

January 21, 2014

Dear Colleagues,

Thank you for your interest in the mutational screening and linkage analysis of the families of infants with Bilateral Renal Agenesis/Potter Syndrome. We appreciate your review of these documents with your interested participant family. However, we would like to stress that you are not obtaining consent for us, but are acting as a consultant on behalf of the participant.

To date, genes associated with Bilateral Renal Agenesis [OMIM 191830] in humans have not been identified. It is our intent to identify novel genes and mutations associated with Bilateral Renal Agenesis (BRA) and other causes of Potter Syndrome by performing mutational and linkage analysis in participating families. The samples from participants are kept indefinitely and may be used to identify mutations in genes which could be associated with BRA, unilateral renal agenesis (URA), renal adysplasia (RA), renal hypoplasia and other causes of Potter Sequence/Syndrome.

This genetic analysis is investigational and is performed in the setting of a research laboratory. There are no universal standards for the performance of these studies. The investigators endeavor to attain the highest standards in their analysis. These analyses should not be considered diagnostic tests, but rather investigational genetic tests, not intended to replace other clinical or laboratory evaluations or treatments that would otherwise be considered the standard of care.

As these genetic tests are a part of a research protocol, there should be no cost for the blood draw, shipping or processing of the samples to the participants or their family members. If the participant family or their insurance company is billed for the office visit or blood draw, this is an error and should be brought to our attention by contacting either myself or the study coordinator using the information provided in the last section of the Informed Consent Document. We will then take steps to either pay for the office visit and/or blood draw, or remove the participant(s) from the study. Due to the prohibitive costs of office visits for the withdrawal of blood we ask that local physicians include these blood draws as part of the course of the participant's normal office visit where other blood withdraws would normally take place. We will pay for the shipping and handling of the blood and all materials required for the families to participate in this study. Office visits for physicians or genetic counselors are not paid for by this study, nor are any other laboratory tests.

Results from the genetic analysis vary depending on pedigree size, number of affecteds/carriers and phenotypes. Results defining the genetic mutation responsible for a particular cause of BRA/Potter Syndrome are likely to take a year or more. In some cases a genetic cause may never be found. All results are transmitted directly to the corresponding physician via written letter and not to individual participants. Participants will therefore need to depend upon their local physician to communicate and explain the results of the genetic tests. The investigators would be more than willing to discuss the results of the genetic testing with any local physician who wishes to do so.

The participant should keep a signed copy of the consent.

Please return the following items to the investigators:

1. **Signed consent documents:** One signed consent document for each participant.
2. **Health questionnaire:** One completed health questionnaire for each participant.
3. **Blood samples:** Approximately 10ml EDTA or Na-Heparin blood is required from each participant. See table below for allowable volumes. ***Always store blood at room temperature, do not freeze.***
4. **Relevant records, reports and images:** Copies of medical histories, perinatology, nephrology, urology, pathology, physicals, lab values, genetic counseling, etc., and ultrasounds, MRI, photographs, etc.
5. **Pathological samples:** We can process cell culture and fresh or frozen tissue samples when blood material is limited or not available. While formalin fixed paraffin embedded (FFPE) samples are not optimal, we can analyze these in a limited manner. Please contact us regarding shipment of non-blood samples.
6. **DNA Samples:** We prefer to extract the DNA from the blood samples. However, should you choose to extract the DNA, please record the A_{260} value and concentration in $\mu\text{g}/\mu\text{l}$ on the sample tube. Also, please describe the buffer that the DNA is in (H_2O , TE, etc.).

Shipping Instructions:

1. Before collecting samples, please contact Jason Clarke (email: jason-clarke@uiowa.edu ; phone: 319-384-3040) to obtain our shipping account number. We prefer that you use FedEx, but we have accounts with other carriers if you prefer.
2. Indicate contents as blood or DNA, non-hazardous, non-toxic and non-infectious. The value is \$1.00.
3. Please ship blood samples so that they arrive as soon as possible (ideally within 4 days). Deliveries are not made to our laboratory on the weekends.
4. Please ship samples to:
Jason Clarke
University of Iowa, Dept. of Pediatrics
285 Newton Road
CBRB, Room 1257-1
Iowa City, IA 52242, USA
5. Please email jason-clarke@uiowa.edu with the tracking number so that we can ensure safe delivery.

Thank you again for your participation. These documents and additional information can be found on our website at www.kidneygenes.com . Please do not hesitate to contact us with any questions or concerns.

Best Regards,



Patrick Brophy, M.D.
Associate Professor of Pediatrics

For future contact, and to communicate results from our analysis with you, please provide your contact information below and include in the shipment of samples and other paperwork you will be sending to us.

Name: _____ Title: _____

Organization/Company: _____

Address: _____

Phone: _____

INFORMED CONSENT DOCUMENT

Project Title: **Comprehensive genetic analysis of congenital and developmental abnormalities of the genitourinary (GU) tract.**

Principal Investigator: Patrick Brophy

Research Team Contact: Jason Clarke, 319-384-3040 (Lab), jason-clarke@uiowa.edu

- If you are the parent/guardian of a child under 18 years old who is being invited to be in this study, the word “you” in this document refers to your child. You will be asked to read and sign this document to give permission for your child to participate.

This consent form describes the research study to help you decide if you want to participate. This form provides important information about what you will be asked to do during the study, about the risks and benefits of the study, and about your rights as a research subject.

- If you have any questions about or do not understand something in this form, you should ask the research team for more information.
- You should discuss your participation with anyone you choose such as family or friends.
- Do not agree to participate in this study unless the research team has answered your questions and you decide that you want to be part of this study

I. WHAT IS THE PURPOSE OF THIS STUDY?

This is a research study. We are inviting you to participate in this research study because either you or one of your relatives has a congenital or developmental abnormality of the genitourinary (GU) tract, as determined by a physician.

The purpose of this research study is to identify genes that cause congenital and developmental abnormalities of the genitourinary (GU) tract. The GU tract is composed of the organs and tissues that make up your kidneys, urinary system, and reproductive system.

II. HOW MANY PEOPLE WILL PARTICIPATE?

Approximately 2000 people world wide will take part in this study conducted by the investigators at the University of Iowa.

III. HOW LONG WILL I BE IN THIS STUDY?

If you agree to take part in this study, your involvement will last for about 30 minutes to one hour. The results from our analyses are generally available in 1-2 years. At this time, if you have requested to be notified of results, we will report to your doctor or genetic counselor information about the presence or absence of a mutation in the genes that we have analyzed. The DNA taken from the blood sample will be stored indefinitely for the evaluation of the genetic causes of congenital and developmental abnormalities of the GU tract exclusively as described in this document.

IV. WHAT WILL HAPPEN DURING THIS STUDY?

You will have 10 ml (about 2 teaspoons) of blood drawn in your doctor's office. Your doctor will send the blood to us along with your relevant medical records, this signed consent form, images pertaining to your GU tract abnormality (if you have an abnormality) and your health questionnaire if you choose to complete it. We will pay for the shipping of these items. Your doctor or genetic counselor should contact us to obtain our FedEx account number prior to collecting samples from you. Please keep a copy of this consent form for your own records.

Additionally, if you are a pregnant female about to enroll your fetus/baby, we may ask that your doctor collect a blood sample from the umbilical cord of your baby when it is delivered. Collecting the "cord blood" sample minimizes the trauma and risk to your baby. This cord blood sample will be about 3 to 5 ml (about 1 teaspoon) and will be collected by your physician when the baby is delivered. This sample contains your baby's DNA and will be shipped to us in the same manner as described above.

A. The Health Questionnaire:

Following your consent and blood draw at your doctor's clinic, we are asking you to complete the attached health history questionnaire. Completing this questionnaire is voluntary and you can skip any question you choose not to answer. This health questionnaire asks questions regarding the health history of you and your family. Due to the sensitive nature of these questions and your answers, we ask that you do not refer to your relatives by name when answering these questions, but to instead refer to them by their order of relation to you (i.e., paternal uncle, maternal grandfather, etc.). The health questionnaire will cover the following topics: General Information about you or the person you are enrolling (please complete at least this section), Information about the affected baby/individual, Affected baby pregnancy history section, a Work history and Education section, a Substance Use (Tobacco, Alcohol & Drugs) section, there are also several other sections that ask questions about specific conditions that you or somebody in your family may have or have had. These sections are broken down and arranged by the particular part of the body or body system that may be affected. The information that you provide us in this questionnaire will be anonymously entered into a database where we can look for risk factors and other trends that may be associated with this birth defect. We will not share the information from the questionnaire with your doctor or with researchers not associated with this study.

Sometimes it is necessary for the researchers to ask follow up questions to information you provide in your health questionnaire. This could be due to not being able to read what you have written, or to gather more specific information about a question you have answered. May the Researchers contact you either directly by phone or letter to obtain more specific information regarding you and your family's

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APPROVAL DATE: 10/30/13
EXPIRATION DATE: 10/30/14

health history if they believe it may be helpful to the research?

Please initial in the blank next to either “Yes” or “No”: _____ Yes _____ No

If “Yes”, please provide your contact information:

Phone: _____ (Best Time: _____)

Address: _____

B. Our Analysis and Reporting:

The researchers will analyze the blood sample at the University of Iowa for mutations in the genes known to be critical to the development of the GU tract. We are also searching the entire genome of each sample (in most cases), in an attempt to identify mutations in new candidate genes that are not yet known. These results are generally available in 1 year. At this time if you agree we will report the presence or absence of a mutation in the genes that we have analyzed to your doctor or genetic counselor.

If no mutation is present, then the results will not tell us what the cause of your particular disease was. We will continue to perform analysis on these samples, but finding the genetic mutation responsible for your particular urologic abnormality is likely to take a much longer time (more than 1 year) and in some cases a genetic cause for your abnormality may never be found.

When we have results, these will be communicated to your doctor or genetic counselor if you request it and if you are eligible to receive results. **Results will only be sent for individuals who already have a diagnosis of a congenital or developmental abnormality of the GU tract.** Family members of affected individuals participating in this study who **do not** have evidence of a congenital or developmental abnormality of the GU tract themselves, **are not eligible to receive results.** The DNA taken from the blood sample will be stored indefinitely for the evaluation of the genetic causes of congenital and developmental abnormalities of the GU tract exclusively as described in this document.

C. Your Option to Receive Results:

For individual participants: (Please check only one of the following choices)

I **do have** evidence of a congenital or developmental abnormality of the genitourinary (GU) tract and I would like results to be sent to the doctor or genetic counselor as noted below.

I **do have** evidence of a congenital or developmental abnormality of the genitourinary (GU) tract and I **DO NOT** want results of the genetic testing sent to the doctor or genetic counselor.

I **do not have** any evidence of a congenital or developmental abnormality of the genitourinary (GU) tract at this time and I understand that I am not eligible to receive results of my testing.

For the parents/legal guardians of the participant: (Please check only one of the following choices)

I am the parent/legal guardian of this participant. This participant **does have** evidence of a congenital or developmental abnormality of the genitourinary (GU) tract, and I would like the results to be sent to the doctor or genetic counselor as noted below.

I am the parent/legal guardian of this participant. This participant **does have** evidence of a congenital or developmental abnormality of the genitourinary (GU) tract, and I **DO NOT** want the results to be sent to the doctor or genetic counselor.

I am the parent/legal guardian of this participant. This participant **does not have** evidence of a congenital or developmental abnormality of the genitourinary (GU) tract, and I understand that I am not eligible to receive results from this participant's testing.

Your Doctor's/Genetic Counselor's Contact Information:

Please provide us with the contact information of the Doctor/Genetic Counselor you would like us to communicate the results to.

Name: _____

Institution/Organization/Company Name: _____

Address: _____

Phone Number: _____

D. Autopsy Release Consent:

If you are enrolling a deceased person into this study and there was an autopsy performed on this individual, in addition to this consent document, we ask that you complete the Autopsy Release Consent to release samples and reports that were obtained during a routine autopsy. This will allow us to obtain genetic material from samples that were collected, as well as obtain a copy of the final report.

E. Blood/Data Storage for Future Use

As part of this study, we are obtaining blood, or DNA samples from you (and/or your baby or child) as well as health history data. We would like to study this blood, DNA and data in the future, after this study is over.

In some cases, blood cells removed from the blood samples will be used to make a cell line and DNA. Cell lines are produced by growing blood cells in a laboratory and allow us to have a source of the DNA without having to redraw your blood. These blood cells can be stored for decades or more. The cell lines and DNA and data will be made available to researchers trying to learn more about the cause of diseases.

The tests we might want to use to study your blood, DNA and data may not even exist at this time. Therefore, we are asking for your permission to store your blood, DNA and data so that we can study them in the future. These future studies may provide additional information that will be helpful in understanding congenital abnormalities of the GU tract, but it is unlikely that what we learn from these studies will have a direct benefit to you. It is possible that your blood, DNA and data might be used to develop products or tests that could be patented and licensed. There are no plans to provide financial compensation to you should this occur.

If you agree now to future use of your blood, DNA and data, but decide in the future that you would like to have it removed from future research, you should contact Patrick Brophy at 319-384-3090. However, if some research with your blood, DNA and data has already been completed, the information from that research may still be used.

Please initial in the blank next to either “Yes” or “No” to the question below:

My blood, DNA and data may be stored/shared for future gene research in the study of congenital and developmental abnormalities of the GU tract as well as for other health problems (such as cancer, heart disease, etc).

_____ **Yes** _____ **No**

F. Genetic Research

One purpose of this study is to look at genes (DNA) and how they affect health and disease. Genes are the instruction manual for the body. The genes you get from your parents decide what you look like and how your body behaves. They can also tell us a person’s risk for certain diseases and how they will respond to treatment.

You are being asked to give a blood sample for genetic research. What we learn about you from this sample will not be put in your health record. No one else (like a relative, boss, or insurance company) will be given your test results. Your sample will only be used for research at the University of Iowa and will not be sold.

A single 10 ml blood sample will be drawn from a vein in your arm using a needle. This will take about 20 – 30 minutes of your time. If you are enrolling a fetus/baby from a current pregnancy a cord blood sample may be taken instead of having to draw blood directly from the fetus/baby. A single 3 to 5 ml cord blood sample may be taken and this may take an additional 20 minutes of your time.

Results from the genetic tests will only be reported to your local doctor and only if approved by you or your parent/legal guardian. Results cannot be directly reported to you or your parents/legal guardians by the researchers. No results will be reported for participants without a diagnosis of a congenital or developmental abnormality of the GU tract.

A new federal law called the Genetic Information Nondiscrimination Act (GINA) generally makes it illegal for health insurance companies, group health plans, and employers of 15 or more persons to discriminate against you based on your genetic information. Based on this new law, health insurance companies and group health plans are prohibited from requesting your genetic information that we get from this research. This means that they may not use your genetic information when making decisions regarding your eligibility for insurance coverage or the amount of your insurance premiums. Be aware that this new federal law will not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. The law also does not prohibit discrimination if you already have a manifest genetic disease or disorder.

G. Audio Recording/Video Recording/Photographs

One aspect of this study may involve taking photographs of you. This is done in some cases to further characterize a disease or syndrome that affects more than just the kidneys. Some renal syndromes also affect other areas of the body, such as the face, and by having these photographs we will be able to better understand the nature of the disease. Photographs will only be seen by the researchers of this study and if shared or published, they will be altered to hide your identity. These photographs will remain as part of your research record with us and will be destroyed when the study ends or you withdraw your consent to participate.

These photographs are optional and are not a requirement to participate in this study. Please state your willingness to be photographed by the researchers or your local doctor/genetic counselor should the need for these photographs arise as part of the course of this research project by placing your initials in the blank next to either “Yes” or “No”.

I give you permission to take photographs of me during this study. _____ Yes _____ No

V. WHAT ARE THE RISKS OF THIS STUDY?

You may experience one or more of the risks indicated below from being in this study. In addition to these, there may be other unknown risks, or risks that we did not anticipate, associated with being in this study.

The known or expected risks for people participating in the study include:

- 1) The risks for routine venous blood draw, which includes discomfort, bruising, faintness or lightheadedness and very rarely infection.
- 2) The risk for identifying a genetic cause of disease can include difficulty in obtaining insurance or increased cost of insurance. The investigators will not disclose any findings of this study to anyone other than your private doctor or genetic counselor. If desired the researchers will not report any results to your private doctor or genetic counselor.
- 3) The risk for learning of a genetic cause of disease may cause you emotional distress, which could result in depression or anxiety. However, we are only reporting genetic results to individuals who are already aware that they have a congenital or developmental abnormality of the GU tract and therefore, this information will only provide a definitive diagnosis for people who already know they have an abnormality. If you feel that you should consult a genetic counselor, the costs for genetic counseling or office visits related to the discussion or review of genetic testing performed in this study may generate costs that are not reimbursed by this study.
- 4) If you have your blood samples, photographs, questionnaires and medical information shipped to us via Fed Ex, there is a possibility of these items becoming lost in the mail and a slight risk of loss of confidentiality occurring from this.

As with any research study, though, there may be additional risks of participating that are unforeseeable or hard to predict.

A. Genetic Research

One risk of giving samples for this research may be the release of your name that could link you to the stored samples and/or the results of the tests run on your samples. To prevent this, these samples will be given a code. Only the study staff will know the code. The name that belongs to the code will be kept in a locked file or in a computer with a password. Only the researchers Patrick Brophy and Jason Clarke will have access to your name.

VI. WHAT ARE THE BENEFITS OF THIS STUDY?

We don't know if you will benefit from being in this study. However, we hope that, in the future, other people might benefit from this study because this study will help us to move the general understanding of congenital and developmental abnormalities of the GU tract forward with the hope that effective treatments will be developed in the future.

VII. WILL IT COST ME ANYTHING TO BE IN THIS STUDY?

You will not have any costs for being in this research study. If any charges for blood draws or lab tests requested by the investigators as well as the shipping and handling costs of the samples or documents are billed to you, or your insurance, this is an error and should be brought to the attention of the investigators who will cover these costs. The cost of office visits to local doctors or genetic counselors are not covered by this study, nor is any testing other than the blood draw for the DNA sample requested by this study.

VIII. WILL I BE PAID FOR PARTICIPATING?

You will not be paid for being in this research study.

IX. WHO IS FUNDING THIS STUDY?

The University and the research team are receiving no payments from other agencies, organizations, or companies to conduct this research study. Additionally, this research study is funded in part by donations from the public.

X. WHAT IF I AM INJURED AS A RESULT OF THIS STUDY?

If you are injured or become ill from taking part in this study, medical treatment is available at the University of Iowa Hospitals and Clinics.

No compensation for treatment of research-related illness or injury is available from the University of Iowa unless it is proven to be the direct result of negligence by a University employee.

If you experience a research-related illness or injury, you and/or your medical or hospital insurance carrier will be responsible for the cost of treatment.

XI. WHAT ABOUT CONFIDENTIALITY?

We will keep your participation in this research study confidential to the extent permitted by law. However, it is possible that other people such as those indicated below may become aware of your participation in this study and may inspect and copy records pertaining to this research. Some of these records could contain information that personally identifies you.

- federal government regulatory agencies,
- auditing departments of the University of Iowa, and
- the University of Iowa Institutional Review Board (a committee that reviews and approves research studies)

To help protect your confidentiality, we will assign a unique identifier to your blood sample, health information, records and other materials that we may receive as part of your enrollment (biopsies, images, lab results, etc.). No personal identifiers (such as your name, social security number, etc.) will be used to track your sample. The records will be secured in locked cabinets available only to the researchers. Electronic data related to you will be stored on a password protected computer. Extracted DNA samples are labeled with the unique code only, without personal identifiers, and will be stored in a lockable freezer. When results are shared with other scientists no names or other information that could be used to identify the you will be shared. No results will be disclosed except at the request of the participant. If we write a report or article about this study or share the study data set with others, we will do so in such a way that you cannot be directly identified.

If you are a patient at the University of Iowa Hospitals, the University of Iowa Hospitals and Clinics generally requires that we document in your medical record chart that you are participating in this study. The information included in the chart will provide contact information for the research team as well as information about the risks associated with this study. We will keep this Informed Consent Document in our research files; it will not be placed in your medical record chart.

XII. WILL MY HEALTH INFORMATION BE USED DURING THIS STUDY?

The Federal Health Insurance Portability and Accountability Act (HIPAA) requires your health care provider to obtain your permission for the research team to access or create “protected health information” about you for purposes of this research study. Protected health information is information that personally identifies you and relates to your past, present, or future physical or mental health condition or care. We will access or create health information about you, as described in this document, for purposes of this research study. Once your health care provider has disclosed your protected health information to us, it may no longer be protected by the Federal HIPAA privacy regulations, but we will continue to protect your confidentiality as described under “Confidentiality.”

We may share your health information related to this study with other parties including federal government regulatory agencies, the University of Iowa Institutional Review Boards and support staff, federal funding agencies and colleagues at other institutions that are involved with this study.

You cannot participate in this study unless you permit us to use your protected health information. If you choose *not* to allow us to use your protected health information, we will discuss any non-research alternatives available to you. Your decision will not affect your right to medical care that is not research-related. Your signature on this Consent Document authorizes your health care provider to give us permission to use or create health information about you.

Although you may not be allowed to see study information until after this study is over, you may be given access to your health care records by contacting your health care provider. Your permission for us to access or create protected health information about you for purposes of this study has no expiration date. You may withdraw your permission for us to use your health information for this research study by sending a written notice to Dr. Patrick Brophy, University of Iowa Department of Pediatrics; 285 Newton Rd; CBRB Room 1269; Iowa City, IA 52242. However, we may still use your health information that was collected before withdrawing your permission. Also, if we have sent your health information to a third party, such as the study sponsor, or we have removed your identifying information, it may not be possible to prevent its future use. You will receive a copy of this signed document.

XIII. IS BEING IN THIS STUDY VOLUNTARY?

Taking part in this research study is completely voluntary. You may choose not to take part at all. If you decide to be in this study, you may stop participating at any time. If you decide not to be in this study, or if you stop participating at any time, you won't be penalized or lose any benefits for which you otherwise qualify.

A. Will I Receive New Information About the Study while Participating?

If we obtain any new information during this study that might affect your willingness to continue participating in the study, we'll promptly provide you with that information.

B. Can Someone Else End my Participation in this Study?

Under certain circumstances, the researchers might decide to end your participation in this research study earlier than planned. This might happen because:

- The researcher believes that it is not in your best interest to stay in the study.

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- You become ineligible to participate.
- You do not follow instructions from the researchers.
- The study is suspended or canceled.

XIV. WHAT IF I HAVE QUESTIONS?

We encourage you to ask questions. If you have any questions about the research study itself, please contact the Study Coordinator, **Jason Clarke, at 319-384-3040 or jason-clarke@uiowa.edu**. If you experience a research-related injury, please contact the Study PI, **Patrick Brophy at 319-384-3090**.

If you have questions, concerns, or complaints about your rights as a research subject or about research related injury, please contact the Human Subjects Office, 105 Hardin Library for the Health Sciences, 600 Newton Rd, The University of Iowa, Iowa City, IA 52242-1098, (319) 335-6564, or e-mail irb@uiowa.edu. General information about being a research subject can be found by clicking “Info for Public” on the Human Subjects Office web site, <http://research.uiowa.edu/hso>. To offer input about your experiences as a research subject or to speak to someone other than the research staff, call the Human Subjects Office at the number above.

This Informed Consent Document is not a contract. It is a written explanation of what will happen during the study if you decide to participate. You are not waiving any legal rights by signing this Informed Consent Document. Your signature indicates that this research study has been explained to you, that your questions have been answered, and that you agree to take part in this study. You will receive a copy of this form. If you have questions regarding this form or this study, please contact Jason Clarke at 319-384-3040 or jason-clarke@uiowa.edu.

Subject's Name (printed): _____

Do not sign this form if today’s date is on or after EXPIRATION DATE: 10/30/14.

(Signature of Subject)

(Date)

Parent/Guardian’s Name and Relationship to Subject:

(Name of Parent/Guardian - printed)

(Relationship to Subject - printed)

(Name of Subject being enrolled – printed)

Do not sign this form if today’s date is on or after EXPIRATION DATE: 10/30/14.

(Signature of Parent/Guardian)

(Date)

Method by which consent is being obtained:

Please check the method

 Consent is being obtained at a site other than the University of Iowa, solely based on what is written in the document above. **(External clinician does NOT sign this form)**

Consent is being obtained after discussion by a co-investigator listed at the top of this document. (Co-investigator signs below)

Statement of Co-Investigator Who Obtained Consent

(Only to be used by UI Co-investigators listed at the top of this form.)

I have discussed the above points with the subject or, where appropriate, with the subject's legally authorized representative. It is my opinion that the subject understands the risks, benefits, and procedures involved with participation in this research study.

(Signature of Authorized Co-investigator)

(Date)

Comprehensive Genetic Analysis of Congenital and Developmental Abnormalities of the Genitourinary (GU) Tract

Bilateral Renal Agenesis (BRA)/Potter Syndrome & Other Congenital or Developmental Anomalies of the GU Tract Health Questionnaire

This questionnaire is to assist our research and asks questions regarding the health histories of yourself and your family. We prefer that you **do not** use the names of your family members when answering questions. Instead, describe them by their relation to you. Examples: paternal grandmother, maternal uncle, paternal cousin, etc.

You DO NOT have to provide any information regarding your family if you choose. However, this could limit our ability to gather data and fully understand this disorder. At a minimum we ask that you at least answer the questions regarding yourself.

Thank you for taking the time to complete this questionnaire. Please consult the researchers of this study or your physician if you have questions regarding this form.

We ask that each adult enrolling into the study complete their own health questionnaire. Please ensure that each health questionnaire only contains the answers for one person. Children under 18 years of age do not need to complete a health questionnaire.

ALL INDIVIDUALS (ADULTS & CHILDREN) ENROLLING NEED TO COMPLETE THE SECTION BELOW REGARDING "GENERAL INFORMATION ABOUT THE PARTICIPANT"

Today's Date: _____

GENERAL INFORMATION ABOUT THE PARTICIPANT

Last Name: _____ First Name: _____

What is your relationship to the affected individual being enrolled?: Mother Father Sister Brother
 Other (Maternal relation) Other (Paternal relation) If "Other," describe: _____

DOB: _____ (mm)/_____ (dd)/_____ (yyyy) **Blood Type:** O+ O- A+ A- B+ B- AB+ AB-

Current Height: _____ (circle one: feet +inches, inches, cm) **Current Weight:** _____ (circle one: lbs or kg)

Females Only: Are you currently pregnant? No Yes If "Yes," how many weeks? _____ (wks, days)

If "Yes," what was your weight before becoming pregnant? _____ (circle one: lbs, kg)

Are your parents or grandparents consanguineous? (*consanguineous: related by blood or share a common ancestor*)

No Yes, Parents Yes, Maternal Grandparents Yes, Paternal Grandparents

Ethnicity: Hispanic or Latino Not Hispanic or Latino

Race: White or Caucasian Black or African American American Indian or Alaskan Native Asian Native Hawaiian or Pacific Islander Other _____ Two or more races _____

Jewish or Ashkenazi Jew? No Yes, Jewish Yes, Ashkenazi Jew

Section I: Affected Pregnancy History

Please print out and complete this section for each pregnancy affected with an abnormality of the kidney or genitourinary tract.

Information About the Affected Fetus/Infant/Individual

Last Name: _____ **First Name:** _____

Gender: M F Ambiguous genitalia Unknown

Karyotype: 46, XY 46, XX Other _____

DOB: _____ (mm)/ _____ (dd)/ _____ (yyyy) Time: _____ (A.M./P.M.)

DOD: _____ (mm)/ _____ (dd)/ _____ (yyyy) Time: _____ (A.M./P.M.)

Gestational at birth: _____ (wks + days) **Delivery method:** Vaginal C-section N/A

Presentation: Cephalic Breech N/A

Outcome: Miscarriage Spontaneous Abortion Therapeutic Abortion Stillborn Live birth (preterm) Live birth (term)

Diagnosis: Bilateral Renal Agenesis Unilateral Renal Agenesis Urogenital Adysplasia Hydronephrosis Other

If "Other," describe: _____

Diagnostic Method (check all that apply): US MRI IVP Autopsy Other _____

1. What was your age at the time of conception of this pregnancy? _____ (yrs) _____ (mos)
2. Is/was this affected baby a twin or member of a multiple birth? Yes No
3. Have you had more than one pregnancy/baby diagnosed with BRA or Potter Syndrome? Yes (How many? _____) No

If you checked "Yes," please complete a new Section I for each baby/pregnancy
4. Have you had other pregnancies/babies diagnosed with abnormalities of the kidneys, urinary tract or genitals? Yes (How many? _____) No

If you checked "Yes," please complete a new Section I for each baby/pregnancy
5. What was the gestational age of this baby when he/she was diagnosed with BRA/Potter Syndrome? _____ (wks) _____ (days)
6. Were you taking birth control pills when you became pregnant with this baby? Yes No

If "Yes", which kind? _____
7. In the **3 MONTHS PRIOR** to conception of this pregnancy, did you...:

		YES	NO
7a.	Experience "flu like" symptoms (nausea, etc.)?		
7b.	Have a fever?		
7c.	Take prescription medications		
If yes, describe:			

8. At any time **DURING** the course of this pregnancy, did you...:

		YES	NO
8a.	Engage in strenuous exercise or physical activity?		
8b.	Become dehydrated?		
8c.	Develop Diabetes Mellitus? (Do not include preexisting diabetes.)		
8d.	Develop high blood pressure (hypertension)?		
8e.	Develop preeclampsia?		
8f.	Develop eclampsia?		
8g.	Take prescription medications?		
If yes, describe:			
8h.	Take non-prescription (over the counter) medications?		
If yes, describe:			
8i.	Take vitamins and/or supplements?		
If yes, describe:			
8j.	Experience "flu like" symptoms (nausea, etc.) (Do not include morning sickness.)		
8k.	Have a fever?		
8l.	Become exposed to hazardous fumes, chemicals, materials, substances?		
If yes, describe:			
8m.	Consume food or beverages that contained caffeine? (soda, chocolate, tea, coffee, etc.)		
If yes, describe source and frequency:			
8n.	Consume food or beverages that contained artificial sweeteners?		
If yes, describe source and frequency:			
8o.	Smoke cigarettes or use tobacco products?		
If yes, describe how long and how often:			
8p.	Consume alcoholic beverages?		
If yes, describe how long and how often:			
8q.	Use any of the following (check all that apply):		
	Hot tub or jacuzzi?		
	Sauna?		
	Steam room?		
	Prolonged hot baths of 1 hour or longer?		
	Electric blanket?		

Section III: Questions About the Kidney, Urinary Tract & Reproductive System

15. Have you ever had an ultrasound (US) scan or MRI to detect the presence of, or condition of your kidneys?

Yes, US scan Yes, MRI No

If “Yes”, please describe the findings:

	Check if "Yes"	Please Describe
Normal		
Abnormal Right Kidney		
Abnormal Left Kidney		
Absent Right Kidney		
Absent Left Kidney		
Other Abnormal Finding		

16. Do you, or does anyone in your family have or ever had any of the following?: (Please check either “No”, “Yes, myself” or if “Yes, family member,” write in their relationship to you. Some questions have a description area for more information.)

		No	Yes, myself	Yes, family member (incl. relationship to you)
16a.	Blood in the urine?			
Description:		Circle what applies: Hemoglobinuria, Hematuria, Unknown specifics		
16b.	High protein levels in the urine (proteinuria)?			
16c.	Bladder reflux?			
16d.	Vesicoureteral reflux?			
16e.	Obstruction of the kidney, bladder or urinary tract			
Description:		Circle which one: kidney, bladder or urinary tract		

16. (Continued)

		No	Yes, myself	Yes, family member (incl. relationship to you)
16f.	Unilateral Renal Agenesis:			
	Bilateral Renal Agenesis			
16g.	Small kidney(s) (Renal Hypoplasia)			
16h.	Cystic Kidneys			
16i.	Hydronephrosis			
16j.	Any abnormality or condition of the kidney, bladder or urinary tract?			
Description:				
16k.	Kidney failure for any reason?			
16l.	Surgery to correct an abnormality of the kidney, bladder or urinary tract?			

17. Have you , or has anyone in your family been diagnosed as having any of the ?: (Please check either “No”, “Yes, myself” or if “Yes, family member,” write in their relationship to you. Some questions have a description area for more information.)

FEMALE DIAGNOSIS TABLE

		No	Yes, myself	Yes, family member (incl. relationship to you)
17a.	Pelvic Inflammatory Disease (PID)?			
17b.	Solitary fallopian tube?			
17c.	Polycystic Ovary Syndrome/Disease?			
17d.	Cystic Ovary(ies)?			
17e.	Solitary Ovary?			
17f.	Endometriosis?			
17g.	Unicornuate uterus? (One sided uterus)			
17h.	Uterus didelphys? (Double uterus)			
17i.	Bicornuate uterus? (Uterus with two horns)			
17j.	Septated uterus?			
17k.	Any other abnormality of the uterus?			
Describe:				
17m.	Any type of Mullerian abnormality?			
Describe:				
17n.	Any abnormality of the cervix?			
Describe:				
17o.	Any abnormality of the vagina?			
Describe:				
17p.	Any abnormality of the vulva?			
Describe:				

17. (Continued)

MALE DIAGNOSIS TABLE

		No	Yes, myself	Yes, family member (incl. relationship to you)
17q.	Hypospadias?			
17r.	Epispadias?			
17s.	Cryptorchidism?			
17t.	Monorchism?			
17u.	Anorchia?			
17v.	Absent vas deferens?			
17w.	Abnormality of the penis?			
Describe:				
17x.	Abnormality of the testicles?			
Describe:				
17y.	Abnormality fo the scrotum?			
Describe:				
17z.	Other?			
Describe:				

Section IV: Specific Conditions

A. Diabetes:

		No	Yes, myself	Yes, family member (incl. relationship to you)
18	Diabetes Type I (juvenile)			
19	Diabetes Type II (adult)			
20	Secondary conditions due to Diabetes?			
Describe:				

B. Cancer:

21. Have you, or has anyone in your family ever been diagnosed with cancer? Yes, myself Yes, family member No

If “Yes,” please describe the cancer diagnosis and/or the family member as they are related to you:

C. Thyroid:

22. Have you or has anyone in your family ever been diagnosed with a condition or disorder of the thyroid?

Yes, myself Yes, family member No

If “Yes,” please describe (include family member’s relationship to you): _____

D. Cardiopulmonary & Hematology:

23. Have you or has anyone in your family ever been diagnosed with any of the following heart, lung or blood conditions?

		No	Yes, myself	Yes, family member (incl. relationship to you)
23a.	Anemia?			
23b.	Aneurysm?			
23c.	Asthma?			
23d.	Clotting disorder?			
23e.	Congenital heart defect?			
23f.	Heart attack?			
23g.	Heart murmur?			
23h.	High blood pressure?			
23i.	Other defect or condition of the heart?			
Describe:				
23j.	Other defect or condition of the lungs?			
Describe:				

E. Ears & Hearing:

24. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the ears and hearing?

		No	Yes, myself	Yes, family member (incl. relationship to you)
24a.	A known syndrome or disease affecting the ears or hearing?			
Describe:				
24b.	Any type of deafness or hearing loss?			
Describe:				
24c.	Any type of malformation/abnormality of the external ear(s)?			
Describe:				
24d.	Any type of malformation/abnormality of the internal ear(s)?			
Describe:				
24e.	Any type of skin tag near or on the ear(s)?			
Describe:				
24f.	Any type of pit, cyst, cleft or fistula near or on the ear(s)?			
Describe:				
24g.	Surgery to correct hearing loss or abnormalities of the ear(s)?			
Describe:				

F. Head, Neck & Face:

25. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the head, neck or face?

		No	Yes, myself	Yes, family member (incl. relationship to you)
25a.	A known syndrome or disease affecting the head, neck or face?			
Describe:				
25b.	Skin tags on the face?			
Describe:				
25c.	Skin tags on the neck?			
Describe:				
25d.	Pits, sinuses, clefts or fistulas on the neck?			
Describe:				
25e.	Cleft palate and/or cleft lip?			
Describe:				
25f.	Facial paralysis not due to trauma or accident?			
Describe:				
25b.	Surgery to correct abnormalities of the head, neck or face?			
Describe:				
25c.	Any other malformation/abnormality of the head, neck or face?			
Describe:				

G. Eyes & Vision:

26. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the eyes or vision?

		No	Yes, myself	Yes, family member (incl. relationship to you)
26a.	A known syndrome or disease affecting the eyes or vision?			
Describe:				
26b.	Any type of blindness or vision loss?			
Describe:				
26c.	Any type of color blindness			
26d.	One eye obviously differently colored from the other? (heterochromia iridis)			
26d.	An eye obviously composed of more than one color? (sectoral heterochromia iridis)			
26e.	One, or both eyes that abnormally look outward? (exotropia)			
26f.	One, or both eyes that abnormally look inward? (esotropia)			
26g.	One, or both eyes that abnormally look upward? (hypertropia)			
26h.	One, or both eyes that abnormally look downward? (hypotropia)			
26i.	Been diagnosed as having a "Lazy Eye"? (strabismus)			
Describe:				
26j.	Been diagnosed as having Duane's Syndrome or Duane's Anomaly?			
Describe:				
26j.	Skin tags near the eyes?			
Describe:				
26j.	Surgery to correct malformations/abnormalities of the eye(s)?			
Describe:				

27. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the muscles, bones or growth?

		No	Yes, myself	Yes, family member (incl. relationship to you)
27a.	Adrenal Insufficiency Disorder?			
27b.	Gigantism?			
27c.	Kyphosis? (hump back)			
27d.	Marfan Syndrome?			
27e.	Muscular Dystrophy?			
27f.	Oligodactyly? (too few fingers and/or toes)			
27g.	Polydactyly? (too many fingers and/or toes)			
27h.	Scoliosis? (curvature of the spine)			
27i.	Spina bifida?			
27j.	Syndactyly? (two or more fingers and/or toes fused together)			
27k.	Webbed fingers or toes?			
27l.	Surgery to correct malformations/abnormalities of the muscles, bones or skin? (do not include trauma or cosmetic surgeries)			
Describe:				

H. Gastrointestinal Tract/Digestive System:

28. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the gastrointestinal tract or digestive system?

		No	Yes, myself	Yes, family member (incl. relationship to you)
28a.	Anal or rectal fistula?			
28b.	Colitis, ulcerative?			
28c.	Diverticulitis?			
28d.	Gall stones?			
28e.	GERD?			
28f.	Hirschsprung's Disease?			
28g.	Imperforate anus?			
28h.	Inflammatory Bowel Disease?			
28i.	Irritable Bowel Syndrome?			
28j.	Pyloric stenosis?			
28k.	Volvulus?			
28l.	Surgery to correct malformations/abnormalities of the gastrointestinal tract/digestive system?			
Describe:				

I. Nervous System & Mental Development:

29. Have you or has anyone in your family ever been diagnosed with any of the following conditions affecting the nervous system or mental development?

		No	Yes, myself	Yes, family member (incl. relationship to you)
29a.	Alzheimer Disease before 60 years old?			
29b.	Chorea?			
29c.	Dementia?			
29d.	Epilepsy?			
29e.	Multiple Sclerosis?			
29f.	Parkinson Disease?			
29g.	Schizophrenia?			
29h.	Seizures?			
29i.	Severe Depression?			
29j.	ADD or ADHD? (attention deficit disorder/attention deficit hyperactivity disorder)			
29k.	Dyslexia?			
29l.	Impaired mental development or capacity?			
29m.	An IQ score of less than 70?			
29n.	Any learning disorder?			

30. Have you or has anyone in your family ever been diagnosed with any of the following autoimmune conditions?

		No	Yes, myself	Yes, family member (incl. relationship to you)
30a.	Addison's Disease?			
30b.	Crohn's Disease?			
30c.	Gout?			
30d.	Lupus erythematosus?			
30e.	Psoriasis?			
30f.	Rheumatoid arthritis?			

31. Have you or has anyone in your family ever been diagnosed with any of the following genetic or chromosomal conditions?

		No	Yes, myself	Yes, family member (incl. relationship to you)
31a.	A known genetic disease or disorder?			
31b.	Trisomy 21? (Down's syndrome)			
31c.	An abnormality of the chromosomes?			
31d.	Extra chromosomes?			
31e.	Missing or partially missing chromosomes?			
31f.	Any known disorder due to abnormal chromosomes?			
31g.	Born with any birth defects?			
Describe:				
31h.	An illness, disease or condition that appears to run in the family?			
Describe:				
31i.	Any peculiar trait or physical characteristic that appears to run in the family?			
Describe:				

Section V: Reproductive History:

(For Female & Male Participants)

32. Current marital status: Married Divorced With a partner Single

Please provide information about your pregnancy/reproductive history by completing the below table and checking the relevant boxes:

Pregnancy Order	Pregnancy Outcome						Birth weight less than 5 lbs. (2.26 kg)	Gender	DOB (mm/dd/yyyy)	DOD (mm/dd/yyyy)
	Miscarriage/ Spontaneous Abortion	Therapeutic Abortion	Stillborn	Live Birth	Pre-term	Term				
1										
2										
3										
4										
5										
6										
7										
8										
9										
10										

33. Have you had pregnancies with more than one partner? Yes No

If “Yes,” please describe which ones: _____

34. Have you sought fertility counseling or therapy? Yes No

If “Yes,” please describe: _____