ARPKD Database Study

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age: Up to 18 years old
Healthy Volunteers: This study is NOT accepting healthy volunteers
Inclusion Criteria:

- Demonstration of hepato/renal fibrocystic disease by clinical information, imaging studies, biopsy, autopsy, or genetic testing.

Exclusion Criteria:

- ADPKD Urinary tract malformations Major congenital anomalies of other systems

Conditions & Interventions

Conditions:
Hepato/Renal Fibrocystic Disease, Autosomal Recessive Polycystic Kidney Disease, Joubert Syndrome, Bardet Biedl Syndrome, Meckel-Gruber Syndrome, Congenital Hepatic Fibrosis, Caroli Syndrome, Oro-Facial-Digital Syndrome Type I, Nephronophthisis, Glomerulocystic Kidney Disease
Keywords:
cystic kidney disease, polycystic kidney disease, congenital hepatic fibrosis, genetic disease

More Information

Description:
Hepato-renal fibrocystic diseases (HRFD) is a term developed that encompasses rare diseases such as Autosomal Recessive Polycystic Kidney Disease (ARPKD), and other diseases with common features (Joubert syndrome, Bardet Biedl syndrome, Meckel-Gruber syndrome, congenital hepatic fibrosis (CHF), Caroli syndrome (CS), polycystic liver disease, oro-facial-digital syndrome, nephronophithisis (NPHP), and glomerulocystic Kidney Disease). The lack of enough routinely available resources for these diseases to be well diagnosed and treated, would be best resolved by coordinated case accrual and sharing of clinical data and bio-specimens (DNA and tissues) among participating institutions, thereby leading to the centralization and sharing of clinical and genetic information, as well as bio-materials, providing an important engine for more rapid research progress and community understanding through the creation of research networks. This study aims to build a registry of a clinical database (medical health information), a mutational database (genetic information) and an educational resource about HRFD to eventually provide information about these diseases to families, physicians and genetic counselors via our existing HIPAA- approved study website. Goals for the Core A: The Hepato/Renal Fibrocystic Diseases Translational Resource are: 1. \- Clinical Database: • Expand our comprehensive Clinical Database to include information from all patients who meet the inclusion criteria for hepato/renal fibrocystic diseases. 2. \- Mutational Database: * Test children with ARPKD and other hepato/renal fibrocystic disease to identify genetic mutations, establish a DNA bank for patients with hepato/renal fibrocystic diseases and develop a Mutational Database. This Database will be capable of linking clinical and mutational information via a unique identifier in a searchable format to facilitate genetic research (e.g. genotype-phenotype correlations, new disease gene studies, and modifier gene studies), translational studies, and clinical trials. 3- Tissue Resource: * Much of the research that is performed on diseases of the kidney, including recessive genetic diseases, requires human tissue from both affected as well as non-affected (controls) individuals. In this Core Resource, we are establishing an independent tissue resource which would supply investigators throughout North America with samples of hepato/renal fibrocystic disease affected tissues for studies of these disorders. 4- Educational Resource: * Expand our multi-media, web-based resource to provide a reliable up-to-date, and comprehensive informational resource for ARPKD and Hepato/Renal Diseases families, their physicians, and genetic counselors.

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