
Emory University Consent to be a Research Subject

Title: Characterization of 3q29 Deletion Syndrome and 3q29 Duplication Syndrome

Principal Investigator: Jennifer Mulle, MHS, PhD, Assistant Professor, Department of Epidemiology, Rollins School of Public Health

If you are the parent or 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 research study. This form is designed to tell you everything you need to think about before you decide to consent (agree) to be in the study or not to be in 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 and your child.

Before making your decision:

- Please carefully read this form or have it read to you
- Please ask questions about anything that is not clear

You can print 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.

Study Overview

The purpose of this study is to understand the medical and behavioral consequences of 3q29 deletion syndrome and 3q29 duplication syndrome. In this study, we are looking for people who have been diagnosed with 3q39 deletion syndrome or 3q29 duplication syndrome (these are sometimes called “3q29 microdeletion” or “3q29 microduplication”). We want to understand how these people grow and develop. We will use a set of questionnaires to ask about the behaviors of individuals with 3q29 deletion syndrome or 3q29 duplication syndrome. We will also ask for a copy of their clinical genetics report, with details about how they were diagnosed with 3q29 deletion syndrome or 3q29 duplication syndrome. We will also survey a set of people without 3q29 deletion syndrome or 3q29 duplication syndrome, as a comparison group. If you are the parent of an individual with 3q29 deletion syndrome or 3q29 duplication syndrome, we may ask you to complete the same set of questionnaires for your other children that are siblings of the individual with the 3q29 deletion or 3q29 duplication.

Procedures

If you agree to be in this study, we will ask you to complete a series of online questionnaires asking about medical conditions, development, and behavior in the individual with 3q29 deletion syndrome or 3q29 duplication syndrome. There will be a total of six questionnaires if your child is under the age of

12. If your child is over the age of 12, there will be one additional questionnaire (seven total). These questionnaires can be answered on the study website. Each questionnaire will take between 15-30 minutes to answer. If you would prefer to answer the questions on paper instead of online, we can mail the questionnaires to you and provide postage for you to mail the answers back to Emory.

We will also ask for a copy of the clinical genetics report (the report you received from your doctor) when your child was diagnosed with 3q29 deletion syndrome or 3q29 duplication syndrome. You will be able to scan and upload the report directly to the study website, or you can make a copy and mail it to Emory. If you do not have a copy of your report, we can help you get one. This report will give us information about how your child was initially diagnosed with 3q29 deletion syndrome or 3q29 duplication syndrome. The investigator and research staff are the only people who will view this clinical report.

We will recontact you at the email address you provide to us on a yearly basis for the next five years and ask you to fill out new online forms about how the individual with 3q29 deletion or 3q29 duplication is developing. These questionnaires will be the same as or very similar to the questionnaires you will fill out in the beginning of the study. We will collect this information to learn about the pace of development in individuals with 3q29 deletion syndrome or 3q29 duplication syndrome. We may recontact you in the future to expand the study to additional family members.

We will also ask you to upload some short videos of the individual with 3q29 deletion or 3q29 duplication, from when they were 0- 3 years old. These videos could have been taken with a camera or a cell phone. We want to view these videos to observe how 3q29 deletion individuals communicate and display emotion as babies or very young children. These videos will only be viewed by the research staff. They will never be shared outside of this study.

Risks and Discomforts

The risks associated with this study are believed to be minimal.

The most common risks and discomforts expected in this study are: It is possible you may become upset or uncomfortable by some of the questions asked. If this happens you are free to take a break until you feel better. If you are still upset, you may decide to skip any question that you do not want to answer or to stop the questionnaire.

Rare but possible risks include: The information that you give us will be kept strictly confidential. Although every measure is taken to maintain confidentiality, there is a potential risk for a loss of confidentiality. We will make every effort to minimize this risk.

New 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.

Benefits

This study is not designed to benefit you directly. This study is designed to learn more about the features of 3q29 deletion syndrome and 3q29 duplication syndrome. The study results may be used to help others in the future.

Compensation

You will receive a \$10 gift card for each completed questionnaire. If you do not finish the study, you will be paid for the questionnaires you have completed. You will receive \$60-\$70 total, if you complete all questionnaires for which you are eligible. Your compensation can only be released when we receive your clinical genetics report.

Confidentiality

Certain offices and people other than the researchers may look at your medical charts and study records. Government agencies and Emory employees overseeing proper study conduct may look at your study records. These offices include the Emory Institutional Review Board and the Emory Office of Research Compliance. Emory will keep any research records we create private to the extent we are required to do so by law. A study number rather than your name will be used on study records wherever possible. Your name and other facts that might point to you will not appear when we present this study or publish its results.

Study records can be opened by court order. They may also be produced in response to a subpoena or a request for production of documents.

Any identifiable information that is collected during this study will be confidential. This information will be disclosed only with your permission or as required by U.S. or State law. Examples of information that we legally have to disclose include abuse of a child or elderly person, or certain reportable communicable diseases.

In order to make sure that your information is kept confidential, all records about you and your child in this study (including video data) will be stored on a protected (HIPPA-compliant) server that can only be accessed by the investigators collaborating on this study and their research staff. At no time will we discuss any of the information that you provide with anyone outside the research team. If you specifically request us to provide information about you to a third party, you must provide written permission.

When the results of the research are published or discussed in conferences, no information will be included that would reveal your identity or your child's identity. This information will only be accessible to the investigators collaborating in this study and their research staff.

Research Information Will Not Go Into the Medical Record:

Emory does not control results from tests and procedures done at other places. Thus, these results would not be placed in your child's medical record at Emory or CHOA. Emory also does not have control over any other medical records that your child may have with other providers. So if you decide for you and your child to be in this study, it is up to you to let them know.

The researchers will review the results of certain study tests and procedures only for this research. For this study, only experimental data will be collected thus there will not be any information available to share with you.

Costs

There will be no costs to you for participating in this study, other than basic expenses like internet use. 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. You are free to choose not to take part in this study, and if you do become a subject, you are free to stop and withdraw from this study at any time without penalty. To withdraw, you can revisit this online consent form and uncheck the consent box. You can also call a member of the research team at any time and tell them you no longer want to take part. You may also contact the Principal Investigator using the contact information provided below.

The researchers also have the right to stop your participation in this study without your consent if:

- They believe it is in your best interest;
- or for any other reason.

Contact Information

Contact Jennifer Mulle at 404.727.3042 or jmulle@emory.edu:

- if you have any questions about this study or your part in it,
- if you feel you have had a research-related injury, or
- 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>.