Regione Siciliana
Azienda Ospedaliera
OSPEDALI RIUNITI VILLA SOFIA – CERVELLO
UOSD Genetica Medica
Legge Regionale n.5 del 14/04/2009
Centro Riferimento Regionale per il controllo e la cura della Sindrome di Down e delle altre patologie cromosomiche e genetiche
Responsabile: Prof.ssa Maria Piccione. Professore Associato in Genetica Medica. Università degli Studi di Palermo
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PARENTAL PERMISSION

"***DE***ciphering ***N***eurodevelopmental ***D***isorde***R***s through the ***ITE***ms of undiagnosed rare diseases"

- The DENDRITE Study -

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Researchers' statement

We are asking your permission for your child to be in research sturdy and to answer questions about your family. This consent form gives you the information that you will need to help you decide whether you consent to having your child in the study or not. Please read the form carefully. You may ask questions about the purpose of the research, what we are asking your child to do, the possible risks and benefits, your child's rights as a participant, and anything else about the research or this consent form. When we have answered all of your questions, you can decide if you want to be in the study or not. We will give you a copy of this form for your records.

PURPUSE OF THE STUDY

This study is exploring how genes may affect the physiology of neurodevelopment. Genes are made of material called DNA, which carries instructions for making the body's protein. Genes determine

some of our characteristics, such as eye color. The purpose of the study is to explore genetic differences that may be related to neurodevelopmental disorders.

STUDY PROCEDURES

This study will take about 30 minutes. For participants we will collect 2 tubes of blood and/or 2 vials of saliva from the proband and his/her parents. We will collect demographic background information about your child and family pedigree information. We will also ask you for the names and contact information of people who will know how to contact you. We will use the blood and/or saliva samples to study your child's genes. Your child's DNA may also be used for future research projects that study genes, neurodevelopmental disorders and rare disease.

This study might identify genetic differences in your child's DNA that are related to neurodevelopmental disorders and rare diseases. If, and only if, a genetic difference related in this field is found, we will offer to share these results with your child's doctor, or we will provide your child with a referral to see a medical geneticist. We, the researchers, are not able to directly share any genetic results with you. If genetic results are shared with your child's doctor, this information should be seen as preliminary. If this information is not of interest to you, you may choose not to receive it.

RISKS, STRESS, OR DISCOMFORT

The primary rick of being in the study is potential loss of confidentiality. This means that someone might find out that your child has been in this study and could find out other information about them. We will do everything we can to prevent this from happening.

Your child may experience mild discomfort with the blood draw. After the procedure, they may feel soreness or tingling. They may experience bleeding, a small bruise, clot, or infection at the site. In rare situations, the blood draw may cause your child to feel tired (fatigue), and lightheadedness and/or fainting may occur. A minority of participants may feel nauseous and or vomit during or after the blood draw. A trained phlebotomist will conduct the blood draw. Care will be taken to avoid all complications.

Your child may choose to take breaks, decline to participate in a procedure, or stop your child's participation at any time

ALTERNATIVES TO TAKING PART IN THIS STUDY

The alternative to participation in this study is choosing not to participate. If you would like more information about neurocognitive clinical assessment or other studies investigating the genetic cause of neurodevelopmental disorders and rare diseases in general, you can contact the UOSD Genetica Medica, AOOR Villa Sofia Cervello – Palermo, Italy.

BENEFITS OF THE STUDY

There will be no direct benefit to your child form participating in this study. However, we hope that our study might help us better understand how genes impact the neurodevelopment in rare diseases as well as in idiopathic forms.

CONDIFENTIALITY OF RESEARCH INFORMATION

Your child's name and other identify information will be assigned a code number. Only the code number will be put on the information that we collect. We will keep the link between the code number and your child's identify indefinitely, but only the research team will have access to that link. All the information we will collect will be kept indefinitely on secure, password-protected computers and in locked filing cabinets.

Your child's DNA samples will also be labeled with the code number and will be securely stored indefinitely at the UOSD Genetica Medica, AOOR Villa Sofia Cervello – Palermo, Italy and could be shared across National and International Consortia who are conducting the same field research. All data will be kept indefinitely by the UOSD Genetica Medica, AOOR Villa Sofia Cervello – Palermo, Italy. Data and samples may be shared with collaborators at other institutions; there will be no links to identifying information given to other researchers.

If we feel that your child is in danger of hurting him-or-herself or another person, or is in danger of being hurt by a family member, we are obligated by law to report this information to the appropriate state agency.

University and government offices sometimes review studies such as this one to make sure they are being done safely and legally, including institutional oversight review offices at the research site, or state; and federal regulators. If a review of this study takes place, your child's records may be examined. The reviewers will protect your child's privacy. The study records will not be used to put your child at legal risk of harm.

Information collected from your child could be also shared with other National and/or International Institutions and/or Registries across consortia (such as the USA National Institute of Health - NIH). This will include information about your child that we will collect as part of this study including diagnostic information and genetic data. This information will be stored in a de-identified, coded, way to keep your child's personal identity a secret. Furthermore, the de-identified information could be shared also with an NIH repository called Database of Genotypes and Phenotypes (dbGaP). dbGaP was developed to archive and distribute the results of studies that have investigated the interaction of genotype and phenotype to qualified scientists. In this case and in order to allow researchers to share results, the NIH and other central repositories have developed special data (information) banks that collect the results of studies that have investigated genotype and phenotype. The NIH will store your child's coded genetic information and give it to other qualified researchers to do more studies. Qualified researchers that can access the national databases may be from the government, academic, or commercial institutions. We cannot predict how genetic data will be used in the future. Research using genetic information is important for the study of virtually

all disease and conditions. Therefore, the databank may provide study data for researchers working on any disease or condition.

We do not believe that there will be any further risk to your child's privacy and confidentiality by sharing your child's genetic information with these databanks; however, we cannot predict how genetic information will be used in the future. The information will be sent with only a code number attached. Your child's name and other identifiable information will never be given to the NIH. There are many safeguards in place to protect your child's information while it is stored in repositories and used for research.

There is a small chance that your child's genetic information could be shared with others by mistake. In the unlikely event that your child's information was mistakenly shared and if it were to be linked with a medical condition, this could affect your child's ability obtain or maintain some kinds of insurance. There is also the risk that data could be released to the public, employers, or law enforcement agencies. If family members were to see this information it could affect them as well. This could potentially hurt family relationships. It is also possible that your child could be identified from the DNA sample if someone else has another DNA sample from your child. The two samples could be matched to identify you from the sample given for this study. Strict precautions are taken to prevent these risks, which include de-identifying all data and storing and sharing data in a secure manner.

You will not receive any results from allowing your child's data to be placed in the national databases unless it is considered medically relevant.

You can withdraw your consent at any time if you do not want your child's data in the national database. There will be no consequences for withdrawing consent. However, data that has already been sent to other researchers cannot be retrieved from those researchers.

No direct benefit can be promised from allowing your child's code information to be shared with national databases, though some people might find satisfaction in contributing to scientific knowledge about genetic problems and medical conditions.

OTHER INFORMATION

Your child may refuse to participate in the study, or in any procedures described above, and you are free to withdraw your child from this study at any time without penalty and loss of benefits to which your child is otherwise entitled.

RESEARCH-RELATED INJURY

If you think your child has an injury or other harm related to this study, contact Prof.ssa Maria Piccione at +39 091680282 or maria.piccione@unipa.it right away.

_____

Printed name of study staff obtaining consent Signature Date

Subject's statement

This study has been explained to me. I volunteer for my child to take part in this research. I have had a chance to ask questions. If I have questions later about the research, I can ask one of the researchers listed above. If I have questions about my child's or my rights as a research subject, I can call the UOSD Genetica Medica at +39 091680282. I will receive a copy of this consent form.

Genetic Results

_____ If a genetic difference related to ASD is discovered during this study, I give permission to be informed and for genetic results to be sent to my child's doctor or to be referred to a medical geneticist.

_____ If a genetic difference related to ASD is discovered during this study I do not want to be informed.

Re-contact

_____ I agree to be re-contacted for future studies

_____ I do not agree to be re-contacted for future studies

_____ _____

Printed name of child Printed Name of Parent/Primary Caregiver

_____

Printed name of child's siblings

_____ _____

Signature of Parent/Primary Caregiver Place, Date

Copy to: Researcher, Subject