

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 9/1/1964
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing (Temporary Referral as of 06/09/20)

ARUP test code 2012255

ADPKD Sequencing Specimen whole blood

ADPKD Sequencing Interpretation

Negative

TEST PERFORMED - 2012255
TEST DESCRIPTION - Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the PKD1 or PKD2 genes.

INTERPRETATION
No pathogenic PKD1 or PKD2 gene variants were detected through bidirectional sequencing of the coding regions and intron-exon boundaries. This result significantly reduces the possibility of, but does not exclude, a diagnosis of autosomal dominant polycystic kidney disease (ADPKD). Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. If clinical suspicion for ADPKD remains, consideration should be given to PKD1 and PKD2 deletion/duplication analysis (ARUP test code 2012246). Genetic consultation is recommended.

COMMENTS
Reference Sequences: GenBank # NM_001009944.2 (PKD1), NM_000297.3 (PKD2)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [REDACTED]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing

CHARACTERISTICS: Autosomal Dominant Polycystic Kidney Disease (ADPKD) is typically an adult-onset, multisystem disorder. Renal findings include: bilateral renal cysts, renal insufficiency, renal pain, hypertension, dilated renal tubules, enlarged kidneys, and end-stage renal disease (ESRD). Extra-renal findings include cysts in other organs, including liver, pancreas, seminal vesicles, and arachnoid membrane. Connective tissue findings include intracranial aneurysms, dolichoectasia, dilation of the aortic root, aortic dissections, mitral valve prolapse, and abdominal wall hernias. Fifty percent of individuals with ADPKD will develop ESRD by age 60.

PREVALENCE: 1:500-1:1,000 in the U.S.

INHERITANCE: Autosomal dominant; 5-10 percent of cases are de novo.

PENETRANCE: Age-dependent; nearly all older adults develop multiple renal cysts. The average age of onset for ESRD in individuals with PKD1 and PKD2 mutations is 54 and 74 years, respectively.

CAUSE: Pathogenic PKD1 or PKD2 gene mutations. In cases with an identifiable molecular cause, 85 percent are attributed to PKD1 and 15 percent are attributed to PKD2.

CLINICAL SENSITIVITY: Estimated at 87 percent for ADPKD.

METHODOLOGY: Bidirectional sequencing of the entire coding region and intron/exon boundaries of the PKD1 and PKD2 genes. A large region of PKD1, including exons 1-33, is duplicated six times on the same chromosome; therefore, to distinguish the PKD1 gene from the PKD1-like pseudogenes, long range PCR followed by site-specific PCR is used to sequence PKD1 exons 1-33.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Large deletions/duplications, regulatory region mutations and deep intronic mutations in PKD1 or PKD2 will not be detected. Mosaic mutations in PKD1 or PKD2 may not be detected. Mutations in genes other than PKD1 and PKD2 are not assessed by this assay.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
ADPKD Sequencing Specimen	20-153-402286	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ADPKD Sequencing Interpretation	20-153-402286	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 20-153-402286
Patient Identifiers: 01234567890ABCD, 012345
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