You Are Being Asked to Be in a Research Study

What Is a Research Study?

The main purpose of research studies is to gain knowledge. This knowledge may be used to help others. Research studies are not intended to benefit you directly, though some might.

Do I Have to Do This?

No. Being in this study is entirely your choice. If you decide to join this study, you can change your mind later on and withdraw from the research study.

Taking part in a study is separate from medical care. The decision to join or not join the research study will not affect your status as a patient.

What Is This Document?

This form is an informed consent document. It will describe the study risks, procedures, and any costs to you.

This form is also a HIPAA Authorization document. It will describe how your health information will be used and by whom.

Signing this form indicates you are willing to take part in the study and allow your health information to be used.

What Should I Do Next?

1. Read this form, or have it read to you.
2. Make sure the study doctor or study staff explains the study to you.
3. Ask questions (e.g., time commitment, unfamiliar words, specific procedures, etc.)
4. If there will be medical treatment, know which parts are research and which are standard care.
5. Take time to consider this, and talk about it with your family and friends.

Emory University and Children's Healthcare of Atlanta
Consent to be a Research Subject / HIPAA Authorization

Title: Studies of Genes Involved in Head and Facial Disorders

Principal Investigator: Elizabeth J. Leslie, Ph.D., Department of Human Genetics

If you are the legal guardian of a child who is being asked to participate, the term "you" used in this consent refers to your child.

Introduction

You are being asked to be in a medical research study. This form is designed to tell you everything you need to think about before you decide if you want to be a part of the study. **It is entirely your choice. If you decide to take part, you can change your mind later on and withdraw from the research study.** The decision to join or not join the research study will not cause you to lose any medical benefits. If you decide not to take part in this study, your doctor will continue to treat you.

Before making your decision:

- Please carefully read this form or have it read to you
- Please listen to the study doctor or study staff explain the study to you
- Please ask questions about anything that is not clear

You can take a copy of this consent form, to keep. Feel free to take your time thinking about whether you would like to participate. You may wish to discuss your decision with family or friends. Do not sign this consent form unless you have had a chance to ask questions and get answers that make sense to you. By signing this form you will not give up any legal rights.

What is the purpose of this study?

The purpose of this study is to understand the different factors that contribute to craniofacial differences, including orofacial clefting. We do not yet understand the processes that give rise to these disorders but other studies point to personal genetic factors like family history of clefting and other medical conditions, and environmental factors like exposure to certain substances during pregnancy. The overall goal of the study is to develop a better understanding of genes related to orofacial clefts and how they interact with the environment, in order to improve treatment, management, and prevention of orofacial clefts.

What will I be asked to do?

In this study, we will be collecting information, including biological samples. This study does not involve any experimental treatments or investigational products.

This study consists of one Study Visit, where we will collect information relevant to our study, including surveys, physical measurements, and biospecimen sample collection (a saliva or blood sample). If there are any problems or missing information, we may recontact you after the appointment via phone or email to

clarify. We may also ask you to sign a Medical Records Release Form so that we can obtain more complete and accurate information about your medical history.

We will ask you to submit a sample, usually a saliva sample. If you are willing, we may instead draw blood (about 4 tablespoons, 4-5 vials) from a vein in your arm. You will be asked to complete surveys about your family and medical history. We will ask about and will ask about characteristics of your family, personal medical history and history of medical and genetic conditions that your family may have. These surveys may be completed prior to or during the Study Visit. If you are not travelling to Emory, we will send you a saliva kit and instructions for collecting the sample. If you are willing, we may arrange to have blood drawn near your home. This can be at your doctor's office, or a local clinic, lab, or hospital.

We will be collecting a series of facial photographs (including 3-D photos) during the study visit. We may also take photographs of other traits that may be genetically related to orofacial clefting, such as your ears, hands, or feet. If you are not travelling to Emory, we may ask you to provide current photographs of your face, lips, ears, and hands. If you have had a surgical repair, we will ask you to provide baby photos to document the pre-surgical form of your cleft or craniofacial anomaly. If you agree, the photos may be used in presentations or publications about this research study, including professional conferences or other education purposes related to this study. The photos will not reveal your name or any identifying information. Please initial below to indicate your permission:

_____I give my permission to be photographed.

_____I give my permission for the recordings and/or photos to be used in presentations or publications about this research. I understand that my name and other identifying information will not be revealed.

Who owns my study information and samples?

If you join this study, you will be donating your samples and study information. You will not receive any compensation if your samples or information are used to make a new product. If you withdraw from the study, data and samples that were already collected may be still be used for this study. A code will be used to link your samples to your clinical information, your answers to questionnaires, and any other records you release to the researchers. Your biological samples and your information will be stored at Emory University in secure databases and biospecimen banks.

The materials that we collect including samples, medical information, photographs, and locked files with personal information will be kept indefinitely. We may re-contact you in the future about this study or to ask for permission to use your material for any other purpose other than stated above. You can ask us to destroy your sample, test results, or any other information at any time in this study.

If your child is participating in this study, your child's samples, genomic data, and health information will be stored and used for future research. When your child reaches age 18, we will try to contact him or her to ask whether he or she wants to continue to participate in research. If we cannot find your child, we will remove identifying information, and continue to include his or her samples, genomic data, and health information in research.

In general, we will not give you any individual results from the study of the samples you give us. We may determine additional information about orofacial clefting in your family; this may also uncover information about your carrier status that might be important for your family members or your plans to have additional children. If we find something of urgent medical importance to you, we will inform you, although we expect that this will be a very rare occurrence. If you wish to request your results, you should contact the Principal Investigator, Elizabeth Leslie, PhD. However, this will be research results only and you would need to have these results confirmed by a diagnostic laboratory.

It is possible that we will discover that you have a gene variant/medical issue that is unrelated to the purpose of this study. If we believe that the information is of urgent medical importance, we will share this information with you. You should not assume that if you are not contacted, that you do not have any gene variants that might be related to a disease. If your child is participating and is under 18, we will tell you only information directly related to diseases and disorders that affect children. Your child can request additional information when he or she is 18.

What are the possible risks and discomforts?

The proposed protocol includes only minimal risk procedures. The most common risks and discomforts expected in this study would result from a blood draw, which is optional. If you are unwilling to donate blood, you may provide a saliva sample instead. If a blood sample is taken, possible risks and discomforts associated with the blood draw include slight soreness, bleeding or bruising at the point where the blood is taken, possible infection, and in rare cases, fainting or light-headedness.

The other procedures (including 3-D and 2-D photographs) have no known risks associated with them.

There is a small risk of loss of confidentiality. Your privacy is very important to us and we will use many safety measures to protect your privacy. However, in spite of all of the safety measures we will use, we cannot guarantee that your identity will never become known. Whenever possible, a study number, rather you're your name, will be used on study records, samples, and medical information.

There may be side effects from the study procedures that are not known at this time. It is possible that the researchers will learn something new during the study about the risks of being in it. If this happens, they will tell you about it. Then you can decide if you want to continue to be in this study or not. You may be asked to sign a new consent form that includes the new information if you decide to stay in the study.

In Case of Injury

If you get ill or injured from being in the study, Emory and Children's Healthcare of Atlanta will help you get medical treatment. Emory and Children's Healthcare of Atlanta and the sponsor have not, however, set aside any money to pay you or to pay for this medical treatment. The only exception is if it is proven that your injury or illness is directly caused by the negligence of an Emory and Children's Healthcare of Atlanta or sponsor employee. "Negligence" is the failure to follow a standard duty of care. If you become ill or injured from being in this study, your insurer will be billed for your treatment costs. If you do not have insurance, or if your insurer does not pay, then you will have to pay these costs. If you believe you have become ill or injured from this research, you should contact Dr. Elizabeth Leslie at telephone number 404-727-3505. Because there are no treatments associated with this study, it is not necessary for you to let other healthcare providers who treat you know that you are in a research study, but you may choose to do so.

How is my Genetic Information Protected? What are the Risks?

Although your genetic information is unique to you, you do share some genetic information with your children, parents, brothers, sisters, and other blood relatives. Consequently, it may be possible that genetic information from them could be used to help identify you. Similarly, it may be possible that genetic information from you could be used to help identify them. Since some genetic variations can help to predict future health problems for you and your relatives, this information might be of interest to health care providers, life insurance companies, and others. However, Federal and State laws provide some protections against discrimination based on genetic information.

The Genetic Information Nondiscrimination Act (GINA) is a federal law that 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 generally will 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.

Be aware that GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance, and does not apply to employers with less than 15 employees.

In addition to GINA, the State of Georgia has laws that prohibit insurers from using genetic testing information for any non-treatment purpose. However, like GINA, this state law protection has exclusions: life insurance policies, disability income policies, accidental death or dismemberment policies, Medicare supplement policies, long-term care insurance policies, credit insurance policies, specified disease policies, hospital indemnity policies, blanket accident and sickness policies, franchise policies issued on an insurance policy written as a part of workers' compensation equivalent coverage, or other similar limited accident and sickness policies.

Privilege

In the State of Georgia, your genetic information has special legal protections called "privilege," which means that the information collected here cannot be used as evidence in a court. By signing this form and allowing us to use and disclose your genetic information for the purposes described in this consent, you waive any privilege with regard to that genetic information, meaning that the information loses this legal protection.

How will you protect my private information that you collect in this study?

Emory and Children's Healthcare will keep any research records that it creates private to the extent that this is required to do so by law. We will assign a coded study number to your medical information that you provide, to your biological samples, and to your answers to questionnaires. The samples and information will not be stored with your name or any other information that points to you. We will store files that link your name and code number separately under lock and key or in a safeguarded password-protected database. Only very few,

authorized people, who have specifically agreed to protect your identity, will have access to this database. All other researchers and personnel, including those who will be working with your samples and medical information, will not have access to any of the traditionally-used identifying information about you. Your name and other identifying information will not appear when we present or publish the study results. Study records can be opened by court order. They also may be provided in response to a subpoena or a request for the production of documents.

Certificate of Confidentiality

There is a Certificate of Confidentiality from the National Institutes of Health for this Study. The Certificate of Confidentiality helps us to keep others from learning that you participated in this study. Emory will rely on the Certificate of Confidentiality to refuse to give out study information that identifies you. For example, if Emory received a subpoena for study records, it would not give out information that identifies you.

The Certificate of Confidentiality does not stop you or someone else, like a member of your family, from giving out information about your participation in this study. For example, if you let your insurance company know that you are in this study, and you agree to give the insurance company research information, then the investigator cannot use the Certificate to withhold this information. This means you and your family also need to protect your own privacy.

The Certificate does not stop Emory from making the following disclosures about you:

- Giving state public health officials information about certain infectious diseases,
- Giving law officials information about abuse of a child, elderly person or disabled person.
- Giving out information to prevent harm to you or others.

Giving the study sponsor or funders information about the study, including information for an audit or evaluation.

Storing and Sharing your Information

De-identified data from this study, including your de-identified genetic information, may be shared with the research community at large to advance science and health. Data from this study may be placed into public databases where, in addition to having no direct identifiers, researchers will need to sign data use agreements before accessing the data. We will remove or code any personal information that could identify you before your information is shared.

While the databases will not have information such as your name, address, telephone number, or social security number, it may be possible to identify you based on the information in these databases and other public information (including information you tell people or post about yourself). The risk of this happening is currently very low.

The samples and information will be available for any research question, such as research to understand what causes certain diseases (for example heart disease, cancer, or psychiatric disorders), development of new scientific methods, or the study of where different groups of people may have come from. The data, samples, and health information will be de-identified and/or coded (again, no names or personal identifiers). We will

submit your de-identified or coded data and results to a controlled-access database. This means that only researchers who apply for and get permission to use the information for a specific research project will be able to access the information or samples. Researchers approved to access information in the database will agree not to attempt to identify you. Your genomic data, health information and samples will not be labeled with your name or other information that could be used to identify you. This will ensure that, by current scientific standards and known methods, it is extremely unlikely that anyone would be able to identify you from the information we share. Despite these measures, we cannot guarantee anonymity of your personal data. However, this sharing of resources will help other researchers in the field and make your effort even more valuable.

Medical Record

If you have been an Emory and Children's Healthcare of Atlanta patient before, then you already have an Emory and Children's Healthcare of Atlanta medical record. If you have never been an Emory and Children's Healthcare of Atlanta patient, you do not have one. The information collected for this study will be used only for research purposes and will not be placed in your medical record, including all surveys, medical records, photographs, and biological samples. Tests and procedures done at non-Emory and Children's Healthcare places will not become part of your Emory and Children's Healthcare medical record. Also, if you decide to be in this study, it is up to you to let your other health providers know.

Will I benefit directly from the study?

This study is not designed to benefit you directly. This study is designed to learn more about the genetics of Orofacial Clefting and craniofacial differences. The study results may be used to help others in the future by improving diagnosis or treatment for such conditions.

In general, we will not give you any individual results from the study of the samples you give us. If we find something of urgent medical importance to you, we will inform you, although we expect that this will be a very rare occurrence.

Will I be compensated for my time and effort?

You will get $50 for completing the study visit, to compensate you for your time and effort. You will receive $25 for completing questionnaires and another $25 for providing a sample. Additional family members may also be asked to provide samples, and will receive $25 for providing their sample.

Costs

There will be no costs to you for participating in this study, other than basic expenses like transportation. You will not be charged for any of the research activities.

Withdrawal from the Study

You have the right to leave a study at any time without penalty.

Authorization to Use and Disclose Protected Health Information

The privacy of your health information is important to us. We call your health information that identifies you, your "protected health information" or "PHI." To protect your PHI, we will follow federal and state privacy

laws, including the Health Insurance Portability and Accountability Act and regulations (HIPAA). We refer to all of these laws as the "Privacy Rules." Here we let you know how we will use and disclose your PHI for the study.

PHI that Will be Used/Disclosed:
The PHI that we will use or share for the main research study includes:

- Medical information about you including your medical history and present/past medications.
- Results of exams, procedures and tests you have before and during the study.
- Laboratory test results.
- Facial photographs, including 3-D photographs, other relevant photographs, and photographic records taken before the study (if applicable).

Purposes for Which Your PHI Will be Used/Disclosed:
We will use and share your PHI for the conduct and oversight of the research study. We will use and share your PHI to provide you with study related treatment and for payment for such treatment. We will also use and share your PHI to conduct normal business operations. We may share your PHI with other people and places that help us conduct or carry out the study, such as laboratories, data management centers, data monitors, contract research organizations, Institutional Review Boards (IRBs) and other study sites. If you leave the study, we may use your PHI to determine your health, vital status or contact information.

Use and Disclosure of Your Information That is Required by Law:
We will use and disclose your PHI when we are required to do so by law. This includes laws that require us to report child abuse or abuse of elderly or disabled adults. We will also comply with legal requests or orders that require us to disclose your PHI. These include subpoenas or court orders.

Authorization to Use PHI is Required to Participate:
By signing this form, you give us permission to use and share your PHI as described in this document. You do not have to sign this form to authorize the use and disclosure of your PHI. If you do not sign this form, then you may not participate in the research study.

People Who will Use/Disclose Your PHI:
The following people and groups will use and disclose your PHI in connection with the research study:

- The Principal Investigator and the research staff will use and disclose your PHI to conduct the study.
- Emory and Children's Healthcare of Atlanta may use and disclose your PHI to run normal business operations.
- The Principal Investigator and research staff will share your PHI with other people and groups to help conduct the study or to provide oversight for the study.
- The following people and groups will use your PHI to make sure the research is done correctly and safely:
    - Emory and Children's Healthcare of Atlanta offices that are part of the Human Research Participant Protection Program and those that are involved in study administration and

billing. These include the Emory and Children's Healthcare of Atlanta IRBs, the Emory Research and Healthcare Compliance Offices, and the Emory Office for Clinical Research.
    - Government agencies that regulate the research including: Office for Human Research Protections
    - Public health agencies.
    - Research monitors and reviewer.
    - Accreditation agencies.
- Sometimes a Principal Investigator or other researcher moves to a different institution. If this happens, your PHI may be shared with that new institution and their oversight offices. PHI will be shared securely and under a legal agreement to ensure it continues to be used under the terms of this consent and HIPAA authorization.

Storage of Data/Specimens for Future Research:

PHI That Will be Used/Disclosed for Future Research:
The PHI that we will use and/or disclose (share) for future research use includes: genetic data derived from biological samples, facial data derived from photograph samples, and/or relevant medical records and family history. Any remaining saliva or blood sample may be stored for future use. Outside of this study, any data will be de-identified (will no longer be connected to identifying personal information) and non-identifying phenotypic data (facial data derived from photographs) will be connected to biological samples using coded IDs for use by qualified investigators with IRB approval.

Purposes for which your PHI will be Used/Disclosed for Future Research:
We will use and disclose your PHI for the conduct and oversight of storage and future research.

People Who Will Use/Disclose Your PHI for Future Research:
The following people and groups will use and disclose your PHI in connection with storage and future research. In addition, the following people and groups may also use and disclose your PHI in future research:

- The Principal Investigator and the research staff will use and disclose your PHI to conduct the study.
- Emory and Children's Healthcare of Atlanta may use and disclose your PHI to run normal business operations.
- The Principal Investigator and research staff will share your PHI with other people, researchers, and groups to help conduct the study or to provide oversight for the study.
    - The following people and groups will use your PHI to make sure the research is done correctly and safely: the Emory and Children's Healthcare of Atlanta IRBs, the Emory Research and Healthcare Compliance Offices, and the Emory Office for Clinical Research; Government agencies that regulate the research; Public health agencies; Research monitors and reviewers; Accreditation agencies.
- Sometimes a Principal Investigator or other researcher moves to a different institution. If this happens, your PHI may be shared with that new institution and their oversight offices. PHI will be shared securely and under a legal agreement to ensure it continues to be used under the terms of this consent and HIPAA authorization.

Expiration of Your Authorization
Your PHI will be used until this research study ends.

Revoking Your Authorization
If you sign this form, at any time later you may revoke (take back) your permission to use your information. If you want to do this, you must contact the study team at: Emory Cleft Study c/o Dr. Elizabeth Leslie, 615 Michael Street, Whitehead Bldg #301, Atlanta, GA 30306 or (404)727-3505 or cleftstudy@emory.edu.

At that point, the researchers would not collect any more of your PHI. But they may use or disclose the information you already gave them so they can follow the law, protect your safety, or make sure that the study was done properly and the data is correct. If you revoke your authorization you will not be able to stay in the study.

Other Items You Should Know about Your Privacy
Not all people and entities are covered by the Privacy Rules. HIPAA only applies to health care providers, health care payers, and health care clearinghouses. If we disclose your information to people who are not covered by the Privacy Rules, including HIPAA, then your information won't be protected by the Privacy Rules. People who do not have to follow the Privacy rules can use or disclose your information with others without your permission if they are allowed to do so by the laws that cover them.

To maintain the integrity of this research study, you generally will not have access to your PHI related to this research until the study is complete. When the study ends, and at your request, you generally will have access to your PHI that we maintain in a designated record set. A designated record set is data that includes medical information or billing records that your health care providers use to make decisions about you. If it is necessary for your health care, your health information will be provided to your doctor.

We may remove identifying information from your PHI. Once we do this, the remaining information will not be subject to the Privacy Rules. Information without identifiers may be used or disclosed with other people or organizations for purposes besides this study.

Contact Information
Contact Dr. Elizabeth Leslie at (404) 727- 2825 or cleftstudy@emory.edu:

- if you have any questions about this study or your part in it.
- if you have questions, concerns or complaints about the research

Contact the Emory Institutional Review Board at 404-712-0720 or 877-503-9797 or irb@emory.edu:

- if you have questions about your rights as a research participant.
- if you have questions, concerns or complaints about the research.
- You may also let the IRB know about your experience as a research participant through our Research Participant Survey at http://www.surveymonkey.com/s/6ZDMW75.

If you are a patient receiving care at Children's Healthcare of Atlanta of Atlanta and have a question about your rights, please contact Kristine Rogers, Director of Clinical Research at 404-785-1215.

Consent and Authorization:

TO BE FILLED OUT BY SUBJECT ONLY

Please **print** your name, **sign**, and **date** below if you agree to be in the main study. By signing this consent and authorization form, you will not give up any of your legal rights. We will give you a copy of the signed form to keep.

Name of Subject

Signature of Subject (18 or older and able to consent) **Date** **Time**

Signature of Legally Authorized Representative with authority for research decisions **Date** **Time**

Authority of Legally Authorized Representative or Relationship to Subject

TO BE FILLED OUT BY STUDY TEAM ONLY

Name of Person Conducting Informed Consent Discussion

Signature of Person Conducting Informed Consent Discussion **Date** **Time**