Emory University School of Medicine
Consent to be a Research Subject

Title: Clinical Research in Genetics (CRIG)

Principal Investigator: Stephen Warren, Ph.D. & David Ledbetter, Ph.D.

Introduction/Purpose: You have been asked to volunteer for a research study because you are one of the following.

1) You are being evaluated to find out if there is a genetic reason for your health concerns.
2) A family member of yours is being evaluated to find out if there is a genetic reason for his/her health concerns.

In order to better understand this consent form, the following definitions may be helpful.

Genetic material refers to the inherited instructions that are passed from parents to offspring. Genetic material determines physical characteristics, such as eye and hair color.

Genotype refers to the underlying genetic material of an individual that causes normal or abnormal function. A genotype may refer to an individual gene that is or is not functioning properly or a group of genes that are missing or extra.

Phenotype refers to the physical and health characteristics that are caused by an individual’s genetic material (genotype). A phenotype may be a physical attribute such as eye or hair color, blood type, a developmental disability such as mental illness or mental retardation or other health concerns, such as a metabolic disorder or seizure disorder.

For example, one phenotype is blood type. Saying someone has “type A blood” describes their phenotype. However, an individual with “type A blood” will have one of two genotypes. Either both genes are “A” for blood type, or one gene is “A” and the other gene is “O”. Therefore, knowing the phenotype “type A blood” does not tell the genotype (“AA” or “AO”) without further genetic testing.

The purpose of this study is to help researchers understand if a particular phenotype is caused by a person’s genotype. If so, we also want to understand how this genotype is inherited in a family. Therefore, we are interested in collecting samples not only on individuals with a particular phenotype, but also their family members. A phenotype is sometimes caused by just one gene, as in the example of blood type mentioned above. However, more complex phenotypes, such as autism, may be caused by a combination of many different genotypes.

This consent form asks for your permission to collect your genetic material and provide it to researchers now and/or in the future. If your sample is stored, it will be stored in a repository. Researchers who request samples from the repository will only have access to your genetic material and phenotype information, but not to information that identifies you, such as name or social security number.
Procedures: You will be asked to give a small sample of blood (up to 20cc, or about 4 teaspoons), collected from a vein in your arm by a health care professional. You may be asked to provide a saliva sample (collected by spitting into a cup), or a buccal sample (collected by rubbing a swab on the inside of your cheek for 30 seconds) instead of or in addition to the blood sample. The entire procedure should take approximately an hour. Your blood, saliva, or buccal sample will be processed in several ways, one of which may include making an unlimited source of material for future study. (Your sample will not now or in the future be used for human cloning.)

In addition, we may occasionally request other tissue types, such as skin or amniotic fluid. However, these tissue types require more invasive testing than having your blood drawn. Therefore, we would only ask if we could include your other tissue in the repository if you are having that tissue studied as part of your routine clinical care. For instance, a skin biopsy is sometimes required to make a diagnosis of certain genetic conditions. You may be asked if we can include any remaining skin tissue that is not needed for diagnostic testing. You will be asked to sign a separate consent form for collection of these other types of tissues.

In addition, you may be asked to complete an initial evaluation which includes tests designed to evaluate your developmental skills, vocabulary and ability to solve simple problems. You may be asked to complete a short neurological and/or psychological exam. The complete initial evaluation will take between 20 minutes and about 2 hours. You may be asked to return yearly to complete a similar evaluation. All of your information will be entered into a computer database.

Any biological products that are made from your sample will become the property of Emory University or the researcher studying your sample. In order to protect your privacy, all samples and products made from your blood will be assigned an identification code that does not include any of your personal information. Your sample will be stored for as long as it is useful, unless you ask us to destroy it sooner. You may request that your sample be destroyed at any time, simply by contacting repository administrator, Nikki Justice (404-778-8550). The Principal Investigators of this study will also share stored samples with other scientists for research purposes, but your name or other identifying information will not be given to them.

Risks: Collecting blood from a vein in someone's arm is a standard medical procedure, although sometimes there may be some pain or bruising. Because we will be looking at genetic information in your blood, saliva, or buccal sample, there may also be other risks that we currently do not recognize or expect. Risks could include a disruption in family relationships, such as learning the person stated to be the father is not the biological father of an individual. You can avoid these other potential risks by choosing not to learn the results of these studies, and by choosing not to have these results reported to your health care professional. Please see the results and new finding section below to learn more about how results will be reported.

Benefits: Although your participation in this study may not directly help you or your relatives, the results of this research project could help us understand more about the genetic causes of human diseases. In addition, this research may help you or your family understand more about the cause of phenotype in your family. Please see the results and new finding section below to learn more about how results will be reported.

Confidentiality: People other than those doing the study may look at both medical charts and study records. Agencies and Emory departments and committees that make rules and policy about how research is done have the right to review these records. So do companies and agencies that pay for the study. The government agencies and units within Emory responsible for making sure that studies are conducted and handled correctly that may look at your study records in order to do this job include the Office for Human Research Protections, the Emory University Institutional Review Board, the Emory Office of Research Compliance, and the Emory

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University Clinical Trials Office. In addition, records can be opened by court order or produced in response to a subpoena or a request for production of documents. We will keep any records that we produce private to the extent we are required to do so by law. We will use a study number rather than your name on study records where we can. Your name and other facts that might point to you will not appear when we present this study or publish its results.

If you are or have been a patient at an Emory Healthcare facility, then you will have an Emory Healthcare medical record. If you are not and have never been a patient at an Emory Healthcare facility then no Emory Healthcare medical record will be created for you just because you are participating in a research study.

Results from study tests and procedures that are performed, analyzed and/or read at or for Emory Healthcare facilities that can be used for healthcare purposes will be placed in any medical record that you have with Emory Healthcare facilities. In addition, a copy of the informed consent form and HIPAA authorization form that you sign will be placed in any Emory Healthcare medical record you may have. Persons who have access to your medical record will be able to have access to all results and documents that are placed there, and the results/documents may be used by Emory Healthcare facilities to help provide you with medical care. Any results and documents that are kept as part of your medical record are not covered by certain state and federal laws and regulations that may prevent the disclosure of, research data. However, the confidentiality of the results and other documents in the medical record will be governed by laws such as HIPAA that concern medical records.

Emory University does not have any control over results from tests and procedures performed and/or analyzed or read at non-Emory Healthcare facilities. These results are NOT routinely included in medical records at Emory Healthcare facilities, and they will not necessarily be available to Emory Healthcare providers. Emory University also does not have control over any other medical records that you may have with other healthcare providers and will not send any test or procedure results from the study to these providers. It is up to you to let these healthcare providers know that you are participating in a clinical trial.

Some tests and procedures that may be performed during this study by Emory Healthcare or other facilities or persons may not be looked at or read for any healthcare treatment or diagnostic purposes. These test and procedures will only be looked at for research purposes and the results will not be reviewed to make decisions about your personal health or treatment. The specific types of tests or procedures, if any, that fall within this category are listed below: none.

**Compensation:** There will be no extra costs to you or your insurance company for providing a blood sample in this study. Similarly, you and/or your relatives will not receive any money for participating in this study. If a clinically important result is found and you agree, you will be re-contacted to discuss these results. However, the cost of the genetics clinic evaluation and confirmation of results is not covered by this study. You will be asked to make an appointment with a genetics professional in the Department of Human Genetics to discuss results. In order to release these results to you, confirmation in a clinical laboratory is required.

You should also understand that blood, saliva, or buccal samples removed from you for this study may be valuable for scientific, research, or teaching purposes, or for the development of new medical products. By agreeing to participate in this research, you authorize Emory University and members of its staff to use your blood for these purposes. If this future research leads to the development of new diagnostic tests, new medicines, or other uses that may be commercially valuable, you will receive no financial benefits.
In the very unlikely event that you are injured as a result of the sample collection, medical treatment will be made available to you. Emory University has not set aside any funds for payment of costs associated with any injury resulting from participation in this study.

**Contact Persons:** If you have any questions about this study, please contact a CRIG administrator, Nikki Justice at (404) 778-8550. If you have further questions about your rights as a volunteer, you may contact Dr. Colleen Dilorio, the Chair of the Emory University Institutional Review Board (IRB), at 404-712-0720.

**New Findings:** The information that is learned from studies of your samples may be used scientifically. The results of our studies of your samples may be made available to you or to your referring health care professional. It is your choice whether or not you want to know these results, and whether or not you want to have them reported to your health care provider for possible entry into your medical records. No results will be reported until the Principal Investigators are confident of the accuracy and interpretation of those results.

If a clinically significant result is learned from this research and you tell us you want to learn the results, you will be contacted by a study administrator to discuss the process. If you still wish to learn the results, you will be asked to sign a release form and make an appointment with one of the genetics professionals in the Department of Human Genetics to discuss the results. During this appointment we will discuss how this finding may or may not affect your medical care. Under federal law, results from research laboratories cannot be used in clinical care of an individual. Therefore, before results can be released to you, the research results are confirmed by a clinical laboratory. The cost of a genetics clinical evaluation and confirmation in the clinical laboratory are not covered by this research study. Therefore, you or your insurance company will be billed for these services. Once research results are confirmed in a clinical laboratory, your care will be according to standard of care at the time.

**Voluntary Participation and Withdrawal:** Participation in this study is voluntary. You are free not to participate in this study, or to withdraw your participation at any time. Your decision to participate or not participate in this study will in no way affect your current or future medical treatment. Should you wish to withdraw once you have already donated samples, simply notify Nikki Justice by calling (404) 778-8550. Similarly, you do not have to agree to participate in any follow-up activities that may be asked of you at a later time.

A copy of this consent form will be given to you.

Your signature below indicates that you consent to volunteer either yourself, or the child or adult for whom you serve as guardian (as indicated), for participation in this study.

<table>
<thead>
<tr>
<th>Signature (patient/subject)</th>
<th>Date/Time</th>
</tr>
</thead>
<tbody>
<tr>
<td>Signature (parent or guardian)</td>
<td>Date/Time</td>
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**Signature Date/Time**
(person obtaining consent)

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**IRB#: 1317-2004**

**Consent Form Approval Period**
FROM: 02-01-08 TO: 1-2-09

**AUTHORIZATION:** RO

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For Phone Consent

I have read this authorization form and have been given the chance to ask questions about it.

I spoke with a study investigator, ________________________________, on

_____/_____/_______ at _________ to discuss the study and ask questions.
(date) __________________ (time) __________________

If a clinically significant finding is discovered, would you like to be contacted by one of the CRIG administrators to discuss how to receive these results? Please initial and date your choice.

_______ Yes ________ No _______/_____/_______ Date

Are you willing to be contacted to discuss other research studies being conducted by the Department of Human Genetics at Emory University? You may choose to participate or not participate at that time.

_______ Yes ________ No _______/_____/_______ Date

May we contact you as needed to get updated clinical information?

_______ Yes ________ No _______/_____/_______ Date
Emory University School of Medicine
Assent to be a Research Subject

Title: Clinical Research in Genetics (CRIG)

We are asking you to volunteer to be in a medical research study. The study is to help us explain why some children have medical or learning problems. We hope this study will help us find ways to help other children with medical or learning problems in the future. To be in the study, we will need to draw a tube of blood from your arm, collect a sample of your saliva in a cup, and/or rub a small brush on the inside of your cheek to collect some skin cells. Most people say that taking blood from their arm hurts, sort of like a bee sting; collecting the saliva sample or the sample from the inside of your cheek does not hurt.

However, it is important to us that you participate because you want to. You can refuse (say no) to be in this study. Your doctors or your parent(s)/legal guardian(s) cannot make you be in the study if you do not want to be in it. If you agree to be in the study but change your mind about it later, you can stop being in the study.

Your doctor will talk to you about what it means to be in a research study. He or she will also talk to you about what it means to have medical or learning problems. You should ask your doctor all of the questions you have. You should also talk to your parent(s)/legal guardian(s) about the study and tell them if you want to be in it.

Before you sign, you will be asked to speak with someone at the doctor's office. You can ask this person any questions you want about the study. After you speak with him/her if you agree to be in the study, please sign here:

Participant's Name

For the person administering this consent form: Please fill out the following section only if the study involves children.

1. _____ This child is less than 6 years old. No assent required.

2. _____ This child is 6-10 years old. Verbal assent required, written assent requested. The study should be explained to the child in language he/she can understand, with any questions addressed thoroughly. The child should be asked verbally if they agree to participate in the study. If the child agrees, he/she should be asked if they would please sign their name on the line below to indicate their assent. If the child agrees to participate in the study, but cannot, or does not want to sign their name, the consenter may document (here and in the medical record) that the assent has been obtained verbally.

Signature of participant Date/Time
(indicating assent for 6-10 year olds)

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Consent Form Approval Period
FROM: 2-1-08 TO: 1-2-09

Authorization: RD
3. _____ This child is 11-17 years old. Written assent required. The study should be explained to the child in language he/she can understand, with any questions addressed thoroughly. The child should then be asked if they agree to participate, and if so, should be asked to sign their name on the line below to indicate their assent.

__________________________ Date/Time
Signature of participant (indicating assent for 11-17 year olds)

4. _____ This child is unable to provide informed assent. In accordance with Emory University IRB policy, if the child is too immature or otherwise unable to give informed assent, it is the investigator's prerogative to state the following:

In my opinion this child cannot give informed assent.

Reasons:

__________________________ Date/Time
Person obtaining assent

IRB#: 1317-2004
Consent Form Approval Period
FROM: 2-1-08 TO: 1-2-09
AUTHORIZATION: RD