SickKids®
THE HOSPITAL FOR SICK CHILDREN
Research Ethics Board

Protocol Title: Personal Genome Project Canada
Full Consent Form

Principal Investigator:
Stephen Scherer, PhD, FRSC
The Hospital for Sick Children
(416) 813-7613
stephen.scherer@sickkids.ca

Co-Investigators & Staff

Michael Brudno, PhD
The Hospital for Sick Children, University of Toronto
(416) 978-2589
brudno@cs.toronto.edu

George Church, PhD
Harvard Medical School
(617) 432-7562
gmc@harvard.edu

Jill Davies, MS, CGC
MedCan Clinic
416-350-5908
JillDavies@medcan.com

Peter Ray, PhD
The Hospital for Sick Children
(416) 813-6497
peter.ray@sickkids.ca

Norman Rosenblum, MD
The Hospital for Sick Children
(416) 813-5667
norman.rosenblum@sickkids.ca

Cheryl Shuman, MS, CGC
The Hospital for Sick Children
(416) 813-7550
cheryl.shuman@sickkids.ca

Michael Szego, PhD, MHSc
Hospital for Sick Children
(416) 894-4347
michael.szego@utoronto.ca

Purpose of the Research:

The Personal Genome Project Canada (also the "PGP" or the "study") is an independent public genomics research project and a joint venture between the Hospital for Sick Children and the University of Toronto McLaughlin Centre for Molecular Medicine. The main scientific goal of this study is to explore ways to connect human genetic information with human trait information (i.e., human DNA sequence, medical information, tissue samples and physical traits) in a public fashion so that such data may be used for hypothesis-generating research and other scientific, clinical and commercial development efforts worldwide. Additional goals include (i) developing a fully consented and public dataset to aid in the development of computational tools and user interfaces for scientists, clinicians and individuals; and (ii) the education of participants and the general public about the potential benefits, risks, and uncertainties posed by the widespread availability of genetic and related information. Upon successful registration and full enrolment in the PGP, your genetic and trait information will be made available on a publicly accessible website and database.

Description of the Research:

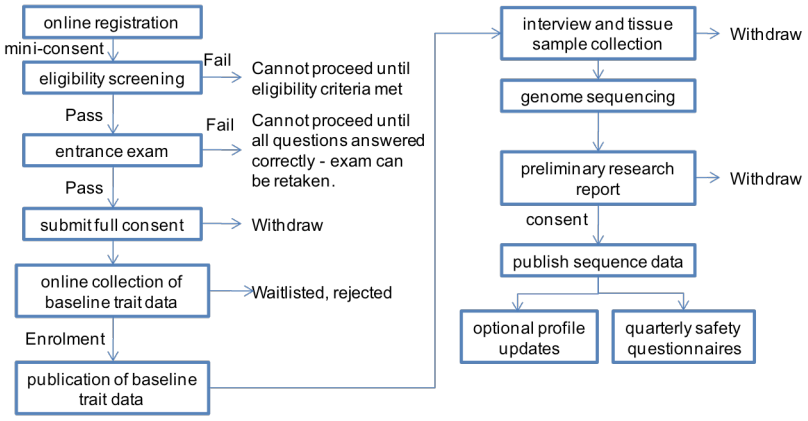
I. Overview

The PGP will collect tissue samples and personal and trait information from each participant. If you choose to participate in the study, your genetic and trait information will be made available through a publicly accessible website and database, according to the procedures described below. You may be selected for whole genome sequencing in which case you will receive certain research data from the PGP (further described below). This data or other information that you receive due to your participation in the study, including DNA sequence data, is not intended to replace in any way professional medical advice, diagnosis or treatment. You may not use any information you receive from the PGP for any medical or clinical purpose unless the relevant DNA sequence or other data, including any interpretations or findings presented in your Preliminary Research Report (described below) are first confirmed by a licensed healthcare professional.

PGP-Canada is an autonomous research project; however, PGP-Canada does have an informal relationship with PGP-USA, a Harvard University-based research project. No confidential research participant information will be shared with PGP-USA. The collective goal of PGP-Canada and PGP-USA is to enrol a total of 100,000 participants in these studies. PGP-Canada will begin with a modest goal of 35 completed genome sequences a year; the pace at which we expand the study to large numbers of enrolees is unknown.

Participation in this study is voluntary. You do not have to participate in the PGP. If you decide to participate you may withdraw from this study at any time, as more fully described in this consent form.

Overview of PGP-Canada



Description of different steps in the PGP-Canada. This multi-step process is designed to ensure that you are as fully informed about the project as possible prior to data publication. Decision points where you will be asked whether you want to continue participating or withdraw from the study have been indicated as "Withdraw".

II. Eligibility Screening & Registration

The initial phase of your participation in the PGP includes registration at the PGP-Canada website, where you are asked to submit your name, email address, date of birth and privacy preferences. Prior to sending us your personal information you are required to read and sign the "mini-consent" form, which more fully describes the Eligibility Screening procedures, and potential risks and benefits.

You are also required to study materials on the PGP website, including this full consent form and to complete an online exam to assess your comprehension of concepts relevant to providing informed consent to participate in the PGP. After completion of the eligibility questionnaire and online exam, you will be informed whether your request for registration was accepted or whether you are ineligible to participate. Accepted individuals can then continue with pre-enrolment.

III. Pre-enrolment

Individuals who are registered in the PGP will then be required to complete this full consent form and submit it to the PGP. Only participants who were informed that they passed eligibility screening should complete and submit this consent form. The next step involves the collection of some personal information about yourself, called baseline trait data.

(a) Baseline Trait Data

Baseline trait data includes date of birth (month and year only), medications, allergies, vaccines, personal medical history, race/ethnicity/ancestry, and vital signs (e.g. height, weight, blood pressure). Your baseline trait data, along with any other information that you voluntarily submit to the PGP, will be made available on the PGP's public website upon enrolment in the study as

described in this consent form. Baseline trait data must be supplied by you to be considered for enrolment in PGP-Canada.

(b) How Baseline Trait Data is Collected

You will be asked to self-report and submit your baseline trait information to the PGP through an online questionnaire form. Collecting and submitting the Baseline Trait Data will take an estimated 1-3 hours, and that time may be lost if you are not selected for enrolment. You will not be compensated for any lost time.

Your baseline trait information will not be immediately published. Once you upload your self-reported trait information to the PGP Canada website you will be asked to complete the pre-enrolment and enrolment procedures outlined below. Candidates that are successfully enrolled will then be invited make their baseline trait information public. Research participants can choose not to link their data and withdraw from the study.

(c) Additional optional information constitues Baseline Trait Data

Facial photographs will be requested but are optional for full enrolment in the PGP. Should you opt to have your photograph taken for this research study, you will be asked to sign a separate Photography Consent Form as well. Additional optional personal data such as existing DNA sequencing, genotyping data and trait information such as lifestyle traits may also be requested by the PGP. Any such information that you submit is optional and will constitute part of your baseline trait data, which is publicly available.

(d) Conversations with Family Members

Consent of family members is not required unless you have an identical twin (see below); however, publishing your personal health information and genome sequence may have consequences for your family. Therefore, we strongly encourage you to discuss your decision to take part in the PGP with your family. In particular, we urge you to discuss your decision to participate in the PGP with family that is most genetically related to you (i.e., parents, siblings and children) since they may be most affected by your decision.

Have you discussed your desire to enrol in PGP Canada with your parents, siblings or children, if applicable [ ] Yes [ ] No [ ] Not yet. If you answered no, can you briefly explain why:

_____

_____

(e) *Identical (Monozygotic) Twin.*

If you have any living siblings who are your identical (monozygotic) twin, such sibling(s) will also need to provide consent for your participation in this research study before the PGP will consider you for enrolment. We require both identical twins consent since they usually share the same DNA sequence.

Do you have a living identical (monozygotic) twin? [ ] Yes [ ] No [ ] Unsure

If you have an identical twin, have they enrolled in the PGP or are they in the process of enrolling? [ ] Yes [ ] No [ ] Unsure

IV Enrolment

(a) Application for Enrolment

Upon completion of all pre-enrolment activities (i.e., submitting your baseline trait information and signing this consent form) your application will be considered by the PGP Executive Committee. You will then be notified that you are either: (1) enrolled, (2) requested to provide additional information to complete your enrolment application, (3) waitlisted, or (4) ineligible.

(b) If You Are Not Enrolled

If you are ineligible for enrolment, the PGP will permanently delete any personal information about you, including any collected Baseline Trait Data that you submitted as part of your enrolment application within 2 weeks from the date the PGP informs you of your ineligibility for enrolment.

(c) Publication of Baseline Trait Data

Once you are enrolled in the PGP, you will be given the opportunity to publish your Baseline Trait Data to the PGP's public website and database. As a participant in the PGP you have the option to publish all your baseline data or none of your baseline data; you will **not** have the option of selecting portions of your Baseline Trait Data to publish or not to publish. If you choose to publish your Baseline Trait Data, they will be made available to the public through the PGP's website and database, and combined with other data provided by the PGP or by you. Risks associated with the publication of your Baseline Trait Data are described in the "Potential Harms" Section of this consent form.

(d) No Obligation

As a participant in the PGP you are under no obligation to publish your Baseline Trait Data. However, you will not be able to participate in other aspects of the study, including tissue and DNA analysis, until you publish your Baseline Trait Data. You are free to withdraw from the PGP at any time. If you withdraw from the study, all your data will deleted within two weeks of your notice to us. After publication of your Baseline Trait Data you are still free to withdraw; however, we cannot control your information once it has been downloaded by a third party from the public website.

(e) Next steps

After your Baseline Trait Data is published you will be placed in a queue for whole genome sequencing. Our current target is to sequence approximately 35 individuals per year. Successful enrolment is not a guarantee that your genome will be sequenced. Due to the current high costs of whole genome sequencing, individuals who agree to pay for their own genome sequence will be preferentially selected from the queue. Individuals who choose not to subsidize their own DNA sequencing may still be selected for sequencing based on the following criteria:

educational purposes (e.g. medical students, genetic counsellors whose whole genome sequence could be used as a high impact teaching tool to teach genomics to medical/genetic counselling students), or under-representation of a group among the PGP participants (e.g. to balance the number of males and females, or members of ethnic minorities). The goal is to have participants accurately representing Canadian diversity. When you are next in the queue, you will be contacted by phone and/or email and asked to make an appointment at MedCan, a private medical clinic in downtown Toronto for an in-person interview and tissue collection**.**

V. In-Person Interview and Tissue Collection at MedCan

(a) In-Person Interview

During your visit to the medical centre for you will meet with a member of the PGP staff to confirm that you are familiar with the study protocols, including this consent form, and to review and confirm your identity and the accuracy of your Baseline Trait Data. You will also be asked to provide government issued identification so we can confirm your identity. You will also be given the opportunity to ask any questions you may have. The interview will take approximately 1 hour.

(b) Blood Sample Collection at the Medical Centre

The PGP will collect a blood sample during your visit to the medical center. Approximately 10 ml of blood (approximately 1.5 tablespoons) will be collected by a trained medical professional appointed by the PGP. The blood sample will be used as a source of your DNA to generate sequence data. We will also save and freeze a small blood sample at the PGP biorepository at The Hospital for Sick Children, which we may use to make a blood cell line at a later date. Cell lines provide a renewable supply of your blood cells and DNA. We will re-contact you (if you consent to recontact, see below) and require your consent prior to creating a blood cell line. All the potential risks and benefits of creating a blood cell line will be outlined if you are recontacted for the purposes of consent to create a blood cell line. Your blood sample will be destroyed after a maximum of 15 years in storage.

(c) No Obligation

As a participant in the PGP you are under no obligation to submit any tissue samples to the study as part of your visit to a medical center or otherwise. However, you may not be able to participate in other aspects of the study, including tissue, cell line and DNA analysis, until you have submitted a blood sample

VI. Generation of DNA Sequence Data

(a) How Analysis is Performed

Genome-sequencing approaches to be used will be state-of-the-art; however, they have been introduced to the laboratory at such a fast rate that the development of computer methods to analyze the data they generate has lagged behind. Computer scientists will be using your published genome sequences (i.e. if you consent to make them public) to develop algorithms to

better analyze genome-wide sequence data, improve data visualization, and improve medical annotation to prioritize genetic variants based on their potential significance. Thus, a variety of analyses will be performed on your sequence data, which are not possible to predict at this stage.

(b) No Guarantees

The PGP cannot make any guarantees about the accuracy or completeness of any such analysis or research or the processing time for any of these activities.

(c) Receipt of Your DNA Sequence Data

Once the PGP has completed DNA analysis of your sample(s), the PGP will make your DNA sequence data available to you via a password protected area on the PGP website. Your sequence data will remain confidential until you consent to publish it (discussed further below). This information is for research purposes only. You may not use this data for any medical or clinical purpose unless the DNA sequence or other data, including any interpretations or findings presented in your Preliminary Research Report (described below), are first confirmed by a licensed healthcare professional. Genome sequencing is not an all-encompassing genetic screen for every possible condition. You can find examples of DNA sequence data similar to what you will receive as an enrolee on the study website.

(d) Receipt and Review of Your Preliminary Research Report

In addition to your DNA sequence data, the PGP will provide you with a preliminary research report (the "Preliminary Research Report" or the "Report") intended to help you make a more informed decision about whether or not to publish your DNA sequence data to the PGP's public website and database. This Report will contain a non-comprehensive list of genetic variants present in your DNA sequence data, as well as any additional information, resources or interpretation that the PGP may decide to provide to you as part of your Report. In preparing your Report, the PGP may review your DNA sequence data in combination with the trait data and other information that you have submitted to the PGP.

The Preliminary Research Report includes preliminary research findings only. The PGP cannot guarantee that the Report is either accurate or complete. The databases, knowledge and tools used to generate the Report are not comprehensive and may change from time to time. Only one Preliminary Research Report will be provided to you. The PGP will not update or supplement the Report.

The Preliminary Research Report is not intended to substitute in any way for professional medical advice, diagnosis or treatment. You may not use the Report for any medical or clinical purpose unless the relevant sequence or other data, including any interpretations or findings presented in your Preliminary Research Report, are first confirmed by a licensed healthcare professional. You may book an appointment with the PGP genetic counsellor at no cost if you have health-related questions arising from your Preliminary Research Report or sequence data.

Examples of other Reports similar to what you may receive as an enrolee are available on the study website.

(e) Decision to Publish DNA Sequence Data

After you receive your DNA sequence data and Preliminary Research Report, you will be asked whether you want to (i) make your DNA sequence data available on the PGP's public website and database, or (ii) withdraw from the PGP. If you choose to publish your DNA sequence data, the PGP will make this data available on its public website and database and this data will be associated with your Baseline Trait Data along with any other data supplied by you or the PGP. If you choose to withdraw from the PGP, your unpublished sequence and published Baseline Trait Data and any other personal information provided to the PGP will be deleted from the PGP within 2 weeks of your decision.

(f) No Obligation

You are not obligated to publish or to take any other action with respect to your DNA sequence data. However, if you elect not to publish your DNA sequence data you will not be permitted to continue to participate in the study.

(g) Updating Your DNA Sequence Data

If you choose to publish your DNA sequence data, the PGP may re-process and supplement your DNA sequence data from time to time as new data, information or techniques become available. If you consented to the publication of your original DNA sequence data, then the PGP may publish your re-processed and/or supplemented DNA sequence data directly to the PGP's public website and database without your further consent. You will not be provided an additional or revised Preliminary Research Report or other analysis of your DNA sequence data, and you will not be given an opportunity to choose whether or not to publish your re-processed and/or supplemented DNA sequence data.

(h) What signing this consent means

By signing this consent form, you authorize the PGP to do the following:
if after reviewing your Preliminary Research Report you inform the PGP that you will allow the PGP to publish your DNA sequence data and other personal information, the PGP may publish such data and information (including your DNA sequence data, whether or not re-processed and/or supplemented, and your Preliminary Research Report), along with your trait information and any other information you have submitted to the PGP. This means that the PGP may proceed to publish this data and information without you being asked to sign any additional consent forms. The PGP will publish the data and information on a publicly accessible website and database. It may also publish the data and information in other formats and/or media.

VII. Ongoing Participation

(a) Mandatory Safety Questionnaire

Public genomics projects like the PGP-Canada are relatively new and the actual risks of participation are unknown. Accordingly, upon publishing your trait data and DNA sequence, we will ask you to fill out a safety questionnaire every 3 months so we can rapidly identify any unforeseen consequences of participation. The PGP will circulate the safety questionnaire to you

via email and we ask that you return the completed form within a week. The safety questionnaire is composed of five questions:

> (i) What negative and/or positive events have happened to you and/or your relatives or acquaintances due to your participation in the PGP?
>
> (ii) What are the reactions or responses of your relatives and acquaintances to the publication of your genetic, trait and other data?
>
> (iii) Please report incidents of being contacted by acquaintances or by strangers (including researchers, health care providers or members of the media) regarding your data being published online.
>
> (iv) In what ways has this study positively or negatively influenced your interactions with your medical care providers or your receipt of or access to health care services?
>
> (v) Has your involvement in this study triggered the need for any health or medical care that would not otherwise have been done? If you answer "yes," please describe (a) the specific medical intervention or care you received, and (b) the findings or consequences of the medical intervention or care you received with regard to your health. Health or medical care that would have been performed had you not participated in this study, whether due to symptoms, a personal or family medical history, routine screening or any other reason, should not be included.

(b) *Reporting of Unexpected Events*

You are requested to immediately report any unexpected events that you may experience as a participant in this study. Such events should be reported to the PI or study coordinator directly.

(c) Periodic Reflection

At 5 year intervals, and at the end of your participation in the study, you will be requested to write your thoughts about the PGP overall, including whether this consent form adequately described the procedures and risks associated with your participation.

(d) Changes to the Questionnaire

The Safety Questionnaire, including the number of questions and the frequency of circulation to enrolees, may be modified by the PGP from time to time.

II. Recontact

Other than the Safety Questionnaires already described, you are under no obligation to receive study notices or to participate in the study after providing the tissue samples and the information already described. If you choose YES to the question below, you may be contacted by the PGP at a future date and asked if you would like to (i) voluntarily submit additional tissue specimens and/or trait or other information, or (ii) participate in future research studies or other activities coordinated by the PGP. You may change your choice on this option at any time by notifying the PGP in writing.

Willing to be recontacted? [ ] Yes [ ] No

Potential Harms:

The Personal Genome Project is a new form of public genomics research and, as a result, it is impossible to accurately predict all of the possible risks and discomforts that you might experience as a result of your participation in this study. In this section you will read about the risks that we have identified as potentially relevant to your participation in the PGP.

You are strongly encouraged to think carefully about these risks, as well as any other risks or discomforts that you anticipate might arise as a result of your own unique circumstances. These might include your own health or medical conditions, your family and personal relationships or any other factor that is specific to you. In addition to understanding the risks outlined in this section, you should feel confident that you have sufficient knowledge (via the educational and testing materials available on the PGP website) of genetics, human subjects research and the benefits and risks of participation in this study to make an informed decision about whether participation is right for you.

You are strongly encouraged to discuss this study and its potential risks with your immediate family members as well as with your physician and/or other qualified health care providers. You are also encouraged to discuss with the Principal Investigator directly any additional concerns that you may have regarding the risks to you of participating in this study.

Finally, because the science in this area is evolving, and data will be collected on an ongoing basis by the PGP, the risks involved due to your participation in this study, as well as the likelihood and severity of such risks, will change over time. You will not be asked to review and re-sign this consent form every time new information related to the risk of participation becomes available. However, the PGP will try to update this consent form and the study website as frequently as possible to reflect the latest information about the risks of participation. Participants and prospective participants are strongly encouraged to check the website regularly and to update their contact information with the PGP in order to obtain the most current information regarding the risks and discomforts of participation. Please remember that you are free not to participate if you have not already enrolled, and to withdraw at any time if you have already enrolled in the study.

I. Risks Associated with the Publication of Your Data.

(a) *Risks Associated with Public Disclosure*

The risks of public disclosure of your genetic and trait data, including your Baseline Trait Data, any additional trait data or other information you provide, and your DNA sequence data, could affect your employment, insurance and financial well-being or social interactions for you and your immediate family. The following is a non-comprehensive list of hypothetical scenarios that could pose risks for you and/or your family:

(i) Data that you provide (such as facial images, other trait data or DNA sequence data) may be used to identify you, resulting in higher than normal levels of contacts from the press and other members

of the public motivated by positive or negative feelings about the study. This could mean a significant loss of privacy and personal time.

(ii) Data that you provide may cause you to learn – either directly or from a family member or other individual – that you are not related to family members or other individuals in the way that you had previously believed. This could also include inferences or allegations of paternity made by individuals you did not previously know or suspect were related to you.

(iii) Anyone with sufficient knowledge and resources could take your DNA sequence data and/or posted trait information and use that data, with or without changes, to:

(1) infer paternity or other genealogical features of you and/or your family,

(2) claim statistical evidence that could affect the ability of you or your family to obtain or maintain employment, insurance or financial services,

(3) claim relatedness to criminals or incriminate relatives,

(4) make synthetic DNA and plant it at a crime scene, or otherwise use it to falsely identify you, or

(5) reveal to you or a member of your family the possibility of a disease or unknown propensity for a disease.

(iv) Whether or not it is lawful to do so, you could be subject to actual or attempted employment, insurance, financial, or other forms of discrimination or negative treatment due to the public disclosure of your genetic and trait information by the PGP or by a third party.

(v) If you have previously made available or intend to make available genetic information in a confidential setting, for example in another research study, the data that you provide as part of the PGP may be used, on its own or in combination with your previously shared data, to identify you as a participant in otherwise confidential genetic research or trials. This means that any data or other information you may have shared pursuant to a promise of confidentiality or privacy may become public despite your intent that they be kept private and confidential. This could result in certain adverse effects for you, including ones not considered by this consent form.

(vi) Your publicly available DNA sequence data, trait data and other information may also include information that applies to your family members. Some people may draw conclusions from your publicly available information, including speculating about what such information might reveal about you and your family members. As a result, the PGP cannot predict all of the risks, or the severity of the risks, that the public availability of this information may pose to you and your relatives. You are strongly encouraged to discuss this study and its potential risks, including the fact that not all of the risks are known, with your immediate family members.

(b) Reproduction or Modification of your Data

If you choose to make your DNA sequence data and other information available, it will be published on the PGP's publicly accessible website and database and be openly available to third parties. As a result,

neither you nor the PGP will be able to control or restrict the access, use, reproduction, modification, or analysis of your data and other public information. Your data and other public information may be made public in other forms in addition to its inclusion in the PGP database. It may also be changed, without either your or the PGP's consent, in a way that might be inaccurate and or upsetting to you. For example, a third party could access your publicly available sequence data or other information, change it and republish it to suggest that you had a propensity for a disease or other detrimental trait. Additional adverse effects are also possible.

II. Risks Associated with Your Receipt of Data From the PGP.

(a) Data quality not guaranteed

The PGP cannot guarantee that any research data, including your DNA sequence data and your Preliminary Research Report, provided to you by the PGP are all true and correct.

*(b) Data should **not** to be used for medical care*

The data provided to you by the PGP, including your DNA sequence data and your Preliminary Research Report, are not an appropriate substitute for professional medical or clinical advice, diagnosis or treatment, and should not be used by you for any medical or clinical purpose unless the relevant sequence or other data, including any interpretations or findings presented in your Preliminary Research Report, are first confirmed by a licensed healthcare professional.

(c) Risks Due to Pursuing Health or Medical Care based on data obtained through PGP Canada

The clinical importance of most of your personal genome data is not known at this time. In addition, although there is considerable information about the possible connections between genetic information some of these connections, especially when screened for in the general population, remain uncertain. If your Preliminary Research Report contains information about potentially harmful genetic variants, you may experience anxiety or stress. As a result, you may want to seek health or medical care or counseling to verify the accuracy of such interpretations. The PGP Canada has a genetic counselor on staff to answer any medical questions you may have regarding your personal genome information. This service is free of charge to research participants. You may be referred to a physician or sent for additional testing if appropriate. However, no additional health or medical care will be made available to you by the PGP and no special arrangements, for compensation or otherwise, will be made by the PGP should you require or choose to pursue any health or medical care as a result of your participation in the PGP.

(i) The PGP is not responsible for any part of your health or medical care, including, accurately of predicting disease or disease risk, informing you of genetic variations, or providing you with accurate and valid DNA sequence data or interpretations of your DNA sequence data.

(ii) In addition to genetic counseling, you should talk to your doctor or other qualified health care provider if you have questions about any information provided to you by the PGP, including your DNA sequence data or Preliminary Research Report. You should not ignore professional medical advice from your doctor or any other qualified health care provider about any information included or not included in your Preliminary Research Report or in other information provided to you by the PGP. You should not interpret your DNA sequence data or your Report as recommending or

discouraging any specific treatment plan, product, or course of action with respect to your health or medical care.

(iii) If your doctor or other qualified health care provider is directly or indirectly involved with the PGP, as either a researcher or a participant, any health or medical care that you receive from such health care provider, including medical advice or clinical management, represents health or medical care provided by that provider pursuant to your existing doctor-patient relationship, and is not health or medical care provided by the PGP.

Potential Discomforts or Inconvenience:

a) *Physical Discomforts*.
There are no known or foreseeable risks or side effects associated with obtaining saliva, hair, inner cheek swab, skin swab, urine, or fecal samples. The blood draw and skin biopsy may involve a small amount of pain, bleeding and/or fainting, and may also cause temporary bruising and/or infection at the site of puncture. Some degree of permanent scarring can be expected from the optional skin biopsy.

b) Inconvenience.
Pre-enrolment and enrolment including reading online background materials, this consent form, and taking the entrance exam is expected to take 4-5 hours. Uploading your trait information is expected to take 1-2 hours. You will also need to travel to our downtown Toronto clinic for your 1 hour medical interview and blood draw/optional skin punch. Once your sequence is published the quarterly questionnaires should take up to 30 minutes each depending on the level of detail you choose to provide.

Potential Benefits:

I. To Individual subjects:
Participation in the project will likely provide a unique educational experience for participants.

II. To society:
The assessment of associations between human genetic variation, physiology, and disease risk has been an important and growing area of recent scientific research. The Personal Genome Project will contribute to this research by creating a dataset of personal genome sequences that can be evaluated along with the biological, population, medical, and physical data required for association statistics. This pool of combined data may make possible preliminary screening of proposed associations prior to more rigorous or focused data collection. Personal genomics will also supply data for additional areas of scientific research by providing new information on the kinds and levels of variation that exist generally throughout and between individual genomes. It will also provide opportunities to assess non-medical associations that may not have as high priority as medical studies, e.g. biometric associations.

The PGP hopes to increase understanding of human biology, health and disease, which may lead to the development of new diagnostics and therapeutics, improve clinical decision-making, and positively

impact global health. This project could have a signficant impact on personalized medicine and pharmacogenomics.

Confidentiality:

a) Preserving Confidentiality Prior to Publication

During the enrollment process, and before your publication of your Baseline Trait Data or DNA sequence data, as applicable, the PGP will take all reasonable efforts to preserve the privacy and confidentiality of such data, as well as other information you provide to the PGP (your Preliminary Research Report, for example). For example, all data will be stored on a password protected computer (with encryption software) in a locked office. Sick Kids Clinical Research Monitors, employees of the funder or sponsor, or the regulator of the study may see your research record to check on the study. By signing this consent form, you agree to let these people look at your record.

However, you should be aware that public disclosure of such information, even if you have not yet completed enrollment, ultimately determine not to publish such data, or decide to withdraw from the study, may still happen due to unintended data breaches, including hacking or other activities outside of the procedures described in this consent form. For this reason the PGP cannot absolutely guarantee that information you provide to the study, or that is generated about you by the study, will be maintained in a confidential manner.

b) No confidentiality after publication

If you are enrolled in the PGP and choose to publish any of your data to the PGP's public website and database, your data will no longer be kept confidential. The PGP will not require any collaborators or other individuals accessing your information to keep the information in a confidential or anonymous fashion. Unless you withdraw from the study before your data are published, your genetic and trait data will be made available via a publicly accessible website and database. If you withdraw after publication, we will remove all your sequence, trait information and personal information from the PGP; however, we cannot compel third parties who may have downloaded your data from deleting it.

c) Association of your Name with your Data

The PGP will not intentionally associate your name with your genomic or trait data or other information that is published to the PGP's public website and database. The PGP will not intentionally publicly identify you by name as a participant in the PGP without your prior consent. However, as described above, because of the identifiable nature of the information you are providing to the study and generated about you by the study, it is possible that one or more third parties may identify you as a participant in the PGP and associate your published data and information with your name or other information that you have not provided to the PGP and may not have wished to be publicly disclosed.

d) No Direct Disclosure to your Health Care Provider

Your genetic and trait data will not be sent to your health care provider directly by the PGP and will not become part of your medical record due to any activities of the PGP. However, because this information will be publicly available, and may be identified as yours, it could become part of your medical record or be shared with your doctor or other health care provider or your health care insurance provider, or provided to others due to the activities of one or more third parties.

e) Replies to Safety Questionnaires

Your replies to the Safety Questionnaires will be confidential. Data will be used by PGP staff, Clinical Research Monitors, and the Sickkids Research Ethics Board to assess the risks to you and any other study participants. Data may be used to modify this consent form and PGP policies as appropriate. Any changes must be approved by the Sickkids research ethics board. De-identified responses may be used on the public website or elsewhere for purposes of public education, risk management or publication. If you would like your answers to be identified as yours, you may indicate that preference as part of your response to the Safety Questionnaires.

f) Publication of Research Results

The results of this study may be published in a medical book, journal, website or webpage, or used for teaching purposes. Identifying information that is associated with your data and/or the results of this study (such as your photograph, DNA sequence data, and medical or trait information) may be used in such publications or teaching materials. The PGP will not notify you prior to such uses.

g) You will be able to print a copy of this completed form for your records.

Reimbursement:

The PGP will not reimburse you for any costs you may incur, including traveling to or from the medical center. You will not be compensated for any loss of personal time associated with participating in this project.

Participation:

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

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 you any of this money now or in the future because you took part in this study.

If you become ill or are harmed because of study participation, 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 Sponsor of this research is Dr. Stephen Scherer and the Funder of this research is the University of Toronto McLaughlin Centre for Molecular Medicine.

Conflict of Interest:

Dr. Stephen Scherer and the other research team members have no 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 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 (or that of my family) at any healthcare institution, including Sick Kids.
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 except as described to me.
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 have read and understood pages 1 to 16 of this document. I agree, or consent, to take part in this study.

Printed Name of Subject & Age	Subject's signature & date

If you have any questions about this study, please call Steve Scherer, PhD at (416) 813-7613

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."