

## Information Sheet: Parent/Guardian of Control (v2)

### **TITLE: Genetic Characterization of Neurodevelopmental Disorders in South African Populations (NeuroDEV South Africa)**

REFERENCE NUMBER: 810/2016

INVESTIGATOR: Dr. Kirsten Donald

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Your child is invited to take part in a research study. Please take some time to read the information here. Please ask any questions about any part of this project that you do not understand. It is very important that you feel you understand what this research is about and how you could be involved. Also, being in the study is voluntary. You are free to choose not to join. If you say no, this will not affect you negatively in any way whatsoever.

This study has been approved by the University of Cape Town Human Research Ethics Committee (HREC ref 801/2016) and the Harvard School of Public Health, in the United States of America. The study will be carried out according to the highest ethical standards.

#### **What is the study about?**

This study looks at development in childhood. The study will try to find out about genes that may be related to why some children develop differently than others. We will be asking over 5,000 people to take part in this study. The reason is to better understand the cause of delays in development and other social and thought problems and make better treatments.

#### **Background**

Our body is made up of many tiny parts called cells. These cells contain DNA, which tell the cells how to grow and work. We get our DNA from our parents. Nobody else in the world has the same DNA as you, unless you are a twin. Differences in DNA can be why some people are tall and others are short. Some illnesses are caused by problems with DNA. The study of DNA is called genetics. DNA can be found in blood and in saliva.

A cell line is a culture made from cells of the same type. Under the right conditions, these cells can make copies of themselves. So without having to draw blood again, researchers can keep studying the cells in your blood for many years. This makes cell lines very useful. A stem cell is a special type of cell. Stem cells can form many different types of cells in the human body. Stem cells are made of the same DNA as all other cells in your body. A stem cell made from your blood is called an induced pluripotent stem cell (iPS cell).

#### **Do I have to take part?**

It is your choice for you and your child to be in this study. This study does not have anything to do with the care you or your child receives at this hospital or school. If you do not want to answer some questions or if you do not want some tests to be done you can tell us. If you would like to stop at any time, you can. You can say "yes" and join the study; or you can also say "No," you don't want to join.

### **What does being in this study include?**

If you choose to join your child in this study, we will ask you some questions about your child to assess your child's behavior, reasoning and movements. We will also examine your child's body and look at your child's medical records to find out about his or her health. The whole process could take up to 2 hours. We would be happy to provide the names of the tests and explain these tests to you before we start.

A nurse will ask your child to give some blood. Your child will give up to two teaspoons (10 ml) of blood. If you agree, we may also ask for your child to give us some saliva in a small plastic tube. If we collect saliva, we will ask for less than half a teaspoon (2 ml or less) of saliva. DNA is in blood and saliva, which we call a 'sample'.

The sample will be sent for DNA processing in the United States of America. We will analyse your child's DNA using chemicals and computers. Then we will compare your child's DNA to the DNA of other children in the study to look for differences. No one testing your child's DNA will know your child's name.

You also have the choice of contributing your child's sample for cell line and stem cell research. Stem cells are important because they can become all types of cells in the body, such as muscle cells, brain cells or heart cells. Stem cells also can make copies of themselves, and for this reason a stem cell line growing in a lab could exist for a very long time, even longer than your child's lifetime.

With your permission, we will also photograph your child's face, hands and feet. The photographs are to help doctors and other health experts recognize children with delays in development. Your child does not have to be photographed to be in the study. If you agree to have your child photographed, the photos will be used for clinical, educational and research purposes.

### **Storage and sharing of your information**

With your permission, your child's sample and health information will be sent to and stored in the United States of America. Some of the sample may also be stored here in South Africa. Your child's donated DNA can last a very long time. The samples will be stored and may be used for other tests in the future. Access to your child's DNA will continue to be guarded into the future by a set of strict controls to make sure that it is only used to study questions that are important to science.

#### *Creating a cell 'bank' or library*

The samples will be stored at the National Institutes of Mental Health Repository and Genomics Resource (NIMH-RGR). This is a 'bank' or library for biological samples funded by the department of health in the United States government. Some of the samples will also be stored in South Africa at the Department of Human Genetics in the University of Cape Town. That way, your and your child's samples can be shared with other researchers at universities, hospitals, government agencies and companies around the world. One place your DNA will be shared with is the Broad Institute, a non-profit American research institute with skilled scientists who know how to analyze DNA.

If you agree for your child to be part of the cell line research, their cells and associated health information will be stored in the cell library and distributed for a long time, possibly forever. None of the researchers who study your cells or health information will know your name.

The samples and information will be available to scientists all around the world, to study for any research question. Examples include research to understand the causes of certain diseases (for example heart disease,

cancer, or psychiatric disorders). Other examples include research to develop new methods, to study human behaviors, or to find out where different groups of people may have come from.

If you agree for you child to participate in the cell line and stem cell research, their samples could be of even greater use. Researchers around the world may use your child's cells to study the biology of stem cells, certain illnesses, and whether it is possible to transplant stem cells (or things made by those cells) to treat diseases. Common examples of what might happen to the stem cells include the following:

- Testing the cells' DNA
- Using cells to test or select drugs to treat disease
- Changing some of the genetic code or sequence within these cells
- Transplanting the cells or things made by those cells to humans or animals
- Sharing cells widely for research, training or to make commercial medical products

Because these samples can last forever, scientists may come up with new ways of working with them in the future. Right now, we do not know what the future uses may be.

#### Creating a photo library

If you allow us to photograph your child, we will use the photos to compare them with children who have malformations of the face, hands or feet. Some photos will be analyzed using computers. For example, the photos may be shared on a mobile application that will improve future technology for diagnosing children with delays in development. One example of a mobile application this kind is Face2Gene. If a photo is shared in this way, a computer breaks it up into thousands of bits of information and compares it to other photos. When the photo is broken up in this way, it cannot be used to identify your child.

If you agree to having your child's photos taken, they will be shared together with some pieces of health information (e.g. age, ethnicity & diagnosis).

#### Putting information in public databases

Some of the genetic and health information from this study will be freely available in public databases that anyone can use. An example of these databases is the Genome Aggregation Database (GnomAD). These public open-access databases may include information about your DNA, as well as your ethnic group and sex. This public information will not be labeled with your name or other information that could identify you.

Your child's DNA and health information will be put in one or more 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 it. Your child's DNA and health information will not be labelled with his or her name or other information that could be used to identify him or her.

Although we will not give researchers your child's name, we will give them basic information such as his or her race, ethnic group, geographic region, age range, and sex. We will also provide them with the medical information collected in this study. This information may help researchers study whether the factors that lead to health problems are the same in different groups of people. It is possible that such findings could one day help people in the same groups as your child.

#### Securing information on a group of computers

Your child's DNA and health information will not be labelled with his or her name or other information that could be used to identify him or her. To ensure safety, the information will be stored and analyzed in the Cloud. "The Cloud" refers to a group of computer servers in a data center that is accessible through the Internet. The Cloud is managed by a company or other institution that is not part of the research team. The study information will be stored on a secure Cloud server to prevent unwanted access. The company or other institution that will process and store your data is located in the United States. It may also come to be located in other countries where the laws may not protect your privacy to the same level as the laws in your country. However, all reasonable steps will be taken to protect your privacy.

#### **What if I change my mind?**

If your child joins the study, you can change your mind later and decide that you don't want your child to be part of it anymore, and you do not want your child's blood or saliva to be used. Please let us know and we will destroy the sample. If your child's sample has already been tested at the time you change your mind, your results and other data may have already been shared with other investigators. In that case, we will not be able to destroy this data. However, the code that links your child's name to their information and samples can be destroyed. Whether or not you decide to join the study, the way we look after your child in the clinic or at

school will be the same. It is your choice whether to be in the study or not.

## **Benefits and Risks**

### *Benefits*

There are no direct benefits to you or your child for taking part in this study. One reason somebody might want to share their child's DNA without direct benefit is that it may help scientists to learn more about the human body, and better understand problems of child development. We hope this will help make better treatments in the future.

### *Risks*

The risks of taking blood include pain, as with a pin prick, a bruise at the point where the blood is taken, and redness and swelling of the vein. There is also a rare risk of infection, if the area is not kept clean, and a rare risk of fainting.

When answering questions about yourself or your child, you may experience distress or discomfort. You do not have to answer questions if they make you uncomfortable. You may also ask to take a break at any time.

There are some risks related to the use of your child's information and samples. It is possible that someone could break into the system that stores information about your child. All the computers with study results are protected by several strong passwords, so this is very unlikely. . It is also possible that your child could be identified through the photographs, which will be stored on the Cloud. Security of the Cloud is high, so breaking into the Cloud is very unlikely. There may be other privacy risks that we do not know.

If someone has another DNA sample from your child, which has not been protected, it is possible that your child could be identified from their sample. This would happen if the two samples were matched, and one of them was linked to your child's name. The risk of this happening is very small but may grow in the future as genetic processing is more widely used.

## **Privacy**

The information we get from you and your child will not be shared with your health insurance. Nobody outside the study will be able to know which results belong to your child. The researchers will not tell anyone else that you are in the study.

## **Reimbursement**

You will be reimbursed for your time, travel costs and the inconvenience of taking part in this study. Once you have been interviewed and your child has given his or her blood, the study staff will reimburse you. You will be reimbursed 250 Rand. As a thank you, we will also give your child a goodie bag, which may include a small toy, snacks and a polaroid photo of your family.

The control over the DNA your child donates will be held by the University of Cape Town and The Broad Institute. Your child's samples will not be sold, but investigators may make products based on studying your child's samples, images or information. If this happens, you will not be able to share in any profits. This is not likely to happen for many years, if at all.

## **Future Contact**

Individual results from this research project will not be given back to you or put into your child's medical records, but we may learn something from this study that we would like to follow up on in the future. We may also want to contact you in the future to see if you would be interested in enrolling your child in another study. Please indicate on the consent form if you are willing to be contacted about any future research studies. You have the right to withdraw your consent for future contact at any point in the future.

When your child turns 18, he/she has the right to decide whether to be in research and has the right to withdraw from this study. We will not get back in touch when your child turns 18, unless you or your child seeks to do so. However, if your child reaches the age of 18, during the course of the study, we will contact you and your child, and offer your child consent. If your child does not have the capacity to consent, we will offer you to re-consent. If we cannot reach you, or your child, or you or your child chooses not to consent, we will break all links to their identity.

## **Questions:**

If you have any questions about this study please ask the research nurse. You can also contact **the Project Manager [emma.eastman@uct.ac.za](mailto:emma.eastman@uct.ac.za) and Primary Investigator [Kirsty.donald@uct.ac.za](mailto:Kirsty.donald@uct.ac.za); 081 810 2614** to have your questions answered. If you wish to withdraw your consent at a later date you may also contact the project manager. For questions about your rights as a research participant, please contact a representative of the **University of Cape Town Human Research Ethics Committee on 021 406 6338**. This can include questions about your participation, complaints or concerns about the study, including if you feel pressure to enroll. You will be given a copy of this information sheet and signature page for your records. Please keep a copy of these documents in case you want to read them again.

## Consent Form: Parent/Guardian of Control

I, \_\_\_\_\_, the legal guardian of \_\_\_\_\_ agree to allow my child to take part in this study, which will use his/her DNA to try to learn about developmental delays and other social and thought problems.

- I give permission to be interviewed and for my answers to be recorded.
- I give permission for a sample of my child's blood and saliva to be taken.
- I give permission for my child's sample to be stored at the Human Genetics Lab at the University of Cape Town, at the National Institute of Mental Health Repository and Genomics Resource in the United States and at the Broad Institute in the United States.
- I agree to my child's DNA being made available on both open and controlled access databases.
- I understand that:
  - a. My child's identity will be protected.
  - b. I will be compensated 250 Rand for the time and inconvenience of being part of the study.

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Additionally, I am choosing to give permission for the following:

1. Do we have your permission to create cell lines and stem cells from the blood we draw?
  - Yes
  - No
2. Do we have your permission to photograph your child?
  - Yes, but the photo can only be shared outside of the study with health care providers or researchers for clinical, educational or research purposes (for example with the Face2Gene application)
  - No
3. Do we have your permission to contact you in the future to follow-up on this study or for future studies?
  - Yes
  - No

Address: \_\_\_\_\_

Telephone/Cell: \_\_\_\_\_

Email/Other: \_\_\_\_\_

4. If you agree to be contacted again, and we cannot find you, may we try to reach you through a friend or relative?

- Yes
- No

Contact information of a relative/friend (1):

Name: \_\_\_\_\_ Relationship: \_\_\_\_\_

Telephone/Cell: \_\_\_\_\_

Email/Other: \_\_\_\_\_

Contact information of a relative/friend (2):

Name: \_\_\_\_\_ Relationship: \_\_\_\_\_

Telephone/Cell: \_\_\_\_\_

Email/Other: \_\_\_\_\_

I have read, or have been read, the accompanying information sheet in my own language. I understand this consent form and the information sheet. Any questions I had have been answered. I understand that I may withdraw my consent from any, or all, aspects of this study without any impact on my medical care, and without any repercussions.

Signed in \_\_\_\_\_ (place) on \_\_\_\_\_ (day) of \_\_\_\_\_ (month) \_\_\_\_\_ (year)

Signature \_\_\_\_\_

Witness 1 Name \_\_\_\_\_ Signature \_\_\_\_\_