



*Targeted Genes and Methodology Details  
for Focused Autosomal Dominant  
Polycystic Kidney Disease 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

<b>Gene</b>	<b>Reference Transcript</b>	<b>Technical Limitations</b>
<i>ALG8</i>	NM_024079.5	-
<i>ALG9</i>	NM_024740.2 [GRCh38(hg38)]	-
<i>DNAJB11</i>	NM_016306.5	-
<i>GANAB</i>	NM_198335.4	-
<i>HNF1B</i>	NM_000458.4	-
<i>PKD1</i>	NM_001009944.3	CNV analysis in exons 1, 26 is not performed
<i>PKD2</i>	NM_000297.4	CNV analysis in exon 9 is not performed
<i>UMOD</i>	NM_003361.3	-