

INFORMATION FOR POTENTIAL PARTICIPANTS

Autosomal recessive polycystic kidney disease (ARPKD) occurs in about 1 in 20,000 live births. Disease expression is widely variable with ~30% of affected infants dying in the newborn period, while others survive to adulthood. Mutations in single gene, *PKHD1*, are responsible for all typical forms of ARPKD.

The University of Alabama at Birmingham Recessive Polycystic Kidney Disease Core Center (UAB RPKDCC) has established a NIDDK-funded, interdisciplinary center of excellence in Polycystic Kidney Disease (PKD)-related research, with specific emphasis on recessive PKD (<http://www.rpkdcc.uab.edu/>). The UAB RPKDCC includes the “**Core A: The ARPKD Clinical and Genetic Resource**”, a Core resource designed to develop educational tools for physicians and patients regarding the natural history, pathogenesis, genetic testing, and clinical trials applicable to ARPKD.

“Core A: The ARPKD Clinical and Genetic Resource” is an internet-based data collection. The study web site is <http://www.arpkdstudies.uab.edu>. To protect the participant confidentiality, unique numerical identifiers will be used to enter and store all clinical and genetic information.

Core A has a clinical (medical information) and a genetic (DNA) component. The participant/representative may choose to participate in one or both of the components.

This study wants to establish a clinical database of patients with ARPKD to compare DNA information with medical information.

If you would like to enroll yourself or your child as a participant in Core A: the ARPKD Clinical and Genetic Resource please follow the instructions provided below.

INSTRUCTIONS:

- Click on [Consent](#) and download our Informed Consent Form.
- After you have reviewed the Informed Consent Form, please click on [Contact Information](#) login and complete the required fields, [submit](#) the information and an E-mail notification will automatically be sent to Teresa Chacana (Research Nurse Coordinator). You can also call Ms. Chacana (205-934-7649) or click on [Teresa Chacana](#) to send an e-mail indicating your interest in participating in this database.
- Ms. Chacana will contact you by phone to discuss the study and answer any questions that you may have regarding the Informed Consent Form.
- After you sign the Informed Consent Form please return it by US mail.
- Once Ms. Chacana receives the signed copy of the Informed Consent and your physician’s contact information, she will verify the contact information and then mail your physician a unique physician password, a unique identifier number for you/your child, and a request that the physician enter clinical data into the ARPKD Clinical Database using the web-based data entry forms.
- If you also provide permission to enter genetic information that was obtained in the UAB Molecular Genomics Laboratory, Dr. Ludwine Messiaen will use the same unique identifier assigned to you/your child to enter these data into the ARPKD Mutational Database.

Thank you for your interest!