

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 1 OF 7	

### 1. PURPOSE

This policy protects the rights and confidentiality of subjects involved in genetic research at Creighton University, while facilitating the investigator's ability to carry out his/her research program.

### 2. TERMS AND DEFINITIONS

- 2.1. Sampling and analysis of genetic material is one of the most active and rapidly changing methods in modern biomedical research. It is also associated with a growing number of ethical, regulatory, and public relations concerns for human subjects who donate DNA for analysis. Concerns exist about proprietary interests in donated genetic materials that lead to profitable biotechnologies. Confidentiality is also a concern because the information gained from genetic testing could have an adverse effect on employability, insurability, and future health care, and may represent a risk of social discrimination or criminal prosecution. In genetic studies, these same risks could apply to family members and ethnic groups associated with the research subjects.
- 2.2. The extent to which a research sample can be linked to the identity of its source is significant in assessing the risks and potential benefits to human subjects. For this reason, the National Bioethics Advisory Commission has developed guidance on the ethical issues associated with research involving human biological materials,<sup>1</sup> including definitions to describe the extent of personal information associated with particular samples of human biological materials. These definitions describe human biological materials that are held in clinical facilities, repositories, or in the laboratories of individual investigators. These definitions will assist the investigator in determining what type of consent/assent document needs to be produced for the investigation in question Any investigator who plans to isolate DNA from patient samples should be familiar with the following definitions:

---

<sup>1</sup> National Bioethics Advisory Committee. [Research Involving Human Biological Materials: Ethical Issues and Policy Guidance, Volume 1](#). August 1999.

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 2 OF 7	

### 2.2.1. Repository Collections

- 2.2.1.1. **Unidentified Specimens**—for these specimens, identifiable personal information was not collected, or, if collected, was not maintained and cannot be retrieved by the repository.
- 2.2.1.2. **Identified Specimens**—these specimens are linked to personal information in such a way that the person from whom the material was obtained could be identified by name, patient number, or clear pedigree location (i.e., his/her relationship to a family member whose identity is known).

### 2.2.2. Research Samples

- 2.2.2.1. **Unidentified Samples**—sometimes termed “anonymous,” these samples are supplied by repositories to investigators from an unidentified collection of human biological specimens.
- 2.2.2.2. **Unlinked Samples**—sometimes termed “anonymized,” these samples lack identifiers or codes that can link a particular sample to an identified specimen or a particular human being.
- 2.2.2.3. **Coded Samples**—sometimes termed “linked” or “identifiable,” these samples are supplied by repositories to investigators from identified specimens with a code rather than with personally identifying information, such as a name or Social Security number.
- 2.2.2.4. **Identified Samples**—these samples are supplied by repositories from identified specimens that have a personal identifier (such as a name or patient number) that would allow the researcher to link the biological information derived from the research directly to the individual from whom the material was obtained.

## 3. IRB REVIEW OF HUMAN GENETIC RESEARCH PROJECTS

All human genetic research projects shall be submitted to the IRB office for review and approval. The type of review required depends on the specific characteristics of the study, as described below.

## **Policies and Procedures**

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 3 OF 7	

### **3.1. Determination of Exempt Status or Expedited Review**

3.1.1. A genetic research project is not subject to full board review if either of the following conditions are met:

- 3.1.1.1. The samples are unidentified or have been irreversibly unlinked from the identity of the subject.
- 3.1.1.2. The subject is deceased and the research would not produce information that can be linked to living groups

### **3.2. Full Board Review**

3.2.1. Full board review is required for a genetic research project if either of the following conditions are met:

- 3.2.1.1. The sources of the samples are living individuals and the source will be identified directly or by code.
- 3.2.1.2. The research is on samples not identified with individuals but nevertheless poses a significant risk of harm to groups associated with the sources. (If the samples are identifiable with relatives, an ethnic group, or a defined class of people such as residents of a specific locale, the study must be designed to avoid harm to the associated group.)

## **4. RESEARCH USING SAMPLES DONATED WITH CONSENT FOR UNSPECIFIED FUTURE USE**

### **4.1. IRB Approval**

4.1.1. IRB approval is required for research that uses samples previously donated with consent for unspecified future research. IRB approval applies only to the research specifically detailed in the submitted protocol. Consent by the research subject to unspecified future use of biological samples collected under an approved protocol does not eliminate the need for IRB review and approval of the future research. IRB review of such research shall include consideration of whether the research falls within the scope of the consent document, whether the consent document adequately informs the subject of risks presented by the new research, and whether the new

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 4 OF 7	

research presents risks that could not have been anticipated by the donor at the time of consent. The IRB may require re-consent of the original donor (research subject) if it feels that the new research entails risks that the donor was not aware of at the time of the original consent.

### 4.2. Sharing Samples with Other Institutions

4.2.1. If samples donated with consent for unspecified future research under a Creighton University-approved protocol are subsequently requested for use by investigators at another institution, the following conditions apply:

- 4.2.1.1. The samples must be sent to the outside investigators or organization without any identifier (anonymized) or with only coded identifiers.
- 4.2.1.2. The original investigator at Creighton University must be the only means of linking the sample to the subject.

## 5. INFORMED CONSENT FOR GENETIC STUDIES

### 5.1. When is Informed Consent Required?

5.1.1. Informed consent is required if **any** of the following conditions are met:

- 5.1.1.1. The research proposes to collect new samples or to use samples collected after January 1, 2000.
- 5.1.1.2. The research involves more than minimal risk to the subject. Risk includes psychosocial risks such as insurance and employment discrimination, familial conflict, stigmatization, and anxiety.
- 5.1.1.3. Failure to seek consent will adversely affect the rights and welfare of the subject. Rights include the right to privacy, the right to control access to personal information about oneself, and the right not to donate materials for research that the subject might consider objectionable.

5.1.2. Informed consent is not required if **all** of the following conditions are met:

- 5.1.2.1. The research involves existing samples (samples already collected

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 5 OF 7	

and stored before January 1, 2000; no new samples obtained during the course of the project).

- 5.1.2.2. The research involves no more than minimal risk to the subject.
- 5.1.2.3. The failure to seek consent will not adversely affect the rights and welfare of the subject. If there is a previous consent document, the proposed use must be consistent with the subject's likely understanding of how the sample would be used.
- 5.1.2.4. The research is scientifically sound and could not be carried out if consent were required.

### 5.2. Elements of Informed Consent

- 5.2.1. Informed consent documents for research involving genetic testing shall include the basic elements required for informed consent as defined by the FDA and the DHHS and as described in IRB Policy 118, "[Informed Consent](#)." In addition, informed consent documents for genetic research shall address some of the specific concerns associated with genetic testing.
- 5.2.2. When preparing an informed consent document for genetic research, investigators shall address the psychosocial risks associated with genetic testing, such as insurance and employment discrimination, anxiety (such as knowledge of predisposition to future disease), familial conflict (e.g., if other family members do not want to know of predisposition to future disease or untreatable disease), stigmatization, and group-related harms. Risks associated with a possible breach of confidentiality also should be addressed. Investigators shall include a clear statement of the scope of the research for which consent is being sought and, if consent is sought for broader use of genetic samples in future research, shall discuss the possible consequences of such consent.
- 5.2.3. Investigators shall clearly state the provisions for maintaining privacy and confidentiality and shall discuss under what circumstances, if any, subjects will be re-contacted. Investigators shall state the circumstances under which findings from the research will be communicated to the subjects. To avoid engendering unnecessary anxiety or medical interventions, findings should be communicated to the subjects only if they are confirmed and reliable and suggest a course of action that can avoid or ameliorate potential injury. If the study is unlikely to produce information that would

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 6 OF 7	

be medically useful to the individual, the consent document should state that no study results will be provided to the subject or his/her doctor.

5.2.4. Investigators shall include a provision for withdrawal of consent and, if the consent is to be sought in a clinical setting, shall state that refusal to consent to the research use of biological materials will not affect the provision of clinical care.

5.3. For more detailed information on what must be included in informed consent documents for genetic research projects and examples of appropriate consent language, refer to "[Model Consent Documents](#)."

## 6. INFORMED CONSENT PROCESS

### 6.1. Type of Genetic Study

- 6.1.1. Genetic studies can be stand-alone studies, part of an associated clinical investigation, or limited to storage of tissue for undefined future use.
- 6.1.2. If the study includes therapeutic or non-therapeutic interventions that are not considered genetic research, a separate consent document shall be drawn up for the clinical investigation aspect of the study.
- 6.1.3. If storage for undefined future use is requested, a separate consent addendum for storage shall be required.
- 6.1.4. In these situations, all consents (clinical investigation, genetic testing and storage for undefined future use) shall be signed if the subject is to participate in all aspects of the study.
- 6.1.5. Investigators shall not require subjects in a clinical investigation to participate in a genetic protocol unless the genetic testing is directly relevant to the subject's disease and is an integral part of the protocol. Otherwise, a potential subject who objects to participating in the genetic testing portion of the study or storage of tissue for undefined future use shall still be allowed to participate in the clinical investigation portion of the study.

## Policies and Procedures

SECTION: Human Research Protection Program	NUMBER: 112	
CHAPTER: Institutional Review Board	ISSUED: 2/2000	REVISED: 9/2004; 6/2006; 4/2009
POLICY: Guidelines for Genetic Research	PAGE 7 OF 7	

### 6.2. Discussion of Risks/Benefits of Genetic Testing

- 6.2.1. Because of the sensitive and uncertain nature of genetic testing, the process of consent to participate in genetic testing studies shall include a thorough discussion of the risks and benefits associated with participation. In some cases, these discussions may be beyond the capabilities of the Principal Investigator of the study and it may be necessary to make arrangements for another qualified party with experience in genetic counseling and testing to represent the Principal Investigator to the potential subjects.

### 6.3. Informing Subjects of Genetic Test Results

- 6.3.1. Investigators should be prepared to inform subjects of the availability of research results in the event that these research results are, in the investigator's opinion, of material interest to the subject. The investigator shall make a reasonable effort to contact the subject and inform the subject that study results are available. The subject has the right to refuse to be informed of the research results. If the results of the research would be, in the opinion of the investigator, of significant health benefit to the subject, the investigator shall ensure that a qualified process of disclosing either direct or incidental results to the subject is conducted by a person qualified to discuss the significance, consequences, and reliability of such results. This qualified person should be a genetic counselor. The investigator, or trained genetic counselor, should remind the subject of the risk associated with genetic testing prior to disclosing those results.