MAYO CLINIC LABORATORIES

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

Gene	Reference Transcript	Additional Evaluations
ALG8	NM_024079.5	-
ALG9	NM_024740.2 [GRCh38(hg38)]	-
DNAJB11	NM_016306.5	-
GANAB	NM_198335.4	-
HNF1B	NM_000458.4	-
PKD1	NM_001009944.3	CNV analysis in exons 1 and 26 is not performed
PKD2	NM_000297.4	CNV analysis in exon 9 is not performed
UMOD	NM_003361.3	-