MAYO CLINIC LABORATORIES | *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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ALG8	NM_024079.5	-	-
ALG9	NM_024740.2 [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	-	-
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_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	-	-
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_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_015102.5	-	-
OFD1	NM_003611.3	-	-
PAX2	NM_003987.4	-	-
PAX2	NM_003988.5	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	-	-