SickKids
THE HOSPITAL FOR SICK CHILDREN
Research Ethics Board

Participant name:

DOB:

HSC #:

Research Ethics Board
Research Consent Form for those consenting for themselves
Genetic diagnosis associated with an increased risk for autism

Title of Research Project:
Molecular and Genomic Analysis of Autism Spectrum and Associated Neurodevelopmental Disorders

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Purpose of the Research:
We hope to use this study to help us find genes that may predispose to, or cause autism spectrum and/or associated neurodevelopmental disorders. Autism spectrum disorders (ASDs) are a group of neurodevelopmental disorders that are characterized by impaired socialization and communication in association with atypical, repetitive behaviors. The spectrum of ASDs is quite variable with a wide range of symptoms and severity. Current estimates suggest that the prevalence of autism is about 1/110

children. This figure represents children with autism as a primary diagnosis. There are also children with specific genetic diagnoses, such as Fragile X syndrome, Rett syndrome and CHARGE syndrome, who are at increased risk for developing autism. Genetic and epigenetic studies of the genes involved in these conditions are likely to further our understanding of the basis of autism.

When we refer to a finding or problem being genetic, that usually means that there has been a change (or mutation) in a gene. Genes are sections of DNA which provide the instructions for how our bodies grow, develop and function. A genetic mutation changes the sequence of the DNA and this can cause an error in the instructions(s) from that specific gene. Epigenetic alterations are different because they do not involve changes in the DNA itself. Rather, the change may surround a section of DNA affecting access to the genes in that section. This can prevent a gene from providing a specific instruction that the body needs.

We all have a number of epigenetic variations and some of these are believed to lead to the normal differences we see between individuals. However, some epigenetic changes may lead to problems with growth and development (such as ASD). We are trying to find out if individuals with specific genetic diagnoses, associated with an increased risk of autism, may have an increased number of epigenetic changes. If we find an increased number of epigenetic changes, this will help us to focus further research on understanding autism.

Description of the Research:

We are asking you to participate in our research on autism, even though you don't have a diagnosis of autism. If you agree to participate, a small blood sample (20-30cc or 1-2 tablespoons) will be taken from you by a trained health care professional. This blood will be used to look at the pattern of epigenetic changes in your DNA. We are also asking parents of children who participate in this study to provide us with saliva (spit) samples if possible. We will extract DNA from the saliva samples. In the case where a child is identified to have an atypical epigenetic pattern, it will be very helpful to compare their epigenetic patterns to that of their parents.

Incidental findings: It is important that you understand that although we will be examining your entire genome, we will not be reviewing all of this information in detail. However, **there is a chance we may uncover health information which would directly impact your care**. If the study identifies such information, **the study clinician or genetic counsellor involved in your care will contact you to discuss the finding**. Should this happen, repeat testing in a clinical laboratory might be recommended to confirm any research results and the benefits and risks as well as possible inconveniences will be discussed with you. Please indicate below if you want to be informed of any incidental findings.

☐ Yes, I would like to be informed of any incidental findings as described above.

☐ No, I do not want to be informed of any incidental findings as described above.

Potential Harms:
There may be a small amount of bleeding when blood is taken from a vein and there may be slight discomfort and bruising or redness that will usually disappear in a few days. EMLA patches, similar to bandaids which contain a cream that will to numb the puncture area will be made available upon request.

Potential Discomforts or Inconvenience:
During the course of the study, if we identify any information that may have clinical significance to you, one of the investigators will contact you about these observations and arrangements will be made for counselling and assistance for you in understanding the personal and family significance should you need it. This knowledge could cause psychological stress to you and your family. In rare cases, knowing about a presence of a genetic problem might possibly affect your health and/or life insurance coverage in the future.

The interpretation of the genetic information will depend in part on the family information that you have provided. If the results of genetic tests do not fit with the information that you have given about your family, it may be that the test is faulty, or that the family information that you gave is wrong. For example, this might happen if the parents do not mention that their child was adopted, or that the biological father is different from the apparent father (this is known as non-paternity).

Potential Benefits:

To individual subjects:
You and your family may or may not benefit from participating in this study. It is possible that we will be able to identify the molecular or genomic cause of the autism spectrum and/or other neurodevelopmental disorder in your family which may help with treatment options. While we cannot know this for sure, we do know that the participation of your family will allow researchers to gain insight into the causes of autism spectrum and/or associated neurodevelopmental disorders. In addition, any incidental findings which are identified in you will be communicated to you if you choose. This could uncover health information which would directly impact your health care.

There will be an annual group information session for parents of children with ASD and/or associated neurodevelopmental disorders, who are enrolled in this study, where any general findings of interest will be shared. A summary of the meeting will be posted on our website (http://www.tcag.ca/researchProjects.html).

To society:
Although you may not benefit directly from this study, results from the study will improve the understanding of autism/ASD and may benefit patients and parents in the future.

Confidentiality:

We will respect your child's privacy. No information about who you are will be given to anyone or be published without your permission, unless required by law. For example, the law could make us give information about you if a child has been abused, if you have an illness that could spread to others, if you or someone else talks about violence (killing themselves or others) or if the court orders us to give them the study papers.

SickKids Clinical Research Monitors, employees of Genome Canada, Autism Speaks, Networks of Centres of Excellence of Canada, Canadian Institutes of Health Research, and the Ontario Ministry of Research and Innovation may see your child's research study records to check on the study. By signing this consent form, you agree to let these people look at your s records. We will put a copy of this research consent form in your patient health record.

The data produced from this study will be stored in a secure, locked location. Only members of the research team (and rarely those individuals described above) will have access to the data. This could include external research team members. Following completion of the research study the data will be kept as long as required then destroyed as required by Sick Kids policy. Published study results will not reveal your identity.

As well, the genetic/epigenetic data collected from you will be kept strictly confidential. Confidentiality will be maintained at all times by assigning number codes rather than names to genetic material (blood DNA sample). The codes will be kept in locked files and available only to Dr. Scherer or those working with him. No information that reveals your identity will be released or published without your consent. In addition, information regarding the results of this research may become part of your s health record if relevant to your medical care.

The DNA isolated from your blood will be stored indefinitely with the number codes, so that as new genes are discovered, which are involved in autism, this DNA can be used to continue the research.

Reimbursement:

We will pay for your parking expense or public transit expenses for visiting SickKids for research assessments and blood collection. If you stop taking part in the study, we will pay these expenses for taking part in the study so far.

Participation:

If you choose to take part in this study you can withdraw from the study at any time. The care you get at SickKids will not be affected in any way by whether you take part in this study. If you choose at anytime to withdraw your genetic material (blood DNA sample) from participation in the study, you will have the option of having the identifying labels removed and the DNA material left for research or having the material destroyed.

During this study we may create new tests, new medicines, or other things that may have monetary value. Although the program may raise money from these findings, we cannot give you any of this money now or in the future.

Your signing this consent form does not interfere with your legal rights in any way. The staff of the study, any people who gave money for the study, or the hospital are still responsible, legally and professionally, for what they do.

Sponsorship:
This study is directed by Dr. Steve Scherer and the Hospital for Sick Children. The current funders of this research are Genome Canada, Autism Speaks, Networks of Centres of Excellence of Canada, Canadian Institutes of Health Research, and the Ontario Ministry of Research and Innovation.

Conflict of Interest:
Dr. Steve Scherer and the research team members have no obvious conflict of interest to declare.

Consent:

By signing this form, I agree that:

1. You have explained this study to me. You have answered all my questions.
2. You have explained the possible harms and benefits (if any) of this study.
3. I understand that I have the right to refuse take part in the study and the right to stop at any time. My decision about taking part in the study will not affect my health care at SickKids.
4. I am free now, and in the future, to ask questions about the study.
5. I understand that no information about who I am will be given to anyone or be published without first asking my permission.
6. I will provide a blood sample and both parents (when available) will provide saliva samples.
7. Such samples can be used as a source of DNA, and stored indefinitely, for research into the genetics of autism/ASD and/or associated neurodevelopmental disorders
   I have read and understood pages 1 to6of this consent form. I agree, or consent, to take part in this study.

_____	_____
Printed Name of Subject & Age	Subject's signature & date
_____	_____
Printed Name of person who explained consent	Signature & date

Printed Witness' name (if the subject/legal guardian does not read English)	Witness' signature & date

If you have any questions about this study, please call:
Dr. Rosanna Weksberg at 416-813-6386

If you have questions about your rights as a subject in a study or injuries during a study, please call the Research Ethics Manager at (416) 813-5718.