



Targeted Genes and Methodology Details for Nephrology Custom Gene Panel

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
<i>ABCC6</i>	NM_001171.5	-	CNV analysis in exons 1–9, 30–31 is not performed
<i>ACE</i>	NM_000789.4	-	-
<i>ACTN4</i>	NM_004924.6	chr:39138352C> T (c.-34C>T)	-
<i>ADAMTS13</i>	NM_139025.4	-	-
<i>ADCY10</i>	NM_018417.6	-	-
<i>AGTR1</i>	NM_031850.3	-	CNV analysis in exon 3 is not performed
<i>AGXT</i>	NM_000030.3	-	CNV analysis in exon 1 is not performed
<i>AHI1</i>	NM_001134831.2	-	-
<i>ALG1</i>	NM_019109.5	-	-
<i>ALG8</i>	NM_024079.5	-	-
<i>ALG9</i>	NM_024740.2 [GRCh38(hg38)]	-	-
<i>ALMS1</i>	ENST00000264448.6	-	-
<i>ALPL</i>	NM_000478.6	chr1:21896768_21896787del (c.793-30_793-11del)	-
<i>ANKS6</i>	NM_173551.5	-	CNV analysis in exon 1 is not performed
<i>ANLN</i>	NM_018685.5	-	-
<i>ANOS1</i>	NM_000216.4	-	-
<i>AP2S1</i>	NM_004069.5	-	-
<i>APOA1</i>	NM_000039.2	-	-
<i>APOE</i>	NM_000041.4	-	-
<i>APOL1</i>	NM_003661.4	<i>APOL1</i> 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.
<i>APRT</i>	NM_000485.3	-	-
<i>AQP2</i>	NM_000486.5	-	-
<i>ARHGAP24</i>	NM_001025616.3	-	CNV analysis in exon 10 is not performed
<i>ARHGDIA</i>	NM_001185077.3	-	-
<i>ARL6</i>	NM_032146.5	-	-
<i>ARL13B</i>	NM_182896.3	-	-
<i>ATP6V0A4</i>	NM_020632.3	-	-
<i>ATP6V1B1</i>	NM_001692.4	-	-
<i>ATP7B</i>	NM_000053.4	chr13:52586924-52585484 covers 5'UTR region	-
<i>AVP</i>	NM_000490.5	-	-
<i>AVPR2</i>	NM_000054.6	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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	Ecuzumab 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	CD46 (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 gene 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, 4 is not performed
<i>CFHR2</i>	NM_005666.4	-	CNV analysis in exon 1 is not performed

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CFHR3</i>	NM_021023.5	-	CNV analysis in exon 5 is not performed
<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, 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, 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, 64 is not performed
<i>DZIP1L</i>	NM_173543.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>EGF</i>	NM_001963.6	-	-
<i>EMP2</i>	NM_001424.6	-	-
<i>ENPP1</i>	NM_006208.3	-	CNV analysis in exons 10, 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>FOXP1c</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>FXVD2</i>	NM_001680.5	-	-
<i>FXVD2</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>GNA11</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	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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
<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, 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

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
MAPKBP1	NM_001128608.2	-	-
MEFV	NM_000243.2	-	-
MKKS	NM_018848.3	-	-
MKS1	NM_017777.4	-	-
MMACHC	NM_015506.3	-	-
MOCOS	NM_017947.4	-	-
MYH9	NM_002473.5	-	-
MYO1E	NM_004998.4	-	-
NEK1	NM_001199397.2	-	CNV analysis in exons 13, 36 is not performed
NEK8	NM_178170.3	-	-
NLRP3	NM_004895.4	-	-
NOTCH2	NM_024408.4	-	Analyses for sequence variants and CNV are not performed for exons 1–4
NPHP1	NM_000272.4	-	CNV analysis in exon 3 is not performed
NPHP3	NM_153240.5	-	CNV analysis in exons 8, 22 is not performed
NPHP4	NM_015102.5	-	-
NPHS1	NM_004646.3	-	-
NPHS2	NM_014625.4	-	-
NR3C2	NM_000901.5	-	-
NUP85	NM_024844.5	-	-
NUP93	NM_014669.5	-	-
NUP107	NM_020401.4	-	CNV analysis in exons 18, 20 is not performed
NUP133	NM_018230.3	chr1: 229577798A>T (c.3335-11T>A)	
NUP160	NM_015231.2	-	CNV analysis in exon 33 is not performed
NUP205	NM_015135.3	-	-
OCRL	NM_000276.4	-	CNV analysis in exons 3, 16 is not performed
OFD1	NM_003611.3	-	-
PAX2	NM_003987.4	-	-
PAX2	NM_003988.5	-	-
PBX1	NM_002585.4	-	-
PCBD1	NM_000281.4	-	-
PDE6D	NM_002601.4	-	-
PDSS2	NM_020381.4	-	CNV analysis in exon 7 is not performed
PHEX	NM_000444.6	chrX:22266301A>G (c.*231A>G)	CNV analysis in exon 17 is not performed
PKD1	NM_001009944.3	-	CNV analysis in exons 1, 26 is not performed
PKD2	NM_000297.4	-	CNV analysis in exon 9 is not performed
PKHD1	NM_138694.4	-	-
PLCE1	NM_016341.4	-	-
PLCG2	NM_002661.5	-	-
PLG	NM_000301.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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	-	-
<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	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SLIT2</i>	NM_004787.4	-	-
<i>SMARCA1</i>	NM_014140.4	-	-
<i>TBC1D8B</i>	NM_017752.3	-	-
<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	-	-