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## Statement on Storage and Use of Genetic Materials

### *American College of Medical Genetics Storage of Genetics Materials Committee*

#### Introduction

The sensitivities of DNA analytic methods have increased dramatically in the past several years. Use of such tests to analyze an individual's genome could reveal parental origin or provide forensic evidence, as well as determine an individual's complement of normal and abnormal genes. Some of the abnormal genes detected cause diseases in infancy, while the effects of others may become manifest only in adulthood. Finally, detection of specific, acquired genomic changes may indicate increased susceptibility to or herald the onset of certain malignancies.

Many health professionals as well as lay people may not appreciate how frequently biological samples are stored and how easily samples that have been stored for an unrelated reason could be used for genetic analysis in the future. The potential problems posed by such uses were explored at length in a Workshop sponsored by the National Center for Human Genome Research and the Centers for Disease Control and Prevention (Clayton et al., in press). ACMG members, recognizing these issues, may have concerns about access by insurers, employers, and others to samples or test results. Developing practices to be used at the time samples are obtained could alleviate problems that might arise in the future as the breadth and scope of potential genetic analyses increase (McEwen and Reilly 1995).

An important issue in genetic testing is defining the scope of informed consent. The obligation to counsel and obtain consent is inherent in the clinician-patient and investigator-subject relationships. In the case of most genetic tests, the patient or subject should be informed that the test might yield information regarding a carrier or disease state that requires difficult choices regarding their current or future health, insurance coverage, career, marriage, or reproductive options. The objective of informed consent is to preserve the individual's right to decide whether to have a genetic test. This right includes the right of refusal should the individual decide the potential harm (stigmatization or undesired choices) outweighs the potential benefits (Office for Protection from Research Risks 1993; Elias and Annas 1994).

#### Technical Considerations

DNA can be derived from a wide variety of biologic or genetic materials including blood and viable, fixed and/or frozen cells and tissues. For example, archived blood spots from newborn screening, hair roots, bones, teeth, and pathology samples and slides can be used as alternatives to blood or tissue samples as sources of DNA for diagnostic or forensic purposes. Because DNA can be obtained from all these materials, a brief review of guidelines for DNA banking is appropriate.

Stored DNA can be used to perform presymptomatic, carrier, or prenatal diagnoses using molecular analysis. Stored DNA can also provide material for research into the molecular alterations associated with population diversity and genetic disorders. At present, materials from which DNA can be obtained are held at a variety of sites. These include but are not limited to civilian and military research, pathology, forensic, and newborn screening laboratories. Technical guidelines regarding storage of separate DNA aliquots in different locations, temperature of storage, optimal methods of sample identification, duration of storage, sample and data retrieval, and storage of filters and dried blood spots are being developed (Yates et al. 1989; McEwen and Reilly 1994).

Among the legal and ethical issues posed by storage of DNA are deciding which samples to store and defining to whom and under what conditions samples will be made available. In selecting individuals to be sampled, one must consider currently diagnosable disorders or disease susceptibilities as well as the potential for testing yet-to-be-identified genes.

#### Recommendations

- I. *Collection and Storage of Samples That May Be Used for Genetic Analysis in the Future* (Office for Protection from Research Risks 1993; Elias and Annas 1994; Clayton et al., in press)
  - A. *When obtaining samples for clinical tests, clarify the following:*
    1. Description of current test including its purpose, limitations (e.g., possibility of false-positive and -negative results and predictive value), and possible outcomes, as well as methods for communicating and maintaining confidentiality of results.
    2. Anticipated use of samples, including whether samples will be used only for the

purpose for which they were collected and then be destroyed.

3. If samples will be retained after initial use, the following issues should be clarified as well:
  - a. The scope of permission to use samples or results in counseling and testing relatives and if so, which relatives.
  - b. The possibility of future test refinements and subjects' expectations that their samples will be analyzed using these new tests and that the results will be communicated to them.
  - c. Permission to use samples from which identifiers have been removed in research, including what type of research.
  - d. Duration of storage of genetic materials, including provision for future access by patients or their designee; the option to have their samples withdrawn or destroyed at any time; and the possibility of inadvertent sample loss.

B. *When obtaining samples for research, clarify the following:*

1. Description of current research: purpose, limitations as above, possible outcomes, and methods for communicating and maintaining confidentiality of results.
2. Possibility that research will lead to the development of diagnostic tests. If so, the possibility their samples will be tested or made available for testing and the results communicated to them must be disclosed, as well as the extent to which subjects can expect to receive any profits from test sales.
3. Permission to use their samples without identifiers for other types of research.
4. Policy for future recontact if permission for future research is not obtained with the sample.
5. Duration of storage of genetic materials and plans for discarding.
6. Note that the regulations on protection of human subjects applicable to institutions receiving federal funds require that the purpose, duration, procedures and alternative procedures, risks and benefits, compensation, voluntary participation and withdrawal, associated additional costs, and communication of results all be described.

II. *Use of Stored DNA or Genetic Materials Previously Collected for Clinical Tests or Research (Office for Protection from Research Risks 1993; Elias and Annas 1994; Clayton et al., in press)*

- A. The following factors, among others, should be considered in deciding whether it is appropriate to use previously collected samples without contacting the individual: are or will the samples be made anonymous?; the degree to which the burden of contacting individuals may make it impracticable to conduct research; existence and content of prior consent; and risks and benefits.
- B. Contacts regarding new diagnostic tests should address permission to use stored samples; purpose, limitations, and possible outcomes of new tests; methods for communicating and maintaining confidentiality of results; permission to use samples or results in testing relatives; and duration of storage.
- C. Contacts regarding new research should address its purpose, limitations and possible outcomes, methods for communicating and maintaining confidentiality of results, duration of storage, uses of samples or results in studying others (anonymously), and sharing samples with other researchers for other types of research.
- D. Finally, in all research, the regulations related to protection of human subjects must be addressed (see IB, above).

**ACMG Storage of Genetic Materials Committee**

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