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.
Title: Modeling the human neuronal phenotype of 3q29 deletion and 3q29 duplication syndrome

Principal Investigator: Jennifer G. Mulle, MHS, PhD, 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 you and 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 learn more about 3q29 deletion syndrome and 3q29 duplication syndrome. We do not yet understand the processes that give rise to many of these disorders. We will analyze results from behavioral assessments, a medical and physical exam, and an MRI, to understand these syndromes better. We will collect biospecimens like blood, DNA, cells, and tissue to help understand what is occurring in these disorders. We will create a “repository” or a collection of biospecimens and clinical information. This will be used to study basic causes of 3q29 deletion syndrome and 3q29 duplication syndrome and for future research. We plan to enroll about 150 people who either have a 3q29 deletion or duplication or have a family member who has the 3q29 deletion or duplication.

What will I be asked to do?
If you have the 3q29 deletion or duplication, this study should only take a few days of your time. We will look at your medical records if you are a patient at Emory University Hospital, any Emory Clinic, or Children’s Healthcare of Atlanta. If you are not a patient at one of these sites, we may ask you to sign a medical record release form to access your medical records.

You will travel to the Marcus Autism Center in Atlanta, Georgia for the behavioral assessment testing, medical exam, and MRI. This testing will take place over 2 days. If you are unable to travel to Atlanta, we may make arrangements to travel to you to conduct testing at a site convenient to your location. You will receive copies of your clinical assessments, to share with pediatricians and/or school administrators. These assessments will include tests to investigate your cognitive abilities, anxiety levels, and screen for certain psychiatric disorders such as autism and schizophrenia.
We will draw 4 vials (about 3 tablespoons) of blood from a vein in your arm. If we are unable to obtain a blood sample, we may need to take a saliva sample. We will ask for you to give one sample. Rarely we may need to ask for an additional sample if the first sample failed (such as cells did not grow or there was a technical problem with the sample).

With your permission, we will audio and/or videotape portions of the behavioral assessments and audiotape the medical history exam in case we need to confirm any observations after your visit. We will also take a photograph of your face and your profile. If you agree, the recordings and/or photos may be used in presentations or publications about this research study. The use of the recordings and/or photos may include professional conferences or other educational purpose related to this research study. The recordings and/or photos will not reveal your name or any other identifying information. Please initial below to indicate your permission:

_______ I give my permission to audio and/or videotape portions of the behavior assessments.

_______ 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.

If you are the parent or guardian of a person with the 3q29 deletion or 3q29 duplication, we will ask you questions about your family member with the syndrome. These questions will focus on his/her development along with past and current behavior.

If you are a parent/guardian or family member of a person with 3q29 deletion or 3q29 duplication, we will ask you to submit a blood sample. We will draw 4 vials (about 3 tablespoons) of blood from a vein in your arm. If we are unable to obtain a blood sample, we may need to take a saliva sample. We will ask for you to give one sample. Rarely we may need to ask for an additional sample if the first sample failed (such as cells did not grow or there was a technical problem with the sample). If you are traveling with the study participant for behavioral testing at Emory, we can draw your blood sample while you are there. If you are not traveling to Emory, we will arrange to have your blood drawn near your home. This can be at your doctor’s office, or a local clinic, lab or hospital. We may make arrangements to have a trained phlebotomist come to your home to collect the blood.

MRI Procedures:

MRI Safety Questionnaire: We will ask you to complete a MRI Safety Questionnaire before beginning the MRI scan. We do this to make sure that you are eligible for an MRI.

MRI Training: To help prepare for the fMRI scan you will complete an MRI training protocol. The training will take place at the Center for Systems Imaging at Wesley Woods, which is part of Emory Healthcare. The training will take place on a ‘mock scanner.’ This is a machine that looks like an MRI scanner but that does not collect any MRI data. You will have the opportunity to see and touch the MRI machine. You will also get a chance to practice all parts of the MRI study. This includes lying still, wearing headphones, entering the machine, etc. A training session may last up to two hours. MRI training will be performed by an experienced clinician or research assistant and may be recorded. If you experience anxiety or discomfort during the training session, or if you cannot lay still enough for effective MRI data collection, we will not proceed with the MRI portion of the study.

MRI Study: You will look at video clips of people talking and interacting. While watching the videos, pictures are taken of the brain using magnetic resonance imaging, or MRI. MRI is a safe technique. It uses strong magnetic fields and radio
waves to make pictures of your brain. These pictures can show what areas of the brain become active when you see pictures, hear sounds, and make simple judgments or decisions. The MRI Study will last between 30-120 minutes with breaks throughout as needed. It will take place at the Center for Systems Imaging at Wesley Woods. The experimenter will tell you what he/she will see or hear during this study so you will know what to expect. During the experiment, you will lie on your back on a narrow bed. The bed will be pushed into the MRI machine. You will get padding for your head and knees to make you more comfortable while lying down. Because the MRI machine makes a lot of noise when turned on, you will be given earplugs and/or headphones to wear. The technician will position your head and the bed will be pushed into the center of the MRI machine. You will be able to talk with the technician during the experiment using a microphone and speaker. We will watch you closely throughout the experiment. If eligible based upon the safety screener, a family member may present in the room during the MRI.

**Incidental Findings**
The fMRI study is for research only. It is not a clinical examination. The scans done as a part this study are not designed to find anything that may be abnormal. No one on the study team is qualified to interpret the scans. We are not responsible for providing a diagnostic evaluation of the images.

If a worrisome finding is seen on the scan, a trained clinician will be asked to review the relevant images. If they make a recommendation somebody from the study team or another physician will contact you. They will tell you about the finding. They will also recommend that you seek medical advice as a precautionary measure. The decision for additional examination or treatment would lie solely with you and your physician. The investigators, the consulting physician, the Center for Systems Imaging, and Emory University are not responsible for any examination or treatment that you receive based on these findings. The images collected in this study are not a clinical MR exam and for that reason, they will not be made available for diagnostic purposes.

You do not give up any rights by signing the consent form. The data collected by this study are for research purposes only. The data is then owned by the Emory/CHOA research groups. The cost and resources used are billed to this research group. Therefore, unless something of concern is found, we cannot provide raw data (MRI scans, assessments, videos.) If something does warrant concern, we will provide the appropriate information needed to help steer medical decision making to the medical group providing care. Their specialization will help define what information is essential to provide the utmost care.

We may contact you in the future by telephone, mail, or e-mail to ask for more information about your health, medical/family history, or additional research studies.

**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 unless you request that we destroy your sample, test results, or associated data. A code will be used to link your samples to your clinical information, your answers to the 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.

All biospecimens will also be deposited in the Rutgers University Cell and DNA Repository (RUCDR) resource for future studies. These may include genetic testing. Your blood sample may be used to create a living tissue sample (called a “cell line”) that can be grown in the laboratory. This allows researchers to have a large number of your cells in the future without collecting more samples from you. The researchers will not (and are not allowed to) use your samples for cloning a human being.
We may use your DNA sample to determine additional information about the 3q29 deletion or 3q29 duplication in your family. This may also uncover information about your carrier status. We will return this information to you if you are the participant and are over the age of 18. If your child is participating and is under 18, we will store this data and he/she may request this information once they are an adult. To request your results you should contact the Principal Investigator, Jennifer Mulle, PhD. However, this will be research results only and you would need to have these results confirmed by a diagnostic laboratory.

The materials that we collect including samples, medical information, video and audio recordings, photographs, MRI images, data, 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.

**What are the possible risks and discomforts?**

There may be side effects from the study procedures that are not known at this time. The most common risks and discomforts expected in this study are:

**Blood Draw:**

_The most common risks expected are_ slight soreness, bleeding or bruising at the point where the blood is taken, possible infection.

_A rare but possible risk_ of fainting or light-headedness may occur.

**MRI:**

Magnetic resonance (MR) uses magnetism and radio waves. It does not use x-rays. It takes pictures and measures chemicals of various parts of the body. The United States Food and Drug Administration (FDA) has set guidelines for magnet strength and exposure to radio waves. We carefully follow those guidelines. The participant will have to wear earplugs during scanning. This will reduce scanner noise levels below FDA limits.

_The most common risks expected are:_

The participant may become tired when completing the tests and interviews. Breaks will be given throughout the tests and interviews. The machine requires the participant to be in an enclosed space for a prolonged duration of time. If the participant is claustrophobic they may wish to opt out of the MRI portion of the study. Some people may feel uncomfortable or anxious during the MRI scan. If this happens, we will stop the study and take the person out of the MRI scanner.

_Rare but possible risks include:_

A possible but highly unlikely risk is that metal objects could be pulled into the magnet and hit the participant. To reduce this risk we make all people involved with the study remove all metal from their clothing and from their pockets. No metal can be brought into the magnet room at any time. Also, once you are in the magnet room, the door to the room will be closed so that no one from outside accidentally goes near the magnet.

On rare occasions, some people might feel dizzy, get an upset stomach, have a metallic taste or feel tingling sensations or muscle twitches. However, these sensations are rare and usually go away quickly.
If you have a contraindication for MRI: There are some risks with an MRI study for some people. People with a pacemaker cannot participate. People with metal objects inside their body also cannot participate.

Psychological Assessment and Clinical Evaluation:

The most common risks expected are:

The assessments to be used in the study have no known hazards and are comparable with testing procedures that have been used in previous psychological research. During clinical visits, the participant will be supervised and monitored by a parent or by study staff at all times. If a participant is upset, the procedure will be suspended. With parental permission, data gathering will continue when the individual has calmed down. The laboratories at the Marcus Autism Center are thoroughly childproofed. The researchers, clinicians, and research assistants who test the children and interact with the families have a background in child development and thorough knowledge of the experimental procedures.

A rare but possible risk is that, in the course of assessments, our clinical team determines that the participant is in serious danger of harming him/herself, or is at risk of harming others. If this happens, we will obtain help to make sure you and/or others are safe. We will engage the parent or caretaker in these decisions.

Privacy Risks: 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 measure that we will use, we cannot guarantee that your identity will never become known. Whenever possible, a study number, rather than your name, will be used on study records, your samples, and medical information.

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 will help you get medical treatment. Emory 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 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. Mulle at telephone number (404) 727-3042. You should also let any health care provider who treats you know that you are in a research study.

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 test results. 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.

**Storage and release of samples, genomic data, and health information**

Portions of your samples, genomic data, and health information will be stored and shared with other researchers. 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 regulated genetic databases. It is possible that we may share your de-identified or coded samples and cell lines with a regulated biobank such as the National Institute of Mental Health (NIMH). 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. 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 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 patient before, then you already have an Emory and Children’s Healthcare medical record. If you have never been an Emory and Children’s Healthcare patient, you do not have one. The results of these study tests and procedures will be used only for research purposes and will not be placed in your
medical record. 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 3q29 deletion and 3q29 duplication syndromes. The study results may be used to help others in the future. You will receive copies of clinical assessments, to share with pediatricians and/or school administrators. This may be potentially beneficial in developing early intervention programs, individual education plans (IEP), or may help qualify for services in your home state.

**Will I be compensated for my time and effort?**
Each participating family member will receive $150. You will be reimbursed for any travel expenses incurred to travel to the Marcus Autism Center.

**What are my other options?**
If you decide not to enter this study, there is care available to you outside of this research study. You do not have to be in this study to be treated for your medical issues.

**Costs**
There will be no costs to you for participating in this study. You will not be charged for any of the research activities. You will be reimbursed for your travel to the Marcus Autism Center.

**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 this study

**PHI that Will Be Used/Disclosed:**
The PHI that we will use or share for the 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.

**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 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 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 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 IRBs, the Emory Research and Compliance Offices, and the Emory Office for Clinical Research.
  - Rutgers
  - 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.

**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 Principal Investigator in writing at: Jennifer Mulle, PhD, Department of Human Genetics, Emory University School of Medicine, 615 Michael Street Room 305M, Atlanta, GA, 30322

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 the Principal Investigator, Jennifer Mulle, PhD, at 404-727-3042 or jmulle@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](http://www.surveymonkey.com/s/6ZDMW75).

If you are a patient receiving care at Children’s Healthcare 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 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 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