Yale Pathology Labs
A legacy of caring, a passion for excellence

Blood Disease Reference Laboratory
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Consent to Participate in Genetic Testing

PURPOSE OF GENETIC TESTING:

I understand that the purpose of this genetic testing is to determine whether or not I have inherited a change (mutation) within the ANK1, SPTA1, SPTB and SLC4A1(Band3) gene that is involved in the development of human hereditary blood cell membrane disorder including Hereditary Spherocytosis, Hereditary Pyropoikilocytosis, Hereditary Elliptocytosis, Hereditary Stomatocytosis and Southeast Asian Ovalocytosis (SAO) .This testing will require a blood sample. This test is performed by using Ion Torrent Next Generation Sequencing method.

LIMITATIONS OF GENETIC TESTING:

I realize that this type of testing has limitations. Due to technological limitations, it is possible that I may have a mutation in this gene that is not detected by this testing or a mutation in a different susceptibility gene. In addition, this test is intended to determine whether or not I have inherited a mutation related to hereditary red cell disorders. However, it may provide information about my current health status.

PSYCHOLOGICAL CONSULTATION:

Since this testing will have implications for me and my family members, it has been suggested that I consider consulting with a trained psychiatrist, psychologist or social worker to discuss these issues prior to receiving my results.

RESULTS:

Results will be disclosed in person as part of a counseling session and not by telephone, through the mail, or to my physician. I understand that there are three possible outcomes of this testing:

1. I may learn that I carry a deleterious (disease-causing) mutation within the above-described gene, which will have implications for me, my children, and family members.
2. I may learn that a mutation within the above-described gene was not detected. However, this does not mean that I did not inherit some other genetic mutation of the gene which is not detectable by the current testing technology or that I do not have other related susceptibility factors.
3. I may learn that a variant of uncertain significance (a genetic change whose significance is not yet known) was detected and as a result, the implications may be unclear.

BENEFITS OF GENETIC TESTING:

Benefits have been documented for both those individuals who receive a positive test result and those who receive a negative test result.

A positive test result (finding a mutation) may clarify my health problem, as well as risks to my children and other relatives.

A true negative test result (not finding a mutation in me when the mutation in my family has been identified) can offer reassurance and may lead to reduced anxiety since there would be no increased risk to me or my children.

An uninformative negative test result (not finding a mutation in me when the mutation in my family is not yet known) can also offer some reassurance and may lead to some reduction in anxiety as it may decrease the risks to me and my children, somewhat.

RISKS AND DISCOMFORTS OF GENETIC TESTING:

I understand that there are no unusual risks involved in having my blood drawn. The process may include pain, bruising, and bleeding or lead to an infection at the site of the needle stick (although infection is a rare complication).

There are several psychological and discrimination issues that need to be considered.

Receiving a positive test result may lead to heightened anxiety, and fear. A positive test result may also bring about changes in family dynamics or stigmatization by family members or friends. In addition, there may be concerns of discrimination by my employer and/or insurance company (particularly life and disability).

A true negative test result may cause me to feel guilty. This is often called ‘survivor guilt’ and is experienced when one member of the family tests negative, while others test positive.

An uninformative negative test result or variant of uncertain significance test result may cause me to feel frustrated, confused, or disappointed since the results may not provide definitive information about my risks, risks to other family members, or the cause of related blood cell disorder in my family. Uninformative negative or variant of uncertain significance test results may also provide me with either a false sense of reassurance or anxiety about my risks.

I understand the purpose and potential benefits of the procedure. My doctor has explained to me what results to expect, and the chances of getting those results. I understand that no promises or guarantees have been made or can be made about the results of the procedure(s).

VOLUNTARY PARTICIPATION:

I acknowledge that my participation in this testing program is completely voluntary and will not jeopardize the medical care I receive now or in the future. At any time during this testing process I can decide not to receive my results, to postpone receiving my results, or to withdraw from testing altogether. I realize that in doing so, I will not learn the results of my test.

I give permission to the hospital and its Departments to keep my blood or DNA during the procedure and use them to make a diagnosis, after which the blood sample will be destroyed and the DNA may be used for scientific research or teaching by appropriate persons. If my DNA is used for science or teaching, my identity will not be released. I will no longer own or have any rights to it regardless of how it may be used.

I agree to these conditions. _____
(initials)

CONFIDENTIALITY:

After my in-person result appointment, I understand that I will receive a summary letter with a copy of my test results and this letter and test results will also be sent to my referring health care provider. In order to encourage confidentiality, results of this testing will not be given to any other health care providers unless I specifically make this request. I understand that my insurance company may become aware of this information if they are paying for this testing procedure, or if I otherwise give them permission to access the information.

INFORMATION ABOUT GENETIC TESTING:

Dr. _____ , M.D., has explained this testing to me and has offered to answer any questions that I have. I know that I can contact this individual at _____ if I have any new questions about this testing or about my role in the testing process.

CONSENT OF THE CLIENT:

I have read this consent form and I understand it. The purpose and procedure of this testing program have been explained to me, in addition to the risks, benefits, and limitations of such testing. All of my questions have been answered. I have chosen to participate in this testing program and accept the procedures and risks.

CONSENT FOR RESULT DISCLOSURE:

In the event of a medical emergency, if I am unable to receive the results of my genetic test, I give the following individuals permission to have access to this information:

EMERGENCY CONTACT PERSON:

_____
Name

_____
Street Address

_____
City State Zip Code

_____
Telephone Number

_____ _____
Signature of Client Date

_____ _____
Witness Date

_____ _____
Genetic Counselor/Physician Date

CONSENT FOR RELEASE OF RECORDS TO FAMILY MEMBERS:

The information gathered from genetic counseling and testing often provides important information not only for an individual patient but also for their entire family. Therefore, we encourage individuals to share this information with their relatives. By signing this section, you give the ordering physician(s) permission to share your records with your relatives.

I give my permission to share my test results, pedigree and medical records with any of my relatives.

_____ _____
Signature of Client Date