MAYO CLINIC LABORATORIES

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

The following applies to ADPKP/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 will 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.

This test was updated August 2022. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered on this version or prior versions of this test, contact the laboratory at 800-533-1710.

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	-	-

Effective Date	Version	Synopsis of Test Change
9/2/2025	V2	Added Technical Limitations and Additional Evaluations