Discovery DNA Adult Consent Form

You are consenting to have a DNA analysis by Discovery DNA Incorporated (herein "Discovery DNA"). Whether the DNA is your own or you are consenting on behalf of someone for whom you are legally eligible to provide consent, it's important to understand all aspects of the consent. Participation is voluntary. There is no guarantee that Discovery DNA will find or report a DNA finding or, even if there are findings, that the results will lead to a better or worse health outcome. The findings from Discovery DNA are for information purposes only and should not be used to make a decision about any treatments or to make a diagnosis. You should be aware of any other services may be available in your area, including those provided by the health care system that you do not have to pay for, that provide DNA testing for diseases through consultation with a Medical Specialist (such as a doctor or genetic counsellor) that are not associated with Discovery DNA. If you are not sure whether the testing offered by Discovery DNA is what you would like to do, please check with your health care specialist. Review and understand all parts and all pages of the consent.

This consent form is for clients 18 years of age or older providing their own sample for testing, as well as those providing surrogate consent on behalf of an adult client (18 or older) who is unable to provide informed consent for whom you are the legally authorized representative. Please read and complete the Pediatric consent form if you are instead consenting for a child under the age of 18.

What does Discovery DNA offer?

Our DNA contains the instructions our body uses to keep working. The DNA, like letters in a book, is lined up in a "sequence". Groups of sequences are called **genes**. Altogether, there are 3 ½ billion letters in our entire gene sequence – one set from each parent. Sometimes, a change in the letters that make up the DNA, called a "**sequence variant**" can change how a gene works and sometimes the gene may not work at all. Changes in certain genes can lead to changes in health. Sometimes these changes may just lead to the common differences between people – such as one person may have different hair colour than another person. In other situations, the changes can be severe and cause what are called "genetic diseases" like cystic fibrosis, sickle cell anemia or any number of thousands of "rare genetic diseases". Rare diseases may be harder for a health care specialist to find with the usual methods like an X-ray, MRI or other common tests. The information in our genes is not the only thing that affects health - the influence of diet, medication or vitamins that we take, environment and physical activity are some of the and other factors that, combined with genetic factors, play a role in how our genes affect our health.

Discovery DNA looks at thousands of regions of DNA that perform important functions to find these sequence variants. In fact, we check for regions in all the estimated 20,000 genes to look for changes that might lead to a rare disease or sometimes just a change in how the gene works. Regions of the genes that code for special functions of a gene are called exons – therefore, the type of DNA testing performed by Discovery DNA is called an "exome". The detail in an exome may be 100 times higher than DNA tests that just look at your ancestral background. An exome has enough detail to look at ancestral background, but it can also look at entire coding regions of a gene which contains a many hundred million bits of information. This information could be used by your health care specialist to find out if it could be an answer to questions you have about your health. Discovery DNA is not giving you a diagnosis – only your doctor can do that by listening to your concerns, asking questions, checking your family history and performing a physical exam. We leave the diagnosis up to your doctor. We just provide a look into the DNA and this information may help shorten the journey to finding out information about your health.

What will a Discovery DNA result tell me?

It's important to understand what the results in each of these categories means.

The following describes the possible results from the test:

1) **Sequence Variant Detected:** If a sequence variant is reported, it is possible, but not for sure, that the DNA change may result in how the gene works. It does NOT mean the person has a diagnosis. It does not mean the person will get sick or stay healthy. We will not give a diagnosis through a Discovery DNA test. We are just helping you narrow down the search so that you can focus on the DNA changes that your health care specialist may find helpful. The quality of DNA sequencing and interpretation of the data is important to us. We will look to see if the likely impact of the DNA change can be classified using the criteria from American College of Medical Genetics and Genomics. We check the criteria ourselves and do not send the DNA to someone else to check it. It's possible that the same DNA or the sequence variants analyzed in a different method might yield a different result. We recommend reviewing the findings with your health care specialist and more testing will be needed to confirm whether the finding reported by Discovery DNA is genuine.

2) **Sequence Variant Not Detected:** If a sequence variant is not reported, this indicates that we could not find a change in the DNA that, using our methods, is linked to any particular health condition. It does not necessarily mean that there are no serious changes in the DNA – it just means we couldn't find them. A negative result does not guarantee that the person tested is healthy. It might mean that the answer to their health concern does not lie in this type of genetic testing and other causes should be considered with your health specialist. Even the best technology is not fool-proof for finding a DNA sequence variant that might be important – the technology or our process may have missed it. It's also possible that the person who had the test simply does not have a genetic disease. Most of the time, approximately 70% of people, will have a negative result because they simply do not have changes in their DNA that can be linked to a rare disease or simply don't have a rare disease. For some people, it may be reassuring to know that a gene for a rare disease was not reported by this type of testing.

3) **Unexpected results:** In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, the person may be looking for a condition that runs in the family but the DNA sequence variant is for a completely different condition. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

What are the risks and limitations of genetic testing?

- You should not make any decisions about your health based on the results from Discovery DNA. The results are for information purposes only.
- Genetic testing is an important part of the journey in finding whether there are changes in DNA that affect a person's health; however, DNA testing may not always give a definitive answer. In some cases, testing may not find a sequence variant even though one exists. This may be due to limitations in current medical knowledge or testing technology. Some types of disease occur not because of a change in the DNA sequence itself, but because the body adjusts how the DNA region is used – these are called epigenetic diseases. Examples of epigenetic diseases are some causes of Angelman syndrome or RETT syndrome.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in the person whose DNA sample is submitted may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related to each other by a common relative). You should be aware that this information may come out directly from the result of the test or be inferred from the results.
- While genetic testing is relied on by thousands of doctors, errors do occur because of (but not limited to): an incorrect sample being sent to Discovery DNA, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation where the DNA from a blood collection is really that of the transplant donor, the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism), the need to have the testing done from a sample of tissue itself rather than blood or saliva (such as a tissue biopsy) or limitations of the testing methods used by Discovery DNA.

- Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information Discovery DNA used to interpret the results.

Confidentiality and Security

Discovery DNA takes your data confidentiality seriously. We use many methods to maintain confidentiality at Discovery DNA:

1) **DNA and Data Sovereignty.** All the DNA samples and all the information generated stay within Canada. The DNA sample is not sent to any other lab and your DNA data file is not sent to any other lab in any stage of processing.

2) **Data Coding.** The sample will be assigned a digital code instead of a name. Because the data files from DNA testing are large, the most efficient way to store them is cloud data storage. The data is sent from our sequencing facility, within Canada, through a secure connection to cloud storage also within Canada with data security provided by Microsoft. The DNA data is stored in a "scrambled" format called encryption. The data is not stored in any government database and Discovery DNA does not give access to the government or the law enforcement authorities. To maintain this high level of security, once registration is complete and a digital code is given, YOU WILL BE RESPONSIBLE FOR KNOWING WHICH SAMPLE BELONGS TO WHICH CODE. If you lose this information, there is no way for Discovery DNA to retrieve it. Discovery DNA will not give out information by telephone, email or other means. Your secure account login is the only way for you to access the data. When a report is issued, only codes will be used to label the findings and names, addresses or other information that can identify a person will not be used. Discovery DNA can look at the DNA data but cannot link the data to a name, address or identify a person using the DNA data. Once a code is assigned, we simply deal with the code and not names or addresses. Your personal information, such as your name and billing information, is stored entirely separately from your DNA data.

3) **Data Usage.** Discovery DNA does not sell the DNA sequence. Discovery DNA will allow users, whether researchers in universities or commercial users, to ask questions about the data through a secure computer interface. Users can ask, for example, what types of sequence variants exist in the database for cystic fibrosis, or sickle cell anemia, or what proportion of genetic variants may be responsive to a new medication being developed. The replies to their questions through the computer program would give bulk results, what is called aggregate data, and NOT an individual's full DNA sequence. The individual's identity stays protected. The data is stored in such a way that one cannot trace relatives or find parents in cases of adoption using the Discovery DNA database. This allows users to use the DNA data to help understand the links between DNA and health or to discover how new treatments might impact human health.

Specimen Retention and Storage

After testing is complete, the DNA sample is permanently destroyed within 90 days. The DNA sample is not stored. DNA specimens are not shipped out of Discovery DNA facilities to any third party.

What Discovery DNA will NOT do

Testing for certain types of diseases requires counselling before the test is performed to understand the implications of the test result and to prepare the person for the different options if the result is positive or negative. We believe those tests are best ordered through your health care specialist. These include, but are not limited to, conditions such as Huntington disease, some spinocerebellar ataxias, and many forms of cancer to name a few. Discovery DNA will also not test a person under 18 years of age to simply find if they are a carrier for any disease. A carrier is a person who does not have any signs or symptoms of the disease but can have a DNA sequence variant that, when paired with another DNA sequence variant in the same gene from the other parent, can lead to a genetic disease in offspring. If there is a concern about carrier status in a DNA sample in a person under 18 years of age, we suggest that this person contact Discovery DNA once they are 18 years of age to receive counselling on what carrier status means and what his or her options are. This can apply to many types of genetic diseases but most frequently to what are called autosomal recessive diseases. We have this policy because a person under 18 years of age will carry the implications of a positive carrier result for the rest of their lives and we believe they should be old enough to understand how this may affect them later in life.

Review of Consent

I have read this entire Informed Consent document and I give permission to Discovery DNA to perform DNA testing as described in the form above. I understand the limitations of DNA testing and that the results do not provide information that would allow anyone to predict their health outcome in the future or to make any decisions on their health without further testing and consultation with a medical specialist. I understand that I have the legal authority to consent for testing of this sample. I also give permission for information where the identity has been removed to be used for scientific publication or to advance the technologic testing capabilities of Discovery DNA. Discovery DNA has no obligation to provide me with any payment or remuneration for use of the data. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. Permission is also given for Discovery DNA to inform my pre-designated health care provider that I have authorized to receive the results from Discovery DNA. I understand that my full DNA sequence will not be sold or transferred outside the Discovery DNA database. No other warranties, verbal or otherwise, are implied other than the agreement made in this consent form. I release Discovery DNA from any liabilities that are the result of not finding a DNA sequence variant, not reporting a sequence variant, an alternative interpretation of the sequence variant or any delays or inaccuracies beyond the control of Discovery DNA.

Client Name	Client Signature	Date (mm/dd/yyyy)

Physician Name	Physician Signature	Date (mm/dd/yyyy)

Surrogate Consent (if applicable)

If the client being tested is unable to provide informed consent, a legally authorized representative (LAR) may sign below. By signing below, this person hereby testifies that the client being tested understands the contents of this form to the best of their ability and provides their assent where applicable. Please only complete this if you are providing surrogate consent.

LAR Name	LAR Signature	Date (mm/dd/yyyy)

If you wish to change your decisions or have any questions please contact the laboratory via email at info@discoverydna.ca