

PhenomeCentral Assent Form

Title of Research Project:

PhenomeCentral: A portal for phenotypic and genotypic matchmaking for patients with rare genetic disorders

Investigator(s):

Michael Brudno, PhD
Senior Scientist, Genetics & Genome Biology,
Director, Centre for Computational Medicine,
Hospital for Sick Children Research Institute
Associate Professor, Department of Computer Science, University of Toronto
416-813-7654
ext. 308179

Kym Boycott, MD
Senior Scientist, CHEO Research Institute
Clinical Geneticist, Department of Genetics, CHEO
Associate Professor Department of Pediatrics, Faculty of Medicine, University
of Ottawa
613-737-7600
ext.4139

Research Contact:

Brittney Johnstone, MSc, CGC, CCGC
Genetic Counsellor, The Centre for Computational Medicine,
The Hospital for Sick Children
416-813-7654
Ext. 414028

Why are we doing this study?

You are being invited to be part of a research study because you have an undiagnosed genetic illness. This means that your doctors do not know exactly what is causing you to be sick but they know that there is a problem with your genes. Genes are parts of your cells which contain the instructions for making a person. We inherit our genes from our parents. In some people, there is a problem with how the genes work and they become sick.

As part of this study we would like to make a database where information about rare or undiagnosed genetic conditions will be kept. The database is called PhenomeCentral. It will be housed at SickKids.

Doctors and researchers (people who study things like genetic conditions) can use this stored information to find people with the same or similar features and help their doctors find out what is causing them to be sick. By sharing information they may be able to better understand rare genetic conditions and provide better care.

Doctors and researchers who want to use the information in PhenomeCentral can only do so with special permission from the people who take care of the database at SickKids.

PhenomeCentral is part of a group of databases called the Matchmaker Exchange that work together to find matches between them. With more databases involved, the chance to find a match increases. You can decide if you would like your information to be shared within PhenomeCentral only or more broadly with the Matchmaker Exchange.

What will happen during this study?

If you agree to be part of this study, the study team members will collect information about you and your illness from your medical records at SickKids. You can pick which parts of your records will be included. This will only be done with your consent and may include any of the following information based on your choice:

Information about your genetic condition:

1. Your symptoms (how you feel)
2. Your medical history including visits done at SickKids
3. Results found by your doctor when he/she examines you
4. Results of any lab tests done by your doctor including blood tests, x-rays and other special tests like tests done on your genes.

Information about your family:

Because genes are passed on from parents, we will also collect any information about your family's medical history. This may help information will be used to help researchers better understand what is making you sick and whether it is passed on in the family.

Pictures

Pictures can sometimes help doctors in finding out about health conditions. For rare conditions such as yours, sharing pictures with other study doctors may help your doctor find other children with the same condition you have. This may help your doctor find better ways to treat you. We want to find out if there are other people in the world who have special features that look like yours because this may help us better understand your condition. This may include pictures of parts of your body or your face. Some of these pictures can be so unique that anyone seeing them may know it is you.

As part of this study, the study team may want to take pictures of you especially if you do not have pictures in your medical records. The study doctors may also want to take additional pictures. This is not required and it is up to you to make a choice. You will be asked to give special consent for this and your decision about let your pictures be stored in PhenomeCentral will not affect your participation in this study.

The pictures may be of a part of your body, like your hand or it could be your face. You can tell your doctor if there is a part of your body that you do not want to have photographed.

What happens to my information?

You will be given a special code number which we will use to identify all your information and your pictures. Your name and any information which can identify you will be kept at SickKids only.

All the other information and pictures we collect about you will be kept safely at PhenomeCentral and will not be destroyed unless you tell us you do not want to be in the study anymore.

If a doctor or researcher uses your information and pictures, they may find other people with the same or similar condition that you have and they may want to contact your parent/guardian for more information. If we find a

diagnosis (what causes your condition), we may want to contact you and your parents to let you know. You and your parents can decide whether you can be contacted for additional information.

Even if you consent now, you can change your mind later. All you need to do is tell your parent/guardian and they will let the study team know. It is important to know that if you change your mind, you can ask to have your stored information and pictures destroyed. However, we can only destroy the information and pictures stored inside PhenomeCentral. Any information or pictures that have been shared with other study doctors cannot be taken back.

Are there good things and bad things about the study?

The good thing about being part of this study is that your doctor might be able to find another person with special features that look like yours. This may help your doctor to learn more about your health condition and find better ways to take care of you.

Your doctor may share your information and pictures with other doctors and scientists on the website. It may be possible for the other doctors and scientists to recognize you or find out who you are because they saw your photo. We have special measures to make sure your privacy is protected and that nobody can look at your information without permission.

Can I decide if I want to be in the study?

Nobody will be angry or upset if you do not want to be in the study. If you agree to be part of this study, you can choose what you want to be shared in the database. If you agree now, you can change your mind at any time. Just let your parent/guardian know and they will tell the study doctor.

When you grow up and can make decisions on your own, you can contact the study team. We will go over the study with you and you can make your own choices, including whether you still want to be in the study or not.

Consent for Information to be included in the study:

You can choose which information you want included in the database (read list to child and check)

Yes No

- Year and month of birth: we need this so we can calculate your age
- Symptoms and medical history
- Measurements such as height, weight, head circumference, etc.
- Family history
- Results from any available tests including genetic tests
- Diagnosis (if one is discovered in the future)

_____ Parent's Initials _____ Date

Permission to share information within the Matchmaker Exchange

You can choose if you want to share your information with PhenomeCentral only or also with the Matchmaker Exchange.

- My information can be shared with the Matchmaker Exchange.

_____ Initials _____ Date

I do not want my information to be shared with the Matchmaker Exchange.

_____ Initials _____ Date

Consent for Pictures to Be Included:

You can choose which information you want included in the database (read list to child and check choices). Before the pictures are shared, you can look at them and let the study team know if you want them to be included in the database.

Including Pictures Already in My Medical Records

- I do not want any of my pictures included
- All my pictures in my medical record can be included
- Pictures of my face should not be included
- Pictures of my face can be included

Taking New Pictures:

- I do not want any new pictures to be taken of me
- The study team can take pictures of me
- I do not want any new pictures of my face
- The study team can take pictures of my face

_____ Parent's Initials _____ Date

Assent:

I was present when _____ read this form and said that he or she agreed, or assented, to take part in this study.

Printed Name of person who obtained assent

Signature & Date