>MEFV</i>	NM_000243.2	-	-
<i>MKKS</i>	NM_018848.3	-	-
<i>MKS1</i>	NM_017777.4	-	-

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>MMACHC</i>	NM_015506.3	-	-
<i>MOCOS</i>	NM_017947.4	-	-
<i>MYH9</i>	NM_002473.5	-	-
<i>MYO1E</i>	NM_004998.4	-	-
<i>NEK1</i>	NM_001199397.2	-	CNV analysis in exons 13 and 36 is not performed
<i>NEK8</i>	NM_178170.3	-	-
<i>NLRP3</i>	NM_004895.4	-	-
<i>NOTCH2</i>	NM_024408.4	-	Analyses for sequence variants and CNV are not performed for exons 1–4
<i>NPHP1</i>	NM_000272.4	-	CNV analysis in exon 3 is not performed
<i>NPHP3</i>	NM_153240.5	-	CNV analysis in exons 8 and 22 is not performed
<i>NPHP4</i>	NM_015102.5	-	-
<i>NPHS1</i>	NM_004646.3	-	-
<i>NPHS2</i>	NM_014625.4	-	-
<i>NR3C2</i>	NM_000901.5	-	-
<i>NUP85</i>	NM_024844.5	-	-
<i>NUP93</i>	NM_014669.5	-	-
<i>NUP107</i>	NM_020401.4	-	CNV analysis in exons 18 and 20 is not performed
<i>NUP133</i>	NM_018230.3	chr1: 229577798A>T (c.3335-11T>A)	
<i>NUP160</i>	NM_015231.2	-	CNV analysis in exon 33 is not performed
<i>NUP205</i>	NM_015135.3	-	-
<i>OCRL</i>	NM_000276.4	-	CNV analysis in exons 3 and 16 is not performed
<i>OFD1</i>	NM_003611.3	-	-
<i>PAX2</i>	NM_003987.4	-	-
<i>PAX2</i>	NM_003988.5	-	-
<i>PBX1</i>	NM_002585.4	-	-
<i>PCBD1</i>	NM_000281.4	-	-
<i>PDE6D</i>	NM_002601.4	-	-
<i>PDSS2</i>	NM_020381.4	-	CNV analysis in exon 7 is not performed
<i>PHEX</i>	NM_000444.6	chrX:22266301A>G (c.*231A>G)	CNV analysis in exon 17 is not performed
<i>PKD1</i>	NM_001009944.3	-	CNV analysis in exons 1 and 26 is not performed
<i>PKD2</i>	NM_000297.4	-	CNV analysis in exon 9 is not performed
<i>PKHD1</i>	NM_138694.4	-	-
<i>PLCE1</i>	NM_016341.4	-	-
<i>PLCG2</i>	NM_002661.5	-	-
<i>PLG</i>	NM_000301.3	-	-
<i>PMM2</i>	NM_000303.3	chr16:8891573G>T (c.-167G>T) chr16: .8926102C>T (c.640-15479C>T)	
<i>PODXL</i>	NM_001018111.3	-	CNV analysis in exon 3 is not performed
<i>PRKCSH</i>	NM_002743.3	-	-

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>PRPS1</i>	NM_002764.4	-	-
<i>PTPRO</i>	NM_030667.3	-	CNV analysis in exon 18 is not performed
<i>REN</i>	NM_000537.4	-	CNV analysis in exon 8 is not performed
<i>ROBO2</i>	NM_002942.5	-	-
<i>RPGRIP1L</i>	NM_015272.5	-	CNV analysis in exon 23 is not performed
<i>SALL1</i>	NM_002968.3	-	-
<i>SALL4</i>	NM_020436.5	-	-
<i>SARS2</i>	NM_001145901.2	-	-
<i>SCARB2</i>	NM_005506.4	-	-
<i>SCLT1</i>	NM_144643.4	-	-
<i>SCNN1A</i>	NM_001038.6	-	-
<i>SCNN1B</i>	NM_000336.3	-	-
<i>SCNN1G</i>	NM_001039.4	-	-
<i>SDCCAG8</i>	NM_006642.5	-	CNV analysis in exon 7 is not performed
<i>SEC61A1</i>	NM_013336.4	-	-
<i>SEC63</i>	NM_007214.5	-	CNV analysis in exon 2 is not performed
<i>SGPL1</i>	NM_003901.4	-	-
<i>SIX1</i>	NM_005982.4	-	-
<i>SLC2A2</i>	NM_000340.2	-	-
<i>SLC2A9</i>	NM_020041.3	-	-
<i>SLC3A1</i>	NM_000341.4	-	-
<i>SLC4A1</i>	NM_000342.4	-	-
<i>SLC4A4</i>	NM_001098484.3	-	-
<i>SLC5A1</i>	NM_000343.4	-	-
<i>SLC5A2</i>	NM_003041.4	-	-
<i>SLC6A19</i>	NM_001003841.3	-	-
<i>SLC7A7</i>	NM_001126106.2	-	-
<i>SLC7A9</i>	NM_014270.5	-	-
<i>SLC9A3R1</i>	NM_004252.5	-	-
<i>SLC12A1</i>	NM_000338.3	-	-
<i>SLC12A3</i>	NM_000339.3	chr16: 56917770C>T (c.1670-191C>T)	-
<i>SLC17A5</i>	NM_012434.5	-	-
<i>SLC22A12</i>	NM_144585.4	-	-
<i>SLC26A1</i>	NM_213613.4	-	-
<i>SLC34A1</i>	NM_003052.5	-	-
<i>SLC34A3</i>	NM_080877.2	-	-
<i>SLIT2</i>	NM_004787.4	-	-
<i>SMARCAL1</i>	NM_014140.4	-	-
<i>TBC1D8B</i>	NM_017752.3	-	-

Targeted Genes and Methodology Details for Comprehensive Nephrology Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TBX18</i>	NM_001080508.3	-	-
<i>TCTN1</i>	NM_001082538.3	-	CNV analysis in exon 7 is not performed
<i>TCTN2</i>	NM_024809.5	-	-
<i>TCTN3</i>	NM_015631.6	-	-
<i>THBD</i>	NM_000361.3	-	-
<i>TMEM67</i>	NM_153704.6	-	-
<i>TMEM107</i>	NM_032354.5	-	-
<i>TMEM138</i>	NM_016464.5	-	-
<i>TMEM216</i>	NM_001173990.3	-	-
<i>TMEM231</i>	NM_001077416.2	-	-
<i>TMEM237</i>	NM_001044385.3	-	-
<i>TRAF3IP1</i>	NM_015650.4	-	-
<i>TRIM32</i>	NM_012210.3	-	-
<i>TRPC6</i>	NM_004621.6	-	CNV analysis in exon 10 is not performed
<i>TRPM6</i>	NM_017662.5	-	-
<i>TSC1</i>	NM_000368.5	-	-
<i>TSC2</i>	NM_000548.5	-	-
<i>TTC8</i>	NM_198309.3	-	-
<i>TTC21B</i>	NM_024753.5	-	-
<i>TTR</i>	NM_000371.3	-	-
<i>UMOD</i>	NM_003361.3	-	-
<i>VDR</i>	NM_001017535.1	-	-
<i>VHL</i>	NM_000551.3	-	-
<i>VIPAS39</i>	NM_022067.4	-	-
<i>VPS33B</i>	NM_018668.4	-	-
<i>WDR19</i>	NM_025132.4	-	CNV analysis in exon 3 is not performed
<i>WDR35</i>	NM_001006657.2	-	-
<i>WDR72</i>	NM_182758.4	-	CNV analysis in exon 6 is not performed
<i>WDR73</i>	NM_032856.4	-	-
<i>WNK1</i>	NM_018979.4	-	-
<i>WNK4</i>	NM_032387.5	-	-
<i>WNT4</i>	NM_030761.5	-	CNV analysis in exon 1 is not performed
<i>WT1</i>	NM_024426.6	-	-
<i>XDH</i>	NM_000379.4	-	-
<i>XPNPEP3</i>	NM_022098.4	-	-
<i>ZMPSTE24</i>	NM_005857.5	-	-
<i>ZNF423</i>	NM_015069.4	-	-