

## Targeted Genes and Methodology Details for Cystic Kidney Disease 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

ALG8NM_024702,5	Gene	<b>Reference Transcript</b>	Additional Evaluations	Technical Limitations
ALG9     [GRCh38(hg38)]     -     -       ANKS6     NM_173551.5     -     CNV analysis in exon 1 is not performed       BICC1     NM_001080512.3     -     -       CEP83     NM_016122.3     -     -       CEP104     NM_014704.4     -     -       CEP104     NM_014704.4     -     -       CEP104     NM_014956.5     -     -       CEP200     NM_025114.4     cht12:88462434A>T(c.6012·12T>A)     CNV analysis in exons 5, 13, and 53 is not performed       COL4A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCDC2     NM_001836.5     -     -       DXAJB11     NM_0016306.5     -     -       DZIP1L     NM_173543.3     -     -       GLI32     NM_032575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -       IQCB1     NM_0002357.4     -     - <td< td=""><td>ALG8</td><td>NM_024079.5</td><td>-</td><td>-</td></td<>	ALG8	NM_024079.5	-	-
BICC1     NM_001080512.3     -       CEP83     NM_016122.3     -     -       CEP104     NM_014704.4     -     -       CEP164     NM_014956.5     -     -       CEP200     NM_025114.4     chr12:88462434A>T (c.6012-12T>A)     CNV analysis in exons 5, 13, and 53 is not performed       CCL2A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CCL2A1     NM_016356.5     -     -       DCDC2     NM_016356.5     -     -       DCDC2     NM_016356.5     -     -       DZIP1L     NM_173643.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_0032575.2     -     -       INVS     NM_010425.5     -     -       INVS     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_0023357.4     -     -     -       IQCB1     NM_00123570.4     -     -     -       NMEX8     NM_178170.3     -     - <t< td=""><td>ALG9</td><td></td><td>-</td><td>-</td></t<>	ALG9		-	-
CEP83     NM_016122.3     -     -       CEP104     NM_014704.4     -     -     -       CEP164     NM_014956.5     -     -     -       CEP290     NM_025114.4     chr12:88462434A>T (c.6012-12T>A)     CNV analysis in exons 5, 13, and 53 is not performed       COL4A1     NM_01845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCCC2     NM_016326.5     -     -       DNAJB11     NM_016306.5     -     -       DXIPIL     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GANAB     NM_1032575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_001425.5     -     -       IQCB1     NM_0002357.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_002335.4     -     -     -       IRP5     NM_0012860.2     -     -     -       NME488     NM_173170.3 <td>ANKS6</td> <td>NM_173551.5</td> <td>-</td> <td>CNV analysis in exon 1 is not performed</td>	ANKS6	NM_173551.5	-	CNV analysis in exon 1 is not performed
CEP104     NM_014704.4     -     -       CEP164     NM_014956.5     -     -     -       CEP200     NM_025114.4     chrl2:88462434A>T (c.6012-12T>A)     CNV analysis in exon 5, 13, and 53 is not performed       COL4A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCDC2     NM_016356.5     -     -       DXAJB11     NM_016306.5     -     -       DXIP1L     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_002575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -       IQCB1     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_002357.4     -     -     -       IRP5     NM_0012357.4     -     -     -       NMP010128608.2     -     -     -     -       NMEVBP1     NM_00128608.2 </td <td>BICC1</td> <td>NM_001080512.3</td> <td>-</td> <td>-</td>	BICC1	NM_001080512.3	-	-
CEP164     NM_014956.5     -     -       CEP290     NM_025114.4     chrl2:88462434A>T (c.6012-12T>A)     CNV analysis in exon 5, 13, and 53 is not performed       COL4A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCDC2     NM_016356.5     -     -       DXAJB11     NM_016306.5     -     -       DXIPIL     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_002575.2     -     -       INVS     NM_014425.5     -     -       INVS     NM_014425.5     -     -       INVS     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_002355.4     -     -     -       IRP5     NM_00124.3     -     -     -       NKB8     NM_178170.3     -     -     -       NCTCH2     NM_002408.4     -     CNV analysis in exon 3 is not performed       NPHP1     NM	CEP83	NM_016122.3	-	-
CEP290     NM_025114.4     chr12:88462434A>T (c.6012-12T>A)     CNV analysis in exons 5, 13, and 53 is not performed       C0L4A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCDC2     NM_016356.5     -     -       DNAJB11     NM_016306.5     -     -       DZIP1L     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_0032575.2     -     -       INVS     NM_001425.5     -     -       IQC81     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00123570.4     -     -     -       IQC81     NM_00123570.4     -     -     -       IRP5     NM_00214.3     -     -     -       IRP5     NM_0012357.4     -     -     -       NOTCH2     NM_0024408.4     -     -     -       NOTCH2     NM_0024408.4     -     CNV analysis in exon 3 is not performed <td>CEP104</td> <td>NM_014704.4</td> <td>-</td> <td>-</td>	CEP104	NM_014704.4	-	-
COL4A1     NM_001845.6     -     CNV analysis in exon 16 is not performed       CRB2     NM_173689.7     -     -       DCDC2     NM_016356.5     -     -       DNAJB11     NM_016306.5     -     -       DZIP1L     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_032575.2     -     -       INVS     NM_014425.5     -     -       INVS     NM_014425.5     -     -       IQCB1     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00123570.4     -     -       IQCB1     NM_00124.3     -     -       NM_0128608.2     -     -     -       NK78     NM_178170.3     -     -       NOTCH2     NM_024408.4     -     CNV analysis in exon 3 is not performed       NPHP1     NM_000272.4     -     CNV analysis in exon 3 is not performed	CEP164	NM_014956.5	-	-
CRB2     NM_173689.7     -     -       DCDC2     NM_016356.5     -     -       DNAJB11     NM_016306.5     -     -       DZIP1L     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_032575.2     -     -       HNF1B     NM_004458.4     -     -       INVS     NM_014425.5     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_0012357.4     -     -     -       IQCB1     NM_00102357.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_0012357.4     -     -     -       IRP5     NM_00124.3     -     -     -       NMFBP1     NM_001128608.2     -     -     -       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	CEP290	NM_025114.4	chr12:88462434A>T (c.6012-12T>A)	CNV analysis in exons 5, 13, and 53 is not performed
DCDC2     NM_016356.5     -     -       DNAJB11     NM_016306.5     -     -     -       DZIP1L     NM_173543.3     -     -     -       GANAB     NM_198335.4     -     -     -       GLIS2     NM_032575.2     -     -     -       HNF1B     NM_00458.4     -     -     -       INVS     NM_014425.5     -     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_0012357.4     -     -     -       IQCB1     NM_00102357.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_0012357.4     -     -     -       IRP5     NM_002335.4     -     -     -       NMFBP1     NM_001128608.2     -     -     -       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	COL4A1	NM_001845.6	-	CNV analysis in exon 16 is not performed
DNAJB11     NM_016306.5     -       DZIP1L     NM_173543.3     -     -       GANAB     NM_198335.4     -     -       GLIS2     NM_032575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -       INVS     NM_01023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00123570.4     -     -       IQCB1     NM_00123570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_002355.4     -     -     -       IRP5     NM_001235608.2     -     -     -       NEK8     NM_178170.3     -     -     -       NOTCH2     NM_0024408.4     -     CNV analysis in exon 3 is not performed       NPHP1     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_153240.5     -     -     -       OFD1     NM	CRB2	NM_173689.7	-	-
DZIP1L     NM_173543.3     -       GANAB     NM_198335.4     -     -       GLIS2     NM_032575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -       INVS     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_000214.3     -     -       LRP5     NM_002335.4     -     -       MAPKBP1     NM_00128608.2     -     -       NTCH2     NM_024408.4     -     -       NOTCH2     NM_0024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1-4       NPHP1     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_153240.5     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	DCDC2	NM_016356.5	-	-
GANAB     NM_198335.4     -     -       GLIS2     NM_032575.2     -     -       HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_000214.3     -     -     -       LRP5     NM_002357.4     -     -     -       MAPKBP1     NM_000214.3     -     -     -       NM_D0123570.4     -     -     -     -       JAG1     NM_00214.3     -     -     -       MAPKBP1     NM_002335.4     -     -     -       MAPKBP1     NM_001128608.2     -     -     -       NEK8     NM_178170.3     -     -     -       NOTCH2     NM_024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1-4       NPHP1     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_153240.5     -     -     <	DNAJB11	NM_016306.5	-	-
GLIS2     NM_032575.2     -     -       HNF1B     NM_00458.4     -     -     -       INVS     NM_014425.5     -     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_002335.4     -     -       LRP5     NM_001128608.2     -     -       NEK8     NM_178170.3     -     -       NOTCH2     NM_00272.4     -     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_015102.5     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	DZIP1L	NM_173543.3	-	-
HNF1B     NM_000458.4     -     -       INVS     NM_014425.5     -     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_000214.3     -     -     -       LRP5     NM_0023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_00123570.4     -     -     -       LRP5     NM_00214.3     -     -     -       MAPKBP1     NM_002335.4     -     -     -       MAPKBP1     NM_001128608.2     -     -     -       NEK8     NM_178170.3     -     -     -       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_003611.3     -     -       OFD1     NM_003987.4     -     -	GANAB	NM_198335.4	-	-
INVS     NM_014425.5     -     -       IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_000214.3     -     -       LRP5     NM_002335.4     -     -       MAPKBP1     NM_001128608.2     -     -       NEK8     NM_178170.3     -     -       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 exon 3 is not performed       NPHP4     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	GLIS2	NM_032575.2	-	-
IQCB1     NM_001023570.4     -     CNV analysis in exon 7 is not performed       JAG1     NM_000214.3     -     -       LRP5     NM_002335.4     -     -       MAPKBP1     NM_001128608.2     -     -       NKK8     NM_178170.3     -     -       NOTCH2     NM_024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1-4       NPHP1     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_0153240.5     -     CNV analysis in exon 3 is not performed       NPHP4     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	HNF1B	NM_000458.4	-	-
JAG1     NM_000214.3     -     -       LRP5     NM_002335.4     -     -       MAPKBP1     NM_001128608.2     -     -       NEK8     NM_178170.3     -     -       NOTCH2     NM_024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1–4       NPHP1     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_153240.5     -     CNV analysis in exon 3 is not performed       NPHP4     NM_003611.3     -     -       OFD1     NM_003987.4     -     -	INVS	NM_014425.5	-	-
LRP5     NM_002335.4     -     -       MAPKBP1     NM_001128608.2     -     -       NEK8     NM_178170.3     -     -       NOTCH2     NM_024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1-4       NOTCH2     NM_00272.4     -     CNV analysis in exon 3 is not performed       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_003611.3     -     -       PAX2     NM_003987.4     -     -	IQCB1	NM_001023570.4	-	CNV analysis in exon 7 is not performed
MAPKBP1     NM_001128608.2     -     -       NEK8     NM_178170.3     -     -       NOTCH2     NM_024408.4     -     Analyses for sequence variants and CNV are not performed for exons 1-4       NOTCH2     NM_00272.4     -     CNV analysis in exon 3 is not performed       NPHP1     NM_000272.4     -     CNV analysis in exon 3 is not performed       NPHP3     NM_153240.5     -     CNV analysis in exon 3 is not performed       NPHP4     NM_015102.5     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	JAG1	NM_000214.3	-	-
NEK8     NM_178170.3     -     -       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     -     -       OFD1     NM_003987.4     -     -       PAX2     NM_003987.4     -     -	LRP5	NM_002335.4	-	-
NOTCH2NM_024408.4-Analyses for sequence variants and CNV are not performed for exons 1–4NPHP1NM_000272.4-CNV analysis in exon 3 is not performedNPHP3NM_153240.5-CNV analysis in exons 8, 22 is not performedNPHP4NM_015102.5OFD1NM_003611.3PAX2NM_003987.4	MAPKBP1	NM_001128608.2	-	-
NOTCH2     NM_024408.4     -     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     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	NEK8	NM_178170.3	-	-
NPHP3     NM_153240.5     -     CNV analysis in exons 8, 22 is not performed       NPHP4     NM_015102.5     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	NOTCH2	NM_024408.4	-	
NPHP4     NM_015102.5     -     -       OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	NPHP1	NM_000272.4	-	CNV analysis in exon 3 is not performed
OFD1     NM_003611.3     -     -       PAX2     NM_003987.4     -     -	NPHP3	NM_153240.5	-	CNV analysis in exons 8, 22 is not performed
PAX2 NM_003987.4	NPHP4	NM_015102.5	-	-
	OFD1	NM_003611.3	-	-
PAX2 NM_003988.5	PAX2	NM_003987.4	-	-
	PAX2	NM_003988.5	-	-

## Targeted Genes and Methodology Details for Cystic Kidney Disease Gene Panel (continued)

Gene	<b>Reference Transcript</b>	Additional Evaluations	<b>Technical Limitations</b>
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	-	-
RPGRIP1L	NM_015272.5	-	CNV analysis in exon 23 is not performed
SDCCAG8	NM_006642.5	-	CNV analysis in exon 7 is not performed
SEC61A1	NM_013336.4	-	-
SEC63	NM_007214.5	-	CNV analysis in exon 2 is not performed
TMEM67	NM_153704.6	-	-
TRAF3IP1	NM_015650.4	-	-
TSC1	NM_000368.5	-	-
TSC2	NM_000548.5	-	-
TTC21B	NM_024753.5	-	-
UMOD	NM_003361.3	-	-
WDR19	NM_025132.4	-	CNV analysis in exon 3 is not performed
WDR35	NM_001006657.2	-	-
XPNPEP3	NM_022098.4	-	-