

**COLUMBIA UNIVERSITY INSTITUTIONAL REVIEW BOARD
POLICY ON RESEARCH INVOLVING GENETIC TESTING
UNDER SECTION 79-L OF THE NEW YORK STATE CIVIL RIGHTS LAW**

I. BACKGROUND

Section 79-L of the New York State Civil Rights Law (“Section 79-L”) was enacted in 1996 to provide asymptomatic individuals sufficient information to assess the benefits and consequences of predispositional genetic testing. Unlike diagnostic genetic tests that confirm the presence or absence of a disease, predispositional genetic tests cannot predict with certainty the risk of developing a disease. This type of testing can be confusing because the results may not provide answers to when or even if a disease will appear and can have profound implications about an individual’s future or the future of one or more family members.

Section 79-L covers the informed consent requirements for the use of biological samples containing genetic material in both the clinical and research contexts. It also provides safeguards for maintaining the confidentiality of genetic data.

II. SCOPE

This Policy applies only to the use of biological samples for genetic testing covered by Section 79-L in research (“Section 79-L Research”).

Although this Policy only applies to Section 79-L Research, it may be advantageous that the informed consent form for any research that involves the collection of Biological Samples to be used in current research or stored for future research purposes comply with Section V(B) and, if applicable, Section V(D) of this Policy in case genetic testing may be performed on such Samples, even if not planned at the time such Samples were obtained. This will allow the maximum flexibility with respect to the use of the Samples as it is often not possible to predict when such Samples might be used for genetic testing.

III. EFFECTIVE DATE: February 12, 2016; revised March 31, 2016 and March 1, 2017. This Policy replaces the Columbia University Institutional Review Board Guidance on Research Involving Genetic Testing, dated December 23, 2010.

IV. DEFINITIONS

Certain terms used in this Policy are defined as follows:

Biological Sample: any material part of the human body or of discharge therefrom known to contain DNA, including, but not limited to, a tissue specimen, blood or urine.

Genetic Predisposition: the presence of a variation in the composition of the genes of an individual or an individual’s family member which is scientifically or medically identifiable

and which is determined to be associated with an increased statistical risk of being expressed as either a physical or mental disease or disability in the individual or having offspring with a genetically influenced disease, but which has not resulted in any symptoms of such disease or disorder.

Genetic Test: any laboratory test of human DNA, chromosomes, genes or gene products or any DNA profile analysis used 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.

V. POLICY

A. RETURN OF RESULTS FROM GENETIC TESTING

There is no requirement under Section 79-L that research participants be informed of the results of Genetic Tests and the Institutional Review Board ("IRB") should not require that investigators inform participants of such results. The consent form for studies in which results of such Genetic Tests will not be provided to participants should clearly state that fact. If, however, the results of any Genetic Test are to be provided to a participant, it is a University requirement that such results be confirmed by a CLIA or New York State certified laboratory prior to being released to the participant.

Informed consent for genetic testing may be obtained in person by a member of the research team so long as such person has been certified by the University as having completed training in obtaining consent for genetic testing (a "Qualified Consenter"), or remotely through electronic means, so long as a Qualified Consenter is available by telephone or other interactive media to answer questions of potential subjects.

However, results may be returned only to those participants whose informed consent included the elements described in Section V(B) below. If the Genetic Test for which results will be returned was performed on a stored biospecimen for which only General Research Consent had previously been obtained, additional consent that includes these elements is required. The original signed consent form must be retained by the responsible study physician in the research record.

B. GENETIC TESTING ON BIOLOGICAL SAMPLES COLLECTED WITH THE INTENT TO BE USED FOR KNOWN RESEARCH PURPOSES

Prior to collecting a Biological Sample of an individual for a known research purpose that includes Genetic Testing, and prior to performing any Genetic Test on the Sample, the following requirements must be met:

1. A protocol for the research has been approved by the IRB;
2. Written informed consent including the following information has been obtained from such individual:

- (a) a general description of the Genetic Test;
- (b) a statement of the purpose of the Test and that the individual may wish to obtain professional genetic counseling prior to signing the consent form;
- (c) a statement that a positive Test result is an indication that the individual may be predisposed to or have the specific disease or condition tested for and may wish to consider further independent testing, consult his/her physician or pursue genetic counseling;
- (d) a general description of each specific disease or condition tested for;
- (e) the level of certainty that a positive Test result for that disease or condition serves as a predictor of such disease or condition unless no level of certainty has been established;
- (f) the name of the person or categories of persons or organizations to whom the Test results may be disclosed;
- (g) a statement that no test other than that authorized shall be performed on the biological sample and that the sample shall be destroyed at the end of the testing process or not more than 60 days after the Sample was taken, unless a longer period of retention is expressly authorized in the consent; and
- (h) the signature of such individual or, if such individual lacks the capacity to consent, the signature of the person authorized to consent for such individual,

provided that the requirements of subsections (c), (d) and (e) above may be modified by the IRB if the applicable research protocol does not permit such degree of specificity.

C. GENETIC TESTING ON ANONYMOUS BIOLOGICAL SAMPLES TO BE USED FOR RESEARCH

A Genetic Test on an anonymous Biological Sample to be used for research purposes may be performed without the prior written consent of the individual from whom the Biological Sample was collected so long as the research will be conducted pursuant to a protocol approved by the IRB that assures the anonymity of the source of the Sample.

D. GENETIC TESTING ON STORED BIOLOGICAL SAMPLES COLLECTED FOR GENERAL RESEARCH PURPOSES

Prior to performing any Genetic Test on a Biological Sample of an individual who at the time that the Sample was stored provided consent to use the Sample for general research purposes, the following requirements must be met:

1. A protocol for the research has been approved by the IRB;
2. Either (a) the Sample has been permanently stripped of identifying information or (b) a coding system that has been approved by the IRB has been established to protect the identity of such individual; and
3. Written informed consent including items (a) through (e) below has been obtained from such individual, and such consent did not specify time limits or other factors that would restrict the use of the Sample for a Genetic Test.

- (a) a statement that the sample will be used for Genetic Tests;
- (b) the time period during which the Sample will be stored, or if no time limit is specified, a statement that the Sample will be stored for as long as deemed useful for research purposes;
- (c) a description of the policies and procedures to protect patient confidentiality;
- (d) a statement of the right to withdraw consent to use of the Sample at any time and whom should be contacted to withdraw consent;
- (e) a statement allowing the individual to consent to future contact for any or all purposes, including the following:
 - (i) research purposes;
 - (ii) provision of general information about research findings; and
 - (iii) information about the Genetic Test on the individual's Sample that may benefit him/her or his/her family members with respect to his/her/their choices regarding preventive or clinical care; and
 - (iv) a statement explaining the benefits and risks of consenting to such future contact,

provided that in no event shall information about the results of a specific Genetic Test on a Biological Sample be disclosed to the individual from whom the Sample was collected or, subject to Subdivision 4(c) of Section 79-L, any other individual without obtaining an additional written informed consent complying with the provisions of Section V(B) above from the recipient individual.

E. GENETIC TESTING ON STORED BIOLOGICAL SAMPLES COLLECTED WITHOUT CONSENT FOR GENERAL RESEARCH PURPOSES

Prior to performing any Genetic Test on a Biological Sample of an individual who provided consent to store the Sample, but at the time the Sample was stored did not provide specific written consent for the Sample to be used for Genetic Testing or general research purposes, written consent complying with the provisions of Section V(B) must be obtained from such individual, provided that in no case shall Genetic Testing be performed on any Sample that has been stored for more than 10 years, unless explicit consent is obtained.

These requirements also apply for use of residual clinical Biological Samples, when the clinical consent form included a statement that such Samples may be used for the advancement of medical knowledge, and no other consent for future research was obtained.

F. CONFIDENTIALITY PROTECTIONS

1. Records and Release of Information

All records, findings and results of any Genetic Test performed on an individual shall be deemed confidential and shall not be disclosed to any person or organization without the specific written consent of the individual. In no event shall any information (a) about an individual derived from a Genetic Test on a stored Biological Sample collected from such individual or (b) linking such individual with specific results of a Genetic Test be released to any person or organization without the explicit written consent of such individual.

2. Destruction of Sample

If consent of an individual to the storage of his/her Biological Sample is withdrawn at any time, the person or entity storing the Sample shall promptly destroy the Sample or portions thereof that have not already been used for research purposes.

3. Family Member Contact

In no event shall any family member of an individual who provided a Biological Sample for Genetic Testing be contacted for clinical, research or other purposes without the consent of such individual specifically indicating which family members may be contacted and the specific purpose of the contact.