

ADVICE TO INVESTIGATORS GUIDANCE ON GENETIC RESEARCH: INFORMED CONSENT GUIDELINES

Over the past several years, the field of genetics has seen tremendous growth with many new disease causing genes being identified and primary work on gene therapy being established. As new research is performed, it is important to keep in mind how the findings may impact the patients who are providing us with research materials. For example, individuals who once knew they only had a risk for Huntington disease can now be told early in life if they carry the mutation that almost always results in the disease later in life. Researchers realized that for some patients, this would have a devastating effect on their lives, and specialized screening programs were implemented to identify those individuals who would not benefit from the knowledge. As a result, it has been shown that those to whom the genetic information has been released have benefited from it. In other settings, patients who volunteer for research projects may not be aware of the potential impact of the studies on their lives if key genetic information is discovered. In some cases, genetic diagnosis may result in loss of insurance and possible loss of a job and employment prospects. The New York State Department of Health (NYS DOH), therefore, mandates that all individuals be informed about the possible ramifications of research and clinical testing. The NYS DOH further requires that genetic testing be performed in a NYS approved laboratory if results will be provided to the patient or his/her physician.

***New York State Law definition of Genetic Test:**

“Genetic test shall mean any laboratory test of human DNA, chromosomes, genes, or gene products to diagnose the presence of a genetic variation linked to a predisposition to a genetic disease or disability in the individual or the individual’s offspring; such term shall also include DNA profile, analysis.”

Note: Genetic testing for acquired abnormalities does not require the following consent language to be included. However, if the genetic abnormality is inherited, the following additional language is required.

Research subjects participating in an IRB approved research study must be informed about:

1. Whether or not they will be told the test results.
2. The risk to insurability and potential discovery of non-paternity, if the study poses such a risk.
3. Whether or not genetic counseling will be provided under the study and who will provide/pay for the counseling.
4. If the specimen collected will be used and discarded, or if a portion will be stored for future studies. If the specimen is going to be stored for later use, it can only be used for the studies described in the consent document. If the sample might be used for other research, the subject must be informed of this possibility and given a choice whether he/she wishes to allow his/her sample to be used in that way.

The following are samples of appropriate wording which should be included in the informed consent documents which cover research protocols involving genetic testing.

INSURABILITY

Some tests reveal information that may affect a person's ability to get or keep medical and/or life insurance. The tests done under this study may reveal (insert appropriate language), which may affect your ability to get or keep medical insurance.

NON- PATERNITY

Genetic tests may reveal other information unrelated to this study. For example, in cases where parents and children are both tested, the test may disclose the possibility that the father is not the biological parent.

TEST RESULTS & COUNSELING

Your (type of specimen) may be tested for genetic factors and the information obtained may reveal genetic information about you.

AND

(Include one of the following additional paragraphs)

Since the significance of these tests is not known for you, we will not release the results of any genetic testing. No formal counseling will be provided under this study.

OR

The results of these tests will be made available to your primary care physician. No formal counseling will be provided under this study. If you request, you will be referred to a genetic counselor. However, you or your insurance carrier will be responsible for the genetic counselor's fee.

OR

The results of these tests will be made available to your primary care physician and formal counseling will be provided under the study at no cost to you.

Note to the Investigator. If genetic test data are released to a physician, the test must be NYS approved and must have been performed in an NYS licensed laboratory. If the test performed is not NYS approved, results can only be released to a physician if an "orphan disease exemption" is obtained from Dr. Ann Willey, NYS DOH. A separate exemption must be obtained for each patient for whom results will be released. A copy of a sample "orphan disease" request can be obtained from the IRB office. Please note, that in these cases there are additional requirements which must be met as mandated by the New York State Civil Rights Act, Section 79-1.

FURTHER TESTING

The (type of specimen) you provide will be used only for the research described in this document. At the end of the research project, any leftover samples will be destroyed.

OR

The (type of specimen) you provide will be used for the research described in this document. At the end of the research project, if the sample is deemed medically relevant, it may be used as a control for future studies. In this situation, the specimen will be anonymized and all identifying information removed so that it cannot be traced back to you.

FURTHER TESTING AND SPECIMEN STORAGE

See Human Specimen research: Generic consent statement.