Department: **UAMS Institutional Review Board**
Policy Number: **19.1**
Section: **Human Genetics Guidance**
Effective Date: **July 31, 2002**
Revision Date: **January 30, 2004; February 15, 2016; August 15, 2022**

SUBJECT: Human Genetics Guidance

POLICY

The UAMS IRB will review human subjects research studies involving genetic research in accordance with federal regulations and institutional policies. Genetic information's unique nature requires the IRB to consider the potential ramifications of current and future identifiability of genetic information and related privacy concerns. Genetic information may also have implications for blood relatives of research subjects.

Funding agencies may also have additional requirements for research involving genetic materials. While the IRB office and reviewers will assist the study teams in meeting funding agency requirements, study teams are ultimately responsible for ensuring funding agency requirements are met.

DEFINITIONS

CLIA Certification: Certification under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) is required of laboratories performing tests on human specimens and returning patient-specific results.
Constitutional (host) genes: Genes common to all normal tissues in the body.
Genome: The entire set of genetic instructions found in a cell.
Genome Wide Association Study (GWAS): A research approach involving rapidly scanning markers across the complete sets of DNA, or genomes, of many people to find genetic variations associated with a particular disease.
Genotype: The genetic constitution of an individual.
Identifiable: data or specimens are identifiable if the identify of the subject to whom they pertain is or may be readily ascertained by the investigator or associated with the data/specimens.
Incidental findings: Previously unknown genetic results discovered unintentionally and that are unrelated to the research question being studied.
Phenotype: The physical manifestation of a gene function.
Whole Exome Sequencing: The identification of exome DNA sequence; an exome contains sequence data for all of the known genes used by the body to make proteins.
Whole Genome Sequencing: The identification of both coding (exome) and non-coding DNA sequence data for an individual.

PROCEDURE

I. Identifiability of Genetic Data

A. Genetic information that does not include direct identifiers, such as the 18 items listed as identifiers under HIPAA, is not currently considered to be identifiable human subject material under the Common Rule. OHRP has indicated, in its preamble to the 2018 Common Rule, that technologies and analytic techniques that may change this identifiability determination are to be periodically reevaluated, with Whole Genome Sequencing expected to be among the first technologies reviewed.
B. Genetic information is considered protected health information under HIPAA.
C. The IRB, in its review of research, will consider whether technology available at the time of its study review or which the study team may use, or the genetic data's combination with other information the study team will access, will require a reconsideration of whether the data/specimens are identifiable.
D. Certain studies requiring sharing of genetic data to central repositories. These centrally submitted data are submitted without any direct identifiers and are considered "de-identified," though they may include a code, the key to which is kept at the originating site. However, the possibility of new techniques allowing the identification of this deidentified data in the future cannot be eliminated. In addition, differing types of genetic analysis may have different levels of identifiability, even in the absence of direct identifiers. The IRB shall therefore individually

consider each study anticipating the use of genetic information and make a determination as to whether the genetic data are identifiable.

II. GINA Protections

A. The IRB shall ensure that protections described in the Genetic Information Nondiscrimination Act of 2008 (GINA) are addressed as appropriate in genetic research.
B. GINA is a federal law that prohibits discrimination in health coverage and employment based on genetic information.
C. GINA defines a genetic test as an analysis of human DNA, RNA, chromosomes, proteins, or metabolites that detect genotypes, mutations, or chromosomal changes. Routine tests that do not detect genotypes, mutations, or chromosomal changes, such as complete blood counts, cholesterol tests, and liver enzyme tests, are not considered genetic tests under GINA. Also, under GINA, genetic tests do not include analyses of proteins or metabolites that are directly related to a manifested disease, disorder, or pathological condition that could reasonably be detected by a health care professional with appropriate training and expertise in the field of medicine involved.
D. The following is suggested language for inclusion in informed consent forms regarding GINA protections. The language may be revised as appropriate to individual studies:
A Federal law, called the Genetic Information Nondiscrimination Act (GINA), generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. This law generally will protect you in the following ways:
    - *Health insurance companies and group health plans may not request your genetic information that we get from this research.*
    - *Health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums.*
    - *Employers with 15 or more employees may not use your genetic information that we get from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment.*

All health insurance companies and group health plans were to follow this law by May 21, 2010. All employers with 15 or more employees were to follow this law as of November 21, 2009. Be aware that this Federal law does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

III. Sharing of genetic findings with research subjects and/or family members

A. IRB submissions should address whether the study will return genetic findings to study participants and/or their family members.
B. Considerations about the return of findings include, but are not limited to, whether:
    1. The genetic tests were done in a CLIA-certified laboratory. If they are done in a non-CLIA-certified lab, their relevance to the subjects' health should be considered.
    2. The findings are actionable, e.g. subjects may wish to discuss the result with their personal health-care team, undergo additional screening or other procedures that might support their health, let their family members know about possibly inherited findings, etc.
    3. Research subjects should be able to opt in or out of receiving genetic findings.
C. Information that has implications for family members requires additional consideration. Any data sharing or recruitment of family members should be proposed such that neither the subject's nor the family members' privacy is violated.

IV. Genetic studies in distinct cultural or social groups

A. If recruitment for a genetic study is to come from a unique cultural or social group, the recruitment and consent process should take into account the risk of undue influence from others in the group regarding joining the study.
B. Investigators shall address, and the IRB will consider, whether the proposed research is appropriate, given the norms of the unique cultural or social group. The IRB reserves the right to ask a consultant familiar with the group's cultural norms to review the research prior to approval.

V. Incidental Findings

A. Submissions shall address whether the proposed genetic analyses have the potential to reveal incidental findings

B. If incidental findings are a possibility, the submission should address whether such findings will be disclosed to subjects, and either the process for disclosing or the rationale for not disclosing.

VI. Risk Considerations for Studies Involving Genetic Research

A. The IRB shall consider the unique risks created by any study-related genetic research along with other study risks when making risk determinations for studies that involve genetic research.
B. The IRB will consider the proposed data storing and sharing methods in light of the potential sensitivity of genetic data.
C. The IRB may also require the PI to seek a Certificate of Confidentiality for genetic research.