Children's Hospital of Philadelphia

Informed Consent and HIPAA Authorization Form

Study Title: Core A: The Hepato/Renal Fibrocystic Diseases Translational Resource

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You, or your child, may be eligible to take part in a research study. This form gives you important information about the study. It describes the purpose of this research study, and the risks and possible benefits of participating.

In the sections that follow, the word "we" means the study doctor and other research staff. If you are a parent or legal guardian who is giving permission for a child, please note that the word "you" refers to your child.

Study Overview

You or your child are being asked to take part in this research study because you have autosomal polycystic kidney disease (ARPKD) or another hepato/renal fibrocystic disease (HRFD).

ARPKD is a rare inherited disorder that occurs in 1 in 20,000 people. It is part of the Hepato/Renal fibrocystic diseases (HRFD) group. This disease affects boys and girls equally and affects the kidneys and liver. It occurs mainly in infants and children and causes serious health problems but can also occur in adults.

The purpose of this study is to get more information about ARPKD and other hepato/renal fibrocystic diseases. We also want to expand our web-based resources so anyone can learn about ARPKD or other hepato/renal fibrocystic diseases.

If you agree to take part, your participation will last for around two hours. It will involve one study visit and an annual follow-up till the end of the study or until you choose to end participation. As a participant in the research you will asked to participate in

- Medical history review
- Optional genetic testing by giving a blood or saliva sample
- Optional tissue sample
- Optional urine sample

Data and/or samples collected will be used to create a database that may help us to better understand this rare condition. We also hope to create resources that could be used by families, and their physicians and genetic counselors. There are no direct physical risks to this study. If you choose to take part in the optional genetic testing portion of the study, you can expect to experience some momentary pain, bleeding or bruising when your blood is drawn; the discomfort should be minimal. If clinically useful information is

found during genetic testing, we will ask your clinician to repeat the genetic testing in a clinically approved lab.

You will not benefit directly from participating in this study.

If there is anything in this form you do not understand, please ask questions. Please take your time. You do not have to take part in this study if you do not want to. If you take part, you can leave the study at any time.

Please see below for additional details about the study.

How many people will take part?

About 280 people will take part in the study, including approximately 40 participants from Children's Hospital of Philadelphia (CHOP).

What are the study procedures?

Tests that are part of your regular, routine medical care will continue to be performed. The study involves the following tests and procedures.

Medical history review: Information (data) will be collected from your medical records and from a brief interview. The information will include your diagnosis, treatments, and medications. If you are cared for at CHOP, your data will be updated every year until you turn 18 years of age as long as the study is on-going. If you are still cared for at CHOP after you turn 18, we will request that you continue to allow us to update your information. We will remove your name or any other identifiable health information (such as name, address) from your received records before entering your medical data into the Hepato/Renal Fibrocystic Diseases clinical database.

Optional Blood Sample and Genetic Testing: Up to 1 teaspoon of blood will be taken and stored in the biorepository. The amount of blood will depend upon your size.

If you are a CHOP patient, the sample will be collected when you are having blood drawn for your medical care. A separate blood draw may need to be done if we cannot coordinate the collection of the research sample with a clinical blood draw. We can also mail you the supplies and you can take it to the lab or doctor of your choice.

The sample will be stored in the BioRepository of Children's Hospital of Philadelphia. They will be tested to search for a genetic basis for ARPKD and other HRFD. The following genetic testing may be done on a research-only basis, without release of results to participants: exome sequencing, whole-genome sequencing, whole-genome mapping. Whole-genome sequencing is the analysis of the complete set of genetic instructions (DNA) in a cell. This analysis looks for small changes (sequence variants) in the genetic instructions. We will look for single nucleotide variants (SNVs), small deletions and insertions (INDELs) as well as large structural variants (SVs, deletions, insertions, translocations, and inversions). As recommended by the National Institutes of Health, the sequences (without your name or any further identifier) will be submitted to national databases to facilitate research into genetic causes of conditions.

Alternative Optional Saliva Sample for Genetic Testing: If you are interested in participating in the optional genetic testing but are unable or uncomfortable with the blood draw, you can give a saliva sample instead. You will be asked to spit some saliva in a cup. You should not eat, drink, smoke or chew gum for 30 minutes before collection. You should spit until the amount of saliva (not bubbles) reaches the fill line. However, children under the age of 4 might have difficulty providing half a tablespoon of saliva and may choose to instead complete the blood draw for optional genetic testing.

These saliva kits can also be mailed to your home for ease of collection and use.

Optional Tissue Sample: If you choose to participate, you will sign this consent form and will provide the information of the doctor where you will be having the kidney or liver tissue removed or where the autopsy will be performed. This doctor will collect tissue samples. CHOP will send all of the materials that will be needed to your doctor including: a mailer, a tissue collection kit, and instructions on how to collect and store the tissue. These tissue samples will be labeled with an identifier that is unique to you and will be sent to CHOP Tissue Repository to process and store.

The purpose of this is to make it easier for researchers to obtain rare resources such as liver and kidney liver tissue from hepato-renal fibrocystic disease patients to help further our knowledge about these diseases. When researchers are interested in getting tissue for their investigations, they will contact Dr. Lisa Guay-Woodford and will fill out a request form that will be then be reviewed by the Core A Scientific Advisory Committee. This Committee will determine whether to grant permission for tissue samples to be released from the tissue bank for specific research studies.

If you are a CHOP patient, the sample will be collected when you are having a sample collected for your medical care. We can also mail you the supplies and you can take it to the lab or doctor of your choice.

The sample will be stored in the BioRepository of the Children's Hospital of Philadelphia. The following may be done on a research-only basis.

Optional Urine Sample: A 24-hour urine collection is a simple lab test that measures what's in your urine. The test is used to check kidney function. A 24-hour urine collection is done by collecting your urine in a special container over a full 24-hour period.

If you are a CHOP patient, the sample will be collected when you are in clinic for your medical care.

The sample will be stored in the BioRepository of the Children's Hospital of Philadelphia. The following testing may be done on a research-only basis.

What will be done with my data and specimens during this study?

Research study team will be enter your coded medical data into the Hepato/Renal Fibrocystic Diseases clinical database.

Samples (if you choose to do optional testing) will be processed. Sample testing will be performed and stored at CHOP, until the close of study.

By agreeing to participate in the study, you agree to give these samples to CHOP for research purposes.

Will I receive any results from the tests done as part of this study?

Genetic testing done for this study will be done for research-puposes only and may not have valuable health care use. If we think that there are genetic research results that will be beneficial for you, we will contact you and/or your clinician. At that time we will recommend that these tests be re-done by a certified clinical laboratory. The study does not have the funding to complete such testing in a certified clinical laboratory. Consult your physician, professional genetic or other counselor. You or your insurance company will have to pay for those additional services.

What are the risks of this study?

Taking part in a research study involves inconveniences and risks. The main risks of taking part in this study are discussed below.

Risks of blood tests:

Taking blood may cause some pain, bleeding or bruising at the spot where the needle enters your body. Rarely, taking blood may cause fainting or infection. The genetic testing kits will be sent to you to draw blood at the lab of your choosing. If you choose to do the research blood draw at CHOP, it can be combined with clinical blood draws to minimize risk.

Risks of saliva sample:

There are no physical risks but you might experience momentary discomfort.

Risks of urine sample:

There are no physical risks.

Risks of tissue sample:

There are no physical risks since we would only be storing your tissue sample.

Risks to your personal privacy and confidentiality:

Research that uses health information from your medical record or that involves genetic testing can affect your privacy. Your participation in this research will be held strictly confidential and only a code number will be used to identify the stored samples and data. However, because there will be a link between the code and your identity, confidentiality cannot be guaranteed.

Risks of Genetic Studies:

The risks related to genetic analyses can be to individuals or groups. These harms include stigmatization and insurability. To reduce this risk, your samples will be stored and labeled with a code number. If the results are used for future research, the researchers will not be able to identify you. Information about this study will not be recorded in your medical record.

There is a Federal law, called the Genetic Information Nondiscrimination Act (GINA), which generally makes it illegal for health insurance companies, group health plans, and

most employers to discriminate against you based on your genetic information. This law may protect you in the following ways:

- Health insurance companies and group health plans may not request your genetic information that we get from this research.
- Health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums.
- Employers with 15 or more employees may not use your genetic information that we get from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment.

This Federal law does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

New information about parentage may be discovered by this research. This could include unknown adoption and paternity (fatherhood). These types of findings will not be shared with you unless there are medical concerns. We will not reveal this information to any third party, including other family members.

We may wish to share your data or DNA samples with other investigators or national databases. The NIH maintains a national database for genetic material. They collect samples for future research. The shared information will not include information that can identify you. The shared information will include information about your diagnosis and genes. If you withdraw consent for sharing, your information or samples that are still at CHOP will be removed and will no longer be used for future research. However, data and samples that have already been shared with other researchers cannot be taken back.

There may be other risks that are not known at this time. Tell the study investigator or study staff right away if you have any problems.

Are there any benefits to taking part in this study?

There are no direct benefits to you from participating in this study. An indirect benefit would be that results from this research may provide important insight for the future care of people with these conditions.

What about privacy, authorization for use of Personal Health Information (PHI) and confidentiality?

As part of this research, health information about you will be collected. This will include information from medical records, interviews, and research tests (if applicable). Medical records are available to CHOP staff. Staff will view your records only when required as part of their job. Staff are required to keep your information private. Information that could identify you will not be shared with anyone – unless you provide your written consent, or it is required or allowed by law. Genetic test results (if applicable) will not appear in your medical records. We will do our best to keep your personal information private and confidential. However, we cannot guarantee absolute confidentiality. Your personal information may be disclosed if required by law.

The results of this study may be shown at meetings and published in journals to inform other doctors and health professionals. We will keep your identity private in any publication or presentation.

Several people and organizations may review or receive your identifiable information. They will need this information to conduct the research, to assure the quality of the data, or to analyze the data or samples. These groups include:

- Members of the research team and other authorized staff at CHOP.
- People from agencies and organizations that perform independent accreditation and/or oversight of research; such as the Department of Health and Human Services, Office for Human Research Protections.
- Representatives of CHOP who is the study sponsor funding this research, the data coordinating center and center for genetic testing (if applicable);
- Groups monitoring the safety of this study (CHOP Institutional Review Board);
- The National Institutes of Health who is sponsoring this research;
- If you agree, your data will be shared through databases that may be publicly available to anyone. The data will not include identifiers like your name, medical record number or date of birth. To use your data, researchers must promise not to try to re-identify you. You can tell us at the end of this form whether you will allow is to share your data in this way;

By law, CHOP is required to protect your health information. The research staff will only allow access to your health information to the groups listed above. By signing this document, you are authorizing CHOP to use and/or release your health information for this research. Some of the organizations listed above may not be required to protect your information under Federal privacy laws. If permitted by law, they may be allowed to share it with others without your permission.

The identifiable information from this study will be destroyed six years after the study is completed.

Your permission to use and share the information and data from this study will continue until the research study ends and will not expire. Researchers continue to analyze data for many years and it is not possible to know when they will be completely done.

Can you change your mind about the use of personal information?

You may change your mind and withdraw your permission to use and disclose your health information at any time. To take back your permission, it is preferred that you inform the investigator in writing.

Dr. Lisa Guay-Woodford
The Children's Hospital of Philadelphia
Division of Nephrology
34th Street and Civic Center Blvd.
Philadelphia, PA 19104

In the letter, state that you changed your mind and do not want any more of your health information collected. The personal information that has been collected already will be used if necessary for the research. No new information will be collected. If you withdraw

your permission to use your personal health information, you will be withdrawn from the study.

Financial Information

While you are in this study, the cost of your usual medical care – procedures, medications and doctor visits – will continue to be billed to you or your insurance.

Will there be any additional costs?

There will be no additional costs to you by taking part in this study.

CHOP is providing financial support and material for all optional genetic testing procedures, as listed above, for this study.

Will you be paid for taking part in this study?

You will not receive any payments for taking part in this study.

Who is funding this research study?

The National Institutes of Health is providing funding for this study.

Please ask Dr. Lisa Guay-Woodford if you have any questions about how this study is funded.

What if you have questions about the study?

If you have questions about this study or how your samples/data are going to be used, call the study doctor, Dr. Lisa Guay-Woodford at (267) 425-0315. You may also talk to your own doctor if you have questions or concerns.

The Institutional Review Board (IRB) at The Children's Hospital of Philadelphia has reviewed and approved this study. The IRB looks at research studies like these and makes sure research subjects' rights and welfare are protected. If you have questions about your rights or if you have a complaint, you can call the IRB Office at 215-590-2830.

A description of this clinical trial will be available on http://www.ClinicalTrials.gov, as required by U.S. Law. This Web site will not include information that can identify you. At most, the Web site will include a summary of the results. You can search this Web site at any time.

Sharing Data with the National Institutes of Health (NIH)

Why will my data be shared with the National Institutes of Health (NIH)?

The NIH is funding this study. The NIH's goal is to maximize the benefits that come from the research.

The NIH repository stores genetic information and phenotypic data from many studies. The NIH then shares that information with researchers. We will send the information about you and the other participants to a repository at the NIH. The information will be de-identified (no names or other direct information about you will be included). The NIH will not be able to re-identify you or any other individual.

The NIH intends to share the collected information with other researchers for future research. The researchers who receive data must promise to keep the data confidential

and to use it only for the purpose approved by NIH. They must also promise to not try to re-identify anyone.

The goal of genetic studies is to look for genetic connections that may explain how to identify, prevent, and treat health problems. For example, genetic data may be used to find out:

- Who is more likely to develop a certain illness, such as asthma, cancer, or diabetes, or a condition like high blood pressure or obesity;
- What genes affect the progress of a certain disease or condition; and
- What genes may affect treatments which now may or may not work in certain people.

Risks Associated with Sharing Data with the NIH

There are risks associated with sharing your data with the NIH but they are very unlikely to occur. There is only a very small chance that someone could find out that the data came from you. If that happened, it's possible that someone could deny you a job or health insurance. Or you could experience stress, anxiety, or embarrassment.

Benefits Associated with Sharing Data with the NIH

Sharing your information for future research will not directly benefit you. It is hoped that it will lead to a greater understanding of the interaction between genes and health. This knowledge could help others in the future.

Controlled or Unrestricted Access

The data about you will either be made available by the NIH through controlled access or unrestricted. Controlled access means the data are made available for other research only after investigators have obtained approval from NIH to use the requested data for a particular project. Data for unrestricted access are publicly available to anyone (e.g., The 1000 Genomes Project).

PATIENT OPTIONAL CONSENT **for collection of blood or saliva sample for genetic testing**

If you wish to consent to genetic testing, please select only ONE option (either blood or saliva).

_____ (initials) I agree to have **blood** taken for genetic testing.

_____ (initials) I agree to have **saliva** taken for genetic testing.

_____ (initials) I do not wish to take part in genetic testing for this research.

PARENTAL OPTIONAL CONSENT **for collection of blood or saliva sample for genetic testing**

As a parent if you wish to consent and participate in genetic testing, please select only ONE option (either blood or saliva).

_____ (initials) I agree to have **blood** taken for genetic testing.

_____ (initials) I agree to have **saliva** taken for genetic testing.

_____ (initials) I do not wish to take part in genetic testing for this research.

PATIENT OPTIONAL CONSENT **for collection of tissue sample**

_____ (initials) I agree to give a **tissue** sample to the research study

_____ (initials) I do not wish to take part in a tissue sample for this research.

PATIENT OPTIONAL CONSENT **for collection of urine sample**

_____ (initials) I agree to give a **urine** sample to the research study

_____ (initials) I do not wish to take part in a urine sample for this research.

OPTIONAL CONSENT **for Use of Identifiable Data or Specimens for Future Research**

As part of the study, we will collect:

- Information that identifies you such as name, address, telephone number, date of birth, Social Security number, and other details about you
- Information that relates to your health or medical condition from your medical records
- Information obtained from the study procedures outlined in this consent form, for example: things done to see if you can join the study such as physical exams, blood and urine tests, x-rays and other tests, and any other medical information we learn from you about your health history and family history
- Laboratory results obtained on specimens collected from you (blood, saliva, urine, tissue)

We may wish to use and share this information or samples in a future study about ARPKD and other types of HRFDs.

Research could occur at CHOP, or at outside institutions, which could include for profit companies. The information and samples will be given a unique code. A master list with identifiable information that can be linked back to the uniquely coded data or samples, will only be accessed by research staff and can be used to contact you for future research

with your consent. This master list will be maintained by authorized research staff at CHOP only.

You will not receive any results or financial benefit from the future research done on your specimens or data.

If you leave the study, you can ask to have the data collected about you removed or the samples destroyed. You can also ask us to remove information that identifies you from the data or samples. This may not be possible if your samples and data have already been shared.

Please indicate whether you will allow the identifiable data or samples to be used for future research by putting your initials next to one of the following choices:

_____ (initials) NO, my identifiable (data and/or blood/tissue specimen, as applicable with consent) may not be used for future research. They may be used for this study only.

_____ (initials) YES, my identifiable (data and/or blood/tissue specimen, as applicable with consent) may be used for other future research studies.

Consent to Take Part in this Research Study and Authorization to Use and Disclose Health Information for the Research

The research study and consent form have been explained to you by:

Person Obtaining Consent	Signature of Person Obtaining Consent
	Date

By signing this form, you are indicating that you have had your questions answered, you agree to take part in this research study and you are legally authorized to consent to your child's participation. You are also authorizing the use of your/your child's health information as discussed above. If you don't agree to the collection, use and sharing of health information, you cannot participate in this study.

Name of Subject	
Signature of Subject (18 years or older)	Date

Consent to Take Part in this Research Study and Authorization to Use and Disclose Health Information for the Research

The research study and consent form have been explained to you by:

Person Obtaining Consent	Signature of Person Obtaining Consent
	Date

By signing this form, you are indicating that you have had your questions answered, you agree to take part and to allow your child to take part in this research study, and you are legally authorized to consent to your child's participation. You are also agreeing to let CHOP use and share the health information that will be collected for this study, as explained above. If you don't agree to the collection, use, and sharing of health information, you and your child cannot participate in this study.

NOTE: *A foster parent is not legally authorized to consent for a foster child's participation.*

Consent for Child's Participation

Name of Subject	
Name of Authorized Representative	Relation to subject: ☐ Parent ☐ Legal Guardian
Signature of Authorized Representative	Date

Consent for Parents' participation

Name of Mother	
Signature of Mother	Date
Name of Father	
Signature of Father	Date

Child Assent to Take Part in this Research Study

For children capable of providing assent:

I have explained this study and the procedures involved to _____ in terms he/she could understand and that he/she freely assented to take part in this study.

_____
Person Obtaining Assent

_____ _____
Signature of Person Obtaining Assent Date

This study has been explained to me and I agree to take part.

_____ _____
Signature of Subject (optional) Date

For children unable to assent:

I certify that _____ was not capable of understanding the procedures involved in the study sufficiently to assent to study participation.

_____
Person Responsible for Obtaining Assent

_____ _____
Signature of Person Responsible Date

STUDY SUMMARY SIGNATURE PAGES

For Subjects with Limited English Proficiency

Consent to Take Part in this Research Study and Authorization to Disclose Health Information

Name of Subject

Name of Authorized Representative (if different than subject)

Relation to subject:
☐ Parent ☐ Legal Guardian

The research study and consent form have been explained to the subject or parent/legal guardian.

By signing this form, you are indicating that you have answered the subject's or parent's/legal guardian's questions, they have agreed to take part in this research study and they are legally authorized to consent to their or their child's participation. They have also agreed to let CHOP use and share their or their child's health information as explained above. If they don't agree to the collection, use and sharing of their or their child's health information, they cannot participate in this study.

Person Obtaining Consent

Signature of Person Obtaining Consent

Date:

Witness/Interpreter

By signing this form, you are indicating that

- The information in the Summary Document as well as any additional information conveyed by the person obtaining consent was presented to the subject in a language preferred by and understandable to the subject; and
- The subject's questions were interpreted and the responses of the person obtaining consent were presented in a language preferred by and understandable to the subject.
- At the conclusion of the consent conference, the subject was asked in a language preferred by and understandable to the subject if s/he understood the information in the Summary Document as well as any additional information conveyed by the person obtaining consent (including responses to the subject's questions) and responded affirmatively.

Name of Witness/Interpreter

Signature of Witness/Interpreter

Date:

Child Assent to Take Part in this Research Study

For Subjects with Limited English Proficiency

For children capable of providing assent:

I have explained this study and the procedures involved to _____ in terms he/she could understand and that he/she freely assented to take part in this study.

_____
Person Obtaining Assent

_____ _____
Signature of Person Obtaining Assent Date

Witness/Interpreter

By signing this form, you are indicating that

- The information in the Summary Document as well as any additional information conveyed by the person obtaining assent was presented to the subject in a language preferred by and understandable to the subject; and
- The subject's questions were interpreted and the responses of the person obtaining assent were presented in a language preferred by and understandable to the subject.
- At the conclusion of the consent conference, the subject was asked in a language preferred by and understandable to the subject if s/he understood the information in the Summary Document as well as any additional information conveyed by the person obtaining assent (including responses to the subject's questions) and responded affirmatively.

_____ _____
Name of Witness/Interpreter Signature of Witness/Interpreter

Date: _____