

## **GUIDANCE ON RESEARCH INVOLVING GENETIC TESTING**

### **I. SCOPE:**

This guidance applies to all human subjects research that involves Genetic Testing, as defined below. It provides background information on New York's Civil Rights § 79-1, which governs the confidentiality of Genetic Testing records, and discusses the implications of this law for institutional review boards (IRBs) that review human subjects research involving Genetic Testing, particularly with respect to the criteria for IRB approval of such research and the requirements for obtaining informed consent and laboratory certification.

### **II. REVISED DATE:**

**January 1, 2015**

### **III. BACKGROUND:**

New York Civil Rights Law §79-1 was enacted in 1996 to protect the privacy of genetic information by prohibiting the performance of Genetic Tests on biological samples without the prior written informed consent of the individual tested, restricting the disclosure of the results of Genetic Tests, limiting additional tests that can be performed on samples without the individual's prior consent, and requiring the destruction of the sample within 60 days unless the individual consents to a longer period of time.

**Definition of a Genetic Test under §79-1.** The New York Law has a specific and limited definition of Genetic Test. It only applies to tests of human DNA, chromosomes, genes, or gene products conducted to learn whether an asymptomatic person has a genetic predisposition to a disease or disorder.<sup>1</sup> It does not apply to the following:

- Tests performed on a person with symptoms of the disease or disorder; or Tests performed for the purpose of answering a research question, where the ultimate purpose of performing the test is not to identify a genetic variation. An example of such a test would be a test to gauge the efficiency of a diagnostic test by comparing it to another test.

### **IV. GUIDANCE – Review and Approval of Research involving Genetic Testing:**

---

<sup>1</sup> A "genetic predisposition" means the presence of a variation in the composition of the genes of an individual or an individual's family member that is scientifically or medically identifiable and 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.

In reviewing IRB protocols involving genetic research, the IRB must determine, first, whether the genetic research meets the criteria for a Genetic Test as defined by the New York State Law. If so, then the review and approval process must ensure that the protocol complies with the law. In so doing, the IRB must next determine whether the genetic testing is being performed on preexisting anonymous samples, whether it is being performed solely for research purposes (in which case the procedures do not need to meet the requirements of the law that apply to clinical genetic testing), or whether the genetic testing is being done (in part or in whole) for clinical purposes.

These distinctions are important because the requirements of §79-1 differ depending on whether the Genetic Test is being performed on pre-existing anonymous samples, being performed solely for research purposes, or being performed (even in part) for clinical purposes.

**1. Genetic Tests performed for research or statistical purposes on pre-existing anonymous.**

Section 79-1 does not require informed consent for Genetic Tests performed on pre-existing anonymous samples, for which there is no identifiable person from whom to obtain consent, provided that the IRB determines that the research protocol assures the anonymity of the sources of the samples.

**2. Genetic Tests performed for research purposes only.**

Genetic Tests may be performed on samples for general research purposes if (a) the individual providing the sample gave prior written informed consent for the use of their samples for the general research purpose *and* (b) the samples have been permanently stripped of identifying information or an IRB-approved coding system has been established to protect the identity of the individuals who provided, or will provide, the samples. If the protocol contemplates that a portion or all of the samples will be matched up with identifying information to communicate the results of the tests to the individuals who provided the samples, then the provisions of New York's Civil Rights Law §79-1 that apply to clinical genetic testing apply to the research. These provisions include requirements that informed consent as set forth below for "Genetic Tests performed, at least in part, for clinical requirements" must be obtained and the testing must be performed in a NYS-approved laboratory (or a waiver from NYS from this requirement must be provided).

Written informed consent for use of genetic samples for *research purposes only* must include the following:

- a) statement that the sample will be used for future Genetic Tests;
- b) the time period during which the tissue will be stored, or if no time limit is specified, a statement that the tissue 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, at any time, consent to future use of the tissue, and the name of the organization that should be contacted to withdraw consent;
- e) a statement allowing individuals 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 test on their sample that may benefit them or their family members in relation to their choices regarding preventive or clinical care<sup>2</sup>, and
- f) a statement explaining the benefits and risks of consenting to future contact.

**The provisions of New York’s Civil Rights Law §79-1 that apply to clinical genetic testing must be met by the research protocol if the investigator subsequently proposes to do any of the following:**

- contacting the individual’s family members for clinical, research or other purposes;
- disclosing information derived from the Genetic Tests performed on the sample or information linking an individual with specific results of Genetic Tests to any organization or person; or
- disclosing the specific results of a Genetic Test to the individual (in such circumstances the written informed consent requirements set forth below for “Genetic Tests performed, at least in part, for clinical purposes” must be met).

Note that modifying the protocol such that it falls under the clinical testing requirements of the law means that the consent form and consent procedure must meet these requirements. This may require that consent be re-obtained from participants if the original consent was not for clinical genetic testing, and that the laboratory procedures be done in a New York State certified laboratory (or waiver obtained), which may require re-analysis of the samples if they were not originally analyzed in such a laboratory.

Note also that analyses performed in a non-certified laboratory may not be used to screen or triage samples for analysis in a NY State certified laboratory, if the research falls under the requirements for clinical genetic testing. Such a procedure would allow confirmation of true positives but does not address the potential for false negatives. All of the study samples would need to be re-analyzed in a certified laboratory, if not originally done, if such a change in the protocol were to be approved.

---

<sup>2</sup> Information about the test that may benefit subjects or their family members would not necessarily include test results. If the test results are to be disclosed to the subject, then prior written informed consent for clinical testing must be obtained.

There is no requirement under New York law that research participants be informed of the results of genetic tests. In general, the IRB should not require that investigators inform subjects of test results, so long as the consent form states that the test is being done solely for research purposes and that the results will not be provided to the participant or the participant's physician.

### **3. Genetic Tests performed, at least in part, for clinical purposes.**

If any results of the Genetic Test are to be communication to the research participant or the participant's doctor, then the Genetic Test is being performed, at least in part, for a clinical purpose, and all of the requirements of New York Civil Rights Law §79-1 that pertain to clinical genetic testing must be incorporated into the research protocol. The IRB does not have the authority to grant exceptions or exemptions from the NYS law.

#### ***a. Consent***

Written informed consent must be obtained before conducting the test. The informed consent for clinical testing must contain the following elements:

- i) a general description of the test;
- ii) a statement of the purpose of the test;
- iii) a statement indicating that the individual may wish to obtain professional genetic counseling prior to signing the informed consent;
- iv) a statement that a positive 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 their physician or pursue genetic counseling;
- v) a general description of each specific disease or condition tested for;
- vi) the level of certainty that a positive result for that disease or condition serves as a predictor of such disease. If no level of certainty has been established, this subparagraph may be disregarded.
- vii) the name of the person or categories of persons or organizations to whom the test results may be disclosed;
- viii) a statement that no tests other than those 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 sixty days after the sample was taken, unless a longer period of retention is expressly authorized in the consent; and
- ix) the signature of the individual subject of the test, or if that individual lacks the capacity to consent, the signature of the person authorized to consent for such individual.

\*The requirement of elements (iv), (v) and (vi) may be modified by the IRB if the research protocol does not permit such degree of specificity.

The original consent must be retained by the responsible study physician in the research record with a copy in the medical record.

Additional written consent must be obtained prior to:

- disclosing the Genetic Test results to persons or organizations not named on the informed consent form;
- conducting Genetic Tests other than those for which specific written informed consent was obtained (except when necessary to demonstrate the integrity of the sample or to resolve the analysis of a test with a previously indeterminate result);
- retaining the sample for longer than 60 days for scientific research.

***b. Laboratory Certification***

If a genetic test is performed, at least in part, for clinical purposes, the test must be performed in a CLIA certified lab and the test must either be FDA or NYS approved.

- If the genetic testing is on tumor samples please note that somewhat more flexible rules may apply, and the investigator should consult with the PPHS.
- The results of tests which are not FDA or NYS-approved (even if done in a CLIA certified lab) cannot be shared first and confirmed later. A general statement can be made to the subject that preliminary results indicate a confirmatory test is in order, but saying something to the effect that this or that was found in your genome and should be confirmed is NOT allowed.