



THE HOSPITAL FOR  
SICK CHILDREN

Research Ethics Board

Participant name:

DOB:

HSC #:

**Research Ethics Board  
Research Consent Form**

**Title of Research Project:**

Molecular Genetic Analysis of Autism, Pervasive Developmental Disorder and Severe Speech and Language Disorder

**Investigator(s):**

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**Purpose of the Research**

The research to be described is motivated largely by the recent observation that a subset of children with Moebius Syndrome have behavioural, cognitive, language, and/or social difficulties. Some of these difficulties are also seen in children with autism spectrum disorder (ASD).

We hope to use this study to help us find genes that may predispose or cause autism, Autism Spectrum Disorder (ASD), Moebius syndrome, or severe speech and language disorder (SSLD). At present very little is known about the causes of these disorders, although genetic factors are thought to be important. By finding the gene or genes involved, we hope to improve our knowledge of autism and Moebius syndrome, which, in turn, may help in diagnosis as well as the design of new treatments.

**Description of the Research**

1) Your medical records from the Hospital for Sick Children, as well as any additional institutions will be requested for review. Clinical information on you will include a detailed clinical interview and a standardized observation session (involving the observation of a play sequence, providing information about social situations, verbal and gestural communication, and repetitive behaviours), and an evaluation of your cognitive skills and/or adaptive

functioning. This will take between 2-3 hours with you, and an additional few hours with one of your parents. This may be during the same visit or on a separate occasion. Many of these measures may have already been done as part of a previous clinical assessment.

2) Genetic studies: A small blood sample (20-30cc or 1-2 tablespoons) will be taken from you by a trained person to look for genetic factors (DNA) which may give us clues to the genetic links, or the cause to your developmental problems.

New information from this study or other studies may affect whether you want to continue to take part in the study. If this happens, we will tell you about this new information.

**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 to numb the puncture area will be made available upon request.

**Potential Discomfort**

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 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 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 father is different from the apparent father (this is known as non-paternity).

**Potential Benefit**

**To individual subjects:**

You may not benefit directly from participating in this study.

**To society:**

Although you may not benefit directly from this study, results from the study will improve the understanding of autism/ASD/Moebius syndrome/SSLD and may benefit patients in the future. There will be an annual information session for parents where any general findings of interest will be shared.

**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 the law makes us do this. 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 suicide (killing themselves), or
- If the court orders us to give them the study papers

SickKids Clinical Research Office Monitor, employees of the company funding the Molecular Genetic Analysis of Autism, Pervasive Developmental Disorder and Severe Speech and Language Disorder, or the regulator of the study may see your health record to check on the study.

By signing this consent form, you agree to let these people look at your records. We will put a copy of this research consent form in your patient health records.

The data produced from this study will be stored in a secure, locked location. Only members of the research team (and maybe those individuals described above) will have access to the data. Following completion of the research study the data will be kept as long as required by the SickKids "Records Retention and Destruction" policy. The data will be destroyed according to this same policy.

The results of the tests we describe in this form will be used only for this study. If another doctor caring for your needs to see these results, you will have to give us your permission. We will ask you to sign a form saying that you agree that this person can see your results. We recommend that only a registered psychologist or doctor tell you what the results of these tests mean.

The genetic 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 health record.

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, Dr. Scherer can use this DNA to continue research.

**Reimbursement:**

We will pay for your parking expense for visiting SickKids for assessments. If you stop taking part in the study, we will pay you for your parking expense for taking part in the study so far.

**Participation:**

It is your choice to take part in this study. You can stop 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.

New information that we get while we are doing this study may affect your decision to take part in this study. If this happens, we will tell you about this new information. And we will ask you again if you still want to be in the study.

During this study we may create new tests, new medicines, or other things that may be worth some money. Although we may make money from these findings, we cannot give your child any of this money now or in the future because your child took part in this study.

In some situations, the study doctor for the study may decide to stop the study. This could happen even if the service provided by the study is helping you. If this happens, the study doctor will talk to you about what will happen next.

We will give you a copy of this consent form for your records.

If you become ill or are harmed because you took part in this study, we will treat you for free. 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:**

The funder of this research is the Canadian Institutes of Health Research and Genome Canada.

**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 know what I could do instead of taking part in this study. I understand that I have the right not to 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 have been told that my medical records will be kept private. You will give no one information about me, unless the law requires you to.
6. I understand that no information about who I am will be given to anyone or be published without first asking my permission.
7. I will provide consultation of my medical records for any information related to autism/ASD/Moebius syndrome/SSLD;
8. I will provide a blood sample.
9. Such a sample can be used as a source of DNA, and stored indefinitely, for research into the genetics of autism/ASD/Moebius syndrome/SSLD.
10. My DNA can be used in this laboratory or sent to other laboratories for research into autism/ASD/Moebius syndrome/SSLD after all identifying information has been removed.

11. My DNA can be used in coded form in research into the Genetics of Autism and Moebius syndrome.
12. That you will attempt to contact me in the future if any information about the genetics of autism that is specifically relevant to me comes to light if I inform you of any change of address.
13. I have read and understood pages 1 to 5 of this consent form. I agree, or consent, to take part in this study.

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Printed Name of Subject & Age

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Subject's signature & date

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Printed Name of person who explained consent

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Signature & date

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

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Witness' signature & date

If you have any questions about this study, please call **Dr. Wendy Roberts at (416)813-8748**. 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.