

# AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE (ARPKD)

Target Mutation Analysis -**Test 2**

- updated 08-10-09 -

## DESCRIPTION

Mendelian Inheritance in Man number: [\\*606702](#)

Click here for [Gene Reviews](#) Clinical Summary.

Autosomal Recessive Polycystic Kidney Disease (ARPKD) is characterized by enlarged cystic kidneys and hepatic fibrosis. The diagnosis is often made pre- or neonatally, but some patients are still diagnosed later in life. The severity varies widely, with a high mortality rate in the first months of life. ARPKD is one of the more common hereditary childhood nephropathies with an estimated incidence of 1:20,000-1:40,000. The carrier frequency in the general population is estimated to be 1 in 70 to 1 in 100. Mutations in *PKHD1* are scattered throughout the gene. Most families carry their own "private" mutations. For more information on the condition please refer to the review on the [GeneTests](#) website and [Online Mendelian Inheritance in Man](#).

## Genetics of ARPKD

The gene for ARPKD, *PKHD1* (*Polycystic Kidney and Hepatic Disease 1*), resides on chromosome 6p21-p12, spans 470 kb of genomic DNA and is the only gene known to be associated with the wide clinical spectrum of autosomal recessive polycystic kidney disease. 86 exons have been identified and multiple alternative transcripts are known. Over 300 mutations have been reported. Missense, nonsense, frameshift, splicing and multi-exon deletions can occur and the mutations are located throughout the length of the gene, with no major mutational hotspots known, as shown in the [PKHD1 mutation database](#).

## INDICATIONS FOR DIRECT TESTING

- Carrier testing by sequence analysis of 1 or 2 exons in relatives of patients with known mutations
- Confirmation of mutations identified in a research lab by direct sequencing of 1 or 2 exons.
- Carrier testing by sequence analysis of 1 or 2 exons in the parents of patients with known mutations. **Parental testing is performed free of charge, provided that parental samples are submitted the same week as the sample of the proband undergoing Test 1.**

## TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *PKHD1* mutation(s) within the family. From a fresh EDTA blood sample, DNA is extracted directly, the target region(s) is amplified and the sequence is analyzed for family-specific mutations.

## SPECIMEN REQUIREMENTS

We require 5 milliliters of whole blood. Blood samples must be collected in EDTA (purple topped) tubes.

## TRANSPORT

If specimen is from clinics within UAB or Kirklin Clinic, please call 934-7107 for pick-up. If specimens are being sent from some other location, please ship via UPS or Federal Express.

1. Be sure that the shipping air bill is marked “**Priority**”, either Domestic or International.
2. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping companies Diagnostic Envelope). **You can use our collection kits, which we will send to physicians directly upon request.**

## TURN AROUND TIME

2 weeks

## CPT CODES AND PRICES

**Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.**

### One Exon:

\$190, - USD ([currency converter](#))

83891 (x1), 83898 (x4), 83904 (x3), 83912 (x1)

### Two Exons:

\$340, - USD

83891 (x1), 83898 (x8), 83904 (x6), 83912 (x1)

## REQUIRED FORMS

[ARPKD Test Requisition including the phenotypic data form](#)

**Note:** Requests for Molecular Genetic testing for ARPKD will **not** be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician’s or genetic counselor’s names and addresses
- No billing information
- No informed consent

**For more information, test requisition forms, or sample collection and mailing kits, please call: 205-934-5562.**

## REFERENCES

Bergmann C, Senderek J, Kupper F, Schneider F, Dornia C, Windelen E, Eggermann T, Rudnik-Schoneborn S, Kirfel J, Furu L, Onuchic LF, Rossetti S, Harris PC, Somlo S, Guay-Woodford L, Germino GG, Moser M, Buttner R, Zerres K. (2004) PKHD1 mutations in autosomal recessive polycystic kidney disease (ARPKD). *Hum Mutat.* 2004 May;23(5):453-63 ([pubmed](#))

Sharp AM, Messiaen LM, Page G, Antignac C, Gubler MC, Onuchic LF, Somlo S, Germino GG, Guay-Woodford LM. (2005). Comprehensive genomic analysis of PKHD1 mutations in ARPKD cohorts. *J. Med. Genet.* Apr; 42(4): 336-49. ([pubmed](#))

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