

FOR LAB USE ONLY
ACCESSION NO: _____
DATE & TIME RECEIVED: _____
TECHNICIAN: _____

**UAB MEDICAL GENOMICS LABORATORY**  
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**AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE TEST REQUEST FORM**

**THIS FORM AND PHENOTYPIC CHECKLIST MUST BE FILLED OUT COMPLETELY**

PATIENT NAME:		BIRTH DATE:	DAYTIME PHONE:	SEX:	SOC. SEC. NUMBER:
PATIENT'S ADDRESS:		CITY:	STATE:	ZIP CODE:	MED REC NUMBER:
EMAIL ADDRESS:			PARENT OR GUARDIANS NAME (IF MINOR):		
<b>PKHD1 SEQUENCE ANALYSIS</b>		<b>LINKAGE TESTING</b>		Physician's Name:	
<input type="checkbox"/> Complete Sequence and Copy Number Analysis (Test 1): Tier 1 followed by Tier 2 and Tier 3, as needed <input type="checkbox"/> Complete Sequence Analysis (Test 1): Tier 1 followed by Tier 2, as needed <input type="checkbox"/> <b>RUSH?</b> (Tier 1 and Tier 2 only) <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> (Tier 3) <input type="checkbox"/> Please check if parental samples are submitted same week as proband <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____ <input type="checkbox"/> Prenatal Targeted Analysis (Test 3) Proband _____		<input type="checkbox"/> DNA Isolation & Storage <input type="checkbox"/> Informativity (Test 4) <input type="checkbox"/> Prenatal (Test 5)		Physician's Address: _____ _____ _____ Phone: _____ Fax: _____ NPI Number: _____ Email: _____	
Facility where specimen obtained:					
Date:					
<b>REQUIRED</b> Diagnosis (ICD-9) Code (only in US):			ADDITIONAL REPORTS TO:		
Is Patient Pregnant?			Name:		
<input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP: _____		Please check if applicable:		Mailing Address:	
		<input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc)		_____ _____	
Specimen type:			Phone No: _____ Fax No: _____		
<input type="checkbox"/> Cheek Swabs; # Swabs: _____		<input type="checkbox"/> Amniotic Fluid		<input type="checkbox"/> Cultured CVS	
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes: _____		<input type="checkbox"/> Direct CVS		<input type="checkbox"/> Cultured Amniocytes	
<input type="checkbox"/> Other: _____					

**BILLING INFORMATION**

BILL INSTITUTION: (Please, provide name, address, and telephone number of entity responsible for payment)

Purchase Order Number: \_\_\_\_\_ Contact Name: \_\_\_\_\_  
 Billing address: \_\_\_\_\_ Phone #: \_\_\_\_\_  
 \_\_\_\_\_ Fax #: \_\_\_\_\_  
 \_\_\_\_\_ Email: \_\_\_\_\_

PAYMENT ENCLOSED:

- Cashier's Check
- VISA®  MasterCard®  Discover®  American Express®

Card Number: \_\_\_\_\_ Expiration Date: \_\_\_\_\_  
 Name as it appears on card: \_\_\_\_\_ 3-digit Security Code: \_\_\_\_\_  
 Cardholder Signature: \_\_\_\_\_  
 Cardholder Email Address: \_\_\_\_\_

BILL CONTRACTED INSURANCE COMPANY:

Please include a copy of patient's insurance card. For a list of our contracted insurance companies, please visit our website at [www.genetics.uab.edu/medgenomics](http://www.genetics.uab.edu/medgenomics), under "Billing". Please include a copy of pre-approval statement if payment has been authorized. **We also need the patient's credit card information so that any balance left after insurance pays may be applied to it. The RUSH fee must be paid up front by the patient.**

FILE INSURANCE CLAIM WITH NON CONTRACTED COMPANY

Patient must pay full payment for test up front, (credit card or cashier's check) but UAB will file a claim for reimbursement with the patient's insurance company. Please send a copy of patient's insurance card, front and back. **The RUSH fee must be paid up front by the patient.**

**INFORMED CONSENT  
FOR AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE MOLECULAR TESTING**

We have discussed clinical molecular genetic testing for autosomal recessive polycystic kidney disease (ARPKD) as it applies to me and/or my family with my doctor and/or a genetic counselor. We have been given the opportunity to discuss pertinent aspects of the genetic testing and to ask questions.

We understand that there are currently two distinct testing methodologies used for molecular testing for ARPKD. A **haplotype-based molecular genetic test** for ARPKD may not provide definitive information on whether our child possesses the disease gene or if we each carry an altered copy of the gene. We understand that the haplotype-based molecular genetic test is dependent on a confirmed clinical diagnosis of ARPKD and cannot be used to substantiate a clinical diagnosis of ARPKD.

If a confirmed clinical diagnosis is unable to be made, **direct gene sequencing of the PKHD1 gene** is available. We understand that direct gene sequencing detects about 80% of mutations per allele. Thus, in ~95% of cases at least one mutation will be identified substantiating the diagnosis of ARPKD. If only one clearly pathogenic mutation is found in the proband, haplotype-based molecular testing still can be performed in the family allowing prenatal testing in the future, if desired.

We have discussed both of these testing methodologies with our physician/genetic counselor to determine which is most appropriate. We understand that it is important to provide accurate information about our biological family members with regard to whether they have been diagnosed with ARPKD or have undertaken previous genetic testing for polycystic kidney disease (PKD).

Our signatures on this form indicate that we freely and voluntarily consent to have clinical molecular genetic testing performed for the ARPKD gene and give my permission to Dr. \_\_\_\_\_ to forward a sample of blood, paraffin-embedded tissue, whole tissue, cultured cells, amniotic fluid, or chorionic villus tissues to the MGL for such testing. I authorize MGL to report the test results to the physician or genetic counselor named below).

\_\_\_\_\_  
Subject's Signature Date

\_\_\_\_\_  
Physician's Signature Date

\_\_\_\_\_  
Please Print Subject's Name

\_\_\_\_\_  
Please Print Physician's Name

\_\_\_\_\_  
Assent of Parent Date

\_\_\_\_\_  
Genetic Counselor's Signature Date

\_\_\_\_\_  
Assent of Child Date

\_\_\_\_\_  
Please Print Genetic Counselor's Name

**ARPKD - CLINICAL CHECKLIST MEDICAL GENOMICS LABORATORY**

- Updated 03-02-08 -

Phenotypic data reflect the clinical situation in the patient as per date \_\_ / \_\_ / \_\_

Patient name: \_\_\_\_\_

Referring physician: \_\_\_\_\_

A COPY OF THE **HISTOPATHOLOGY REPORT** OF THE AFFECTED PATIENT IS NEEDED TO HELP ADVISE THE MOST APPROPRIATE TYPE OF TESTING FOR THE PKHD1 GENE  
ALWAYS PROVIDE **ETHNIC BACKGROUND**

**I. DEMOGRAPHIC INFORMATION**

- I.1. Gender:  Male  
 Female
- I.2. Date of Birth: \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_
- I.3. Date of Diagnosis: \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_
- I.4. Date of Death (if applicable): \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_
- I.5. Consanguinity:  **yes (please provide details if possible)**  
 no  
 unknown
- I.6. Ethnicity:      Father: \_\_\_\_\_  
                          Mother: \_\_\_\_\_
- I.7. Family History of PKD:  Yes  No  Unknown

For *familial* cases only: draw pedigree and indicate (1) Affected or (2) Unaffected

**II. DIAGNOSIS**

**II.1. PKD-Associated Morbidity**

	Yes	No
Decreased amniotic fluid		
Before 20 weeks gestation		
After 20 weeks gestation		
Kidney related disease		
Hyponatremia		
Hypertension		
Chronic renal insufficiency		
Liver related disease		
Portal hypertension		
Variceal bleeding		
Cholangitis		
Other		
Ventilation		
Chronic lung disease		
Other congenital anomalies		

## II.2 Initial Radiological Studies

### KIDNEY(if applicable)

Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

Imaging Study:

- Ultrasound  CT scan with contrast  CT scan without contrast  
 Other, specify:

- Result:  Echogenic kidneys without cysts  
 Echogenic kidneys with diffuse small cysts  
 Echogenic kidneys with gross cysts

### LIVER (if applicable)

Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

Imaging Study:  Ultrasound  CT scan  Other, specify:

- Result:  Normal  
 Echogenic liver  
 Dilated bile ducts

## II.3 Histopathology

### KIDNEY(if applicable)

Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

Pathology:  Autopsy  Open Biopsy  Needle Biopsy  Nephrectomy

- Result:  Dilated collecting ducts  
 Glomerular dilation  
 Dysplastic elements

### LIVER(if applicable)

Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

Pathology:  Autopsy  Open Biopsy  Needle Biopsy

- Result:  Normal  
 Billiary dysgenesis