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Rutgers Biomedical and Health Sciences
671 Hoes Lane West
Piscataway, NJ 08854

CONSENT TO TAKE PART IN A RESEARCH STUDY

Title of Study: The 3q29 deletion and 3q29 duplication: genetic architecture of behavioral phenotypes

Principal Investigator: Jennifer G. Mulle, MHS, PhD

STUDY SUMMARY: This consent form is part of an informed consent process for a research study and it will provide information that will help you decide whether you want to take part in this study. It is your choice to take part or not.

The **purpose of the research** 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 and interviews to understand these syndromes better. We will also collect biospecimens like saliva 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.

If you have the 3q29 deletion or duplication: this study should only take a few days of your time. We will conduct behavioral assessment testing remotely, while you are in the comfort of your home. This testing will take the form of computerized tests, questionnaires, and interviews with our study team.

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

If you choose to participate, you will be asked to provide a saliva sample.

If you are the parent or family member of a person with the 3q29 deletion or 3q29 duplication: This study should only take a few days of your time. We may ask you questions about your family member with the syndrome, including his/her development as well as past and current behavior. You may also be asked to complete surveys about your family and medical history. We may also ask about characteristics of your family, personal medical history and history of medical and genetic conditions that your family may have.

We will also ask you to complete a testing battery, which may include computerized tests, questionnaires, and interviews with our study team. We will conduct this testing remotely, while you are in the comfort of your home.

If you choose to participate, you will be asked to provide a saliva sample.

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.

With your permission, we will audio and/or videotape portions of the behavioral assessments in case we need to confirm any observations after your evaluation.

Possible harms or burdens of taking part in the study:
The proposed protocol includes only minimal risk procedures. However, there may be side effects from the study procedures that are not known at this time.

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.

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.

An alternative to taking part in the research study Your alternative to taking part in the research study is not to take part in it. 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.

The information in this consent form will provide more details about the research study and what will be asked of you if you choose to take part in it. If you have any questions now or during the study, if you choose to take part, you should feel free to ask them and should expect to be given answers you completely understand. After your questions have been answered and you wish to take part in the research study, you will be asked to sign this consent form. You are not giving up any of your legal rights by agreeing to take part in this research or by signing this consent form.

Who is conducting this study?
Jennifer G. Mulle, MHS, PhD is the Principal Investigator of this research study. A Principal Investigator has the overall responsibility for the conduct of the research. However, there are often other individuals who are part of the research team. Dr. Mulle may be reached at (848) 445-9866 and address 679 Hoes Lane West, Piscataway, NJ, 08854.

The Principal Investigator or another member of the study team will also be asked to sign this informed consent. You will be given a copy of the signed consent form to keep.

Sponsor of the Study: National Institute of Mental Health

Why is this study being done?
This study is being done to learn more about 3q29 deletion syndrome and 3q29 duplication syndrome.

Who may take part in this study and who may not?
Individuals with the 3q29 deletion or the 3q29 deletion may take part in this study. Parents of children with the 3q29 deletion or 3q29 duplication may also take part in this study.

Why have I been asked to take part in this study?
You are being asked to be in this research study because you or one of your family members has 3q29 deletion syndrome or 3q29 duplication syndrome.

How long will the study take and how many subjects will take part?
Participation in this study should only take a few days of your time, and a few hours each day. We will enroll 200 people with the 3q29 deletion and 50 people with the 3q29 duplication. We will also enroll their parents, for a total of up to 750 people in this study.

What will I be asked to do if I take part in this study?
If you have the 3q29 deletion or duplication: this study should only take a few days of your time.

We will conduct behavioral assessment testing remotely, while you are in the comfort of your home. This testing will take the form of computerized tests, questionnaires, and interviews with our study team. Testing will take place over several days. 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.

Videos of these evaluations will be made to record you during these sessions, in case we have any questions about the data. This way we won't have to take any more of your time, we can check the recordings instead. These recordings will only be viewed by our study team, we will never show them to anyone outside the study.

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

If you choose to participate, you will be asked to provide a saliva sample. In rare situations we may ask you to agree to a draw blood (about 4 tablespoons. 4-5 vials) from a vein in your arm. We will ask you to give one sample. Rarely we may need to ask for an additional sample if the first sample failed (such as if there was a technical problem with the sample).

We will send you a saliva kit and instructions for collecting the sample. If you are willing, we may arrange to have your blood drawn near your home. This can be at your doctor's office, or a local clinic, lab or hospital.

If you are the parent or family member of a person with the 3q29 deletion or 3q29 duplication: This study should only take a few days of your time. We may ask you questions about your family member with the syndrome, including his/her development as well as past and current behavior. You may also be asked to complete surveys about your family and medical history. We may also ask about characteristics of your family, personal medical history and history of medical and genetic conditions that your family may have.

We will also ask you to complete a testing battery, which may include computerized tests, questionnaires, and interviews with our study team. We will conduct this testing remotely, while you are in the comfort of your home. These assessments will include tests to investigate your cognitive abilities, anxiety levels, and screen for certain psychiatric disorders such as autism and schizophrenia. The purpose of this is to understand how individuals with the 3q29 deletion or 3q29 duplication are similar to their biological relatives, and how they are different.

If you choose to participate, you will be asked to provide a saliva sample. In rare situations, we may ask you to agree to a draw blood (about 4 tablespoons. 4-5 vials) from a vein in your arm. We will ask you to give one sample. Rarely we may need to ask for an additional sample if the first sample failed (such as if there was a technical problem with the sample).

We will send you a saliva kit and instructions for collecting the sample. If you are willing, in rare situations we may 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 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.

With your permission, we will audio and/or videotape portions of the behavioral assessments in case we need to confirm any observations after your evaluation. The recordings and/or photos will not reveal your name or any other identifying information.

What are the risks of harm or discomforts I might experience if I take part in this study?
The proposed protocol includes only minimal risk procedures. However, 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 would result from a blood draw, which is optional and only performed in rare situations. If a blood sample is taken, the most common risks and discomforts are slight soreness, bleeding or bruising at the point where the blood is taken, and possible infection. A *rare but possible risk* of fainting or light-headedness may occur.

This study will conduct genetic research. There are some unique risks associated with genetic research, as follows:

Psychological or Social Risks Associated with Loss of Privacy:
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 identify you. Similarly, it may be possible that genetic information from you could be used to help identify them. Further, patterns of genetic variation also can be used by agencies to identify a person or his/her blood relatives (for example, to establish relationships between parents and their children).

Economic Risks of Harm:
Since some genetic variations can help to predict the future health problems of you and your relatives, this information might be of interest to health providers, life insurance companies, and others. Therefore, your genetic information potentially could be used in ways that could cause you or your family economic distress.

There is a federal law call the Genetic Information Nondiscrimination Act (GINA) that helps protect against genetic discrimination. In general, this law 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: (1) health insurance companies and group health plans may not request your genetic information that we get from this research; (2) health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums; and (3) 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. However, it does not protect you against discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

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.

Are There Any Benefits To Me If I Choose To Take Part In This 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.

What Are My Alternatives If I Do Not Want To Take Part In This Study?
Your alternative is not to take part in this study. 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.

How Will I Know If New Information Is Learned That May Affect Whether I Am Willing To Stay In The Study?
During the study, you will be updated about any new information that may affect whether you are willing to continue taking part in the study. If new information is learned that may affect you after the study or your follow-up is completed, you will be contacted. 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 Receive The Results Of The Research?
We will give you copies of the results from behavioral assessments, which may be useful to you and may be shared with teachers and/or physicians. Except for information about carrier status, in general, we will not give you any individual results from the study of the samples you give us.

Will There Be Any Cost To Me To Take Part In This Study?
There will be no costs to you for participating in this study. You will not be charged for any of the research activities.

Will I Be Paid To Take Part In This Study?
Each participating family member will receive $100 for participating in behavioral assessments and $25 for providing a saliva or blood sample. If you do not finish the study, we will compensate you for the assessments you have completed.

How Will Information About Me Be Kept Private Or Confidential?
All efforts will be made to keep your personal information in your research record confidential, but total confidentiality cannot be guaranteed.

The research team may use or share your information collected or created for this study with the following people and institutions:
- The Rutgers University Institutional Review Board and Compliance Boards
- The Office for Human Research Protections in the U.S. Dept. of Health and Human Services

This study has a Certificate of Confidentiality, which affords special protections to the data. 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. Rutgers will rely on the Certificate of Confidentiality to refuse to give out study information that identifies you. For example, if Rutgers 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 Rutgers 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.

Rutgers University 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.

What Will Happen To My Information—data, recordings and/or images—And Biospecimens Collected For This Research After The Study Is Over?
After information that could identify you has been removed, de-identified information or biospecimens collected for this research may be used for other research we conduct without obtaining additional informed consent from you.

De-identified data from this study (data that has been stripped of all information that can identify you), including your de-identified genetic information, 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. 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.

Your data and specimens from this study may be useful for other research being done by investigators at Rutgers or elsewhere. To help further science, we may provide your deidentified data and/or specimens to other researchers. If we do, we will not include any information that could identify you. If your data or specimens are labeled with your study ID, we will not allow the other investigators to link that ID to your identifiable information.

We will use your sample and data only for research. We will not sell them. However, the results of this research might someday lead to the development of products (such as a commercial cell line, a medical or genetic test, a drug, or other commercial product) that could be sold by a company. You will not receive money from the sale of any such product.

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 Rutgers University in secure databases and biospecimen banks.

Some biospecimens may 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 can 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 can provide this information to a parent/guardian or store these data for the child to request once they are an adult. Any information obtained 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, 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 the age of 18 years, 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.

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 years of age, 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 turns 18 years old.

What Will Happen If I Do Not Wish To Take Part In The Study Or If I Later Decide Not To Stay In The Study?

It is your choice whether to take part in the research. You may choose to take part, not to take part or you may change your mind and withdraw from the study at any time. If you do not want to enter the study or decide to stop taking part, your relationship with the study staff will not change, and you may do so without penalty and without loss of benefits to which you are otherwise entitled.

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. You may also withdraw your consent for the use of data already collected about you, but you must do this in writing to Dr. Jennifer Mulle, jennifer.mulle@rutgers.edu, 679 Hoes Lane West, Piscataway, NJ 08854.

Who Can I Contact If I Have Questions?

If you have questions, concerns or complaints about the research, wish more information or if you feel you may have suffered a research related injury, you can contact the Principal Investigator: Dr. Jennifer Mulle, Department of Psychiatry, jennifer.mulle@rutgers.edu.

If you have questions, concerns, problems, information or input about the research or would like to know about your rights as a research subject, you can contact the Rutgers IRB Office at: (973) 972-3608 or (732) 235-9806 or (732) 235-2866, or email us at IRBOffice@research.rutgers.edu, or write us at 335 George Street, Liberty Plaza Suite 3200, New Brunswick, NJ 08901.

OPTIONAL AUDIO/VIDEO RECORDING

Do you agree to having portions of the behavioral assessments audio and video recorded?

☐ Yes, I agree to being audio/video recorded.

☐ No, I do not agree to being audio/video recorded.

RETURNING INFORMATION ABOUT CARRIER STATUS

Do you wish to be informed about your 3q29 deletion or 3q29 duplication carrier status? Any results are for research purposes only and would need to be confirmed by a diagnostic laboratory.

☐ Yes, I want to receive information about my carrier status.

☐ No, I do not want to receive information about my carrier status.

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AGREEMENT TO PARTICIPATE

Subject Consent:

I have read this entire consent form, or it has been read to me, and I believe that I understand what has been discussed. All of my questions about this form and this study have been answered. I agree to take part in this study.

Subject Name (Print):_____

Subject Signature:_____ Date:_____

Legally Authorized Representative with Authority for Research Decisions:

Legally Authorized Representative Name (Print):_____

Signature:_____ Date:_____

Signature of Investigator/Individual Obtaining Consent:

To the best of my ability, I have explained and discussed all the important details about the study including all of the information contained in this consent form.

Investigator/Person Obtaining Consent Name (Print):_____

Signature:_____ Date: _____