

DEPARTMENT: Yale University Human Investigation Committee
POLICY NUMBER:
SECTION: Genetic and Related Testing
REVIEW RESPONSIBILITY: IRB Leadership Committee
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Yale University School of Medicine HIC Policy Regarding: Genetic and Related Testing

Introduction

This policy addresses protocols in which there is genetic testing. It has developed out of concern about the individually identifying information that is available from genetic test results. The policy is meant to apply to the various kinds of individually identifying tests and research that use biological materials, e.g., DNA, RNA, proteins, chromosomes, tissues, and cells (that contains DNA, etc.). The policy also has relevance to the banking of these materials.

1. The scope of research with DNA or tissue should be described in the consent form in terms of a general area of biology or human function/behavior (e.g., cancer, aging, embryo development, phobias, intelligence, learning, sexual behaviors) such that the concept would be understandable to the research subject.

Alternatively, the investigator may request completely unrestricted or open-ended current or future use of banked samples. If such a request is made, the consent form should state in easily understood, plain language the wide scope of use that is being requested and the consent process should emphasize the same. If a consent form states a general area of biology as the scope of the research, the investigator is required to return to the subject to request permission to enlarge the scope unless the HIC approves the change without requiring further consent. In this latter circumstance, the HIC would be responsible for deciding whether unforeseen and unrequested uses would necessitate further consent.

2. Length of time that a sample would be stored must be stated (e.g., specific period of years, indefinitely, until exhausted) both in the protocol and in the consent form.

3. That there are risks if genetic information becomes known to the subject or to others must be stated explicitly in the consent form.

Depending on the subjects and the nature of the protocol, the magnitude of the risks and a description of the risks should be given in both the protocol and the consent form. The extent of the description of these risks may be extensive or may be brief, depending on the protocol. These risks may include:

a. Access to or retention of benefits or entitlements (e.g., health insurance, life or disability insurance, educational opportunities, employment, etc.)

b. Stigmatization: views of others, within or without the subject's family, about the subject; possibility of altered family relationships and interactions.

c. Psychological responses to information: altered self-concept; possible feelings of depression, guilt, anger, etc.

d. Detection of biological relationships within a family: paternity, maternity, adoption.

4. Plans for return of information to subject, to medical records, to primary care physicians, or to other places (or not to return information) should be explicitly stated in the protocol and, in most circumstances, in the consent form. If there is a plan or intent to return information, the information should usually be confirmed in a CLIA approved laboratory. The HIC may waive this requirement in some circumstances.

5. Confidentiality and anonymity. Statements should be made in the protocol and in the consent form regarding mechanisms by which confidentiality or anonymity will be accomplished and maintained and who will be able to link names with data. Usually, the term anonymous would not be used in a consent form because of ambiguity in many persons' understanding of the meaning of the term. Alternatively, the term might be used along with a clear definition of its meaning.

The concept of the term anonymous, in this context, is that no one could link the information from the DNA, protein, etc. to a specific subject by any reasonably executed mechanism. If someone could link the information to an individual subject, the proper term is confidential. Individually identifying information does reside within DNA itself. Thus, in addition, the concept of anonymous includes the presumption that an investigator would not make any effort to link information to an individual subject.

If samples will leave Yale (to other investigators, etc.) they must be stripped of identifiers, in most circumstances. If samples go with identifiers or if they are linkable to the subject by the outside investigator or user, this must be explicitly stated to the subject and consent obtained (i.e., hospital based clinical consent is not sufficient). If they remain linkable to the subject only via a Yale investigator, hospital based clinical consent may be sufficient in some circumstances but not in others.

6. Withdrawal. Explicit statements must be made in the consent form about the right to withdraw from a research study. Often, the investigator would explain, that if a subject so requests, withdrawal will result in no further use of the subject's linked or linkable sample (DNA, tissue, etc.), but that data already derived from the sample will continue to be used in the research.

An investigator may wish to offer only the option of rendering a sample anonymous for further use rather than the option of destruction of the sample. This mechanism should be stated in the protocol and offered clearly in the consent form to the subject. The default will be destruction if the protocol does not offer an option or if the subject is not asked to exercise (or does not exercise) the anonymity option upon his or her withdrawal. An investigator could also offer alternatives of destruction or making the sample anonymous.

7. **Successors.** The PI has the responsibility of assuring agreement, from subsequent persons who might exercise control over samples, to the continuation of the protocol and the consent form statements about protections of information. The HIC should inform and educate PI's about this responsibility.

8. Genetics/DNA protocols will be acceptable for expedited review when HIC personnel feel that the scope of the research is readily understood and would not disturb a large majority of subjects and the risks of information becoming public are minimal, the risks are being adequately and realistically stated, and the risks, themselves, are not beyond a generally accepted concept of being qualitatively minimal.