

Investigator Guidance Series

University of Utah Institutional Review Board

GENETIC RESEARCH

Description

The IRB reviews genetic research to ensure that all criteria for approval are met, giving additional attention to the following:

- The plan to ensure the privacy and confidentiality of participants.
- The minimizing of risks associated with genetic research.
- The plan to disclose clinically relevant genetic results to the participant.

Privacy and Confidentiality

Issues regarding privacy and confidentiality constitute a large portion of the potential risks in genetic research. Such risks include

- Discovery of non-paternity
- Negative impact on the participants' insurability or employability
- Social discrimination

The investigator must provide a plan to protect privacy and confidentiality in the new study application and the consent document, addressing the following considerations:

- Investigators must establish a method to secure information related to genetic testing in a highly secure and confidential manner, and communicate this method in a manner satisfactory to the IRB and the participant. The investigator must specify who will have access to genetic results, limiting access only to those who will need the results to appropriately conduct the research.
- For studies that may discover non-paternity, a plan must be described for protecting the rights and privacy of the participants and their family members. The investigator may choose to withhold all knowledge of non-paternity from the participant. This should be adequately conveyed in the consent document.
- Genetic results obtained through the study may be required by the IRB to be excluded from the participants' medical records, in order to prevent inadvertent disclosure to insurance companies or employers. If genetic results will be included in the medical record, participants must be informed of this in the consent document.
- Genetic studies which involve the long-term storage of specimens, genetic material, or data in tissue banks must follow the [Tissue Banking Guidelines](#) to ensure confidentiality.
- Investigators may want to consider acquiring a Certificate of Confidentiality, if appropriate (see IRB guidance on [Certificates of Confidentiality](#)).
- Results must be sufficiently anonymized to ensure privacy to participants (and family members) when the time comes for the investigator to publish the results of the study.

Disclosing Results to Participants

Information developed in the course of genetic studies may vary considerably with respect to its impact and value to subjects. Investigators should not disclose genetic results to participants or their physicians if the results are not clinically relevant (i.e., the claimed association between marker or gene and disease is generally accepted by the medical genetics community). The purpose of the study should be conveyed to the participants appropriately in the consent document and genetic results must not be indicated as medically important if the value and reliability of the results has not been established.

If the investigator intends to disclose results to participants, the method should be described in the

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protocol summary and consent document. Written information in conjunction with other methods of disclosure is preferred by the IRB when disclosing results. Keep in mind that when participants are informed about the results of their genetic testing, complete anonymity is virtually impossible to accomplish, and additional protections for privacy and confidentiality may be needed.

The following considerations need to be addressed in the investigator's plan to disclose results to participants:

- The investigator must indicate the individual (including their title and qualifications) who will disclose results.
- The investigator must indicate what method of disclosure (i.e., written, telephone, in person, etc.) will be used.
- The investigator must indicate what provisions have been made to answer questions that may arise as a result of disclosure. The individual who discloses results must have the appropriate background and expertise to discuss the clinical relevance with the participant. There should be an indication of who will respond to such questions, what hours they are available, and how long the services will be available to subjects. In addition, it should be indicated how the services will be paid for.
- Information about results may only be released to others, including the participant's physician, if the participant gives written permission (usually in the consent document). If the results of the genetic testing yield abnormal test results, investigators should consider whether or not they plan to disclose those findings to the participant's primary care physician for clinical use.
- The investigator must indicate if the participants will be given the option to not receive his/her results. The investigation must also describe situations where it would be appropriate to overrule the participants' decision to not receive results.
- Additional considerations must be included when the study includes genetic testing and involves vulnerable populations (e.g. children, persons with impaired decision-making capacity or impaired mental function, etc.).
- Because of the vulnerability of minor subjects, special attention should be paid to whether it is appropriate to disclose genetic information to children. If children are involved, the investigator should indicate who will receive results and at what age subjects may receive their results directly. Justification for disclosure to a child might include age of onset of the condition and whether therapeutic interventions currently are available.
- The investigator must indicate what plans there are for regular post-disclosure contact or follow-up with subjects, if applicable.
- The investigator must indicate what costs, if any, will be borne by the patient in conjunction with disclosure.

Familial Genetic Studies

The design of many genetic studies involves the study of a certain family pedigree or specific social or ethnic group. Recruitment from such a narrow pool of participants may place undue pressure on individuals to participate. Coercion by family members is a potentially more serious problem in genetic studies than in other types of studies. Therefore, familial genetic studies should be designed to minimize this risk so that family members who are not interested in participating are not burdened to do so.

Because federal regulations direct that the IRB evaluate whether the "selection of subjects is

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equitable” and that “an investigator shall seek such consent only under circumstances that provide the prospective subject...sufficient opportunity to consider whether or not to participate and that minimize the possibility of coercion or undue influence” (45 CFR 46.116), investigators should address this issue in the informed consent process and also in the description of how patients are enrolled in the study.

Considerations that must be addressed in the protocol summary and consent document for familial genetic studies include the following:

- The investigator must describe the method of recruitment that ensures the privacy of family members is not violated. For example, will recruitment information be obtained through the clinical medical records of directly from family members? Will the resultant participants be consented separately, or is the permission of the primary participant sufficient?
- The investigator must indicate how family members be protected against disclosure of medical or personal information about themselves to other family members.

Points to Address

Protocol Summary:

1. **Study Procedures:** Please state whether or not the participant will have access to the results of genetic testing.
2. **Study Procedures, Genetic Counseling:** Please describe what provisions have been made to answer questions (PI, genetic counselor, etc.) that may arise as a result of disclosure (e.g., who will respond to such questions, what hours will they be available, long the services will be available to subjects, etc.), and how costs resulting from counseling will be covered.
3. **Administrative Responsibilities, Confidentiality:** Please provide appropriate details regarding how the results of genetic testing will be stored (e.g. in locked cabinets, on password-protected computers, etc.), and state that they will be accessible only to the investigator and other authorized people.

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Consent Document:

- 1. Study Procedures, Disclosure of Results:** Please state whether or not the participant will be given the results of their genetic tests. Also, state who WILL have access to the results of the testing (e.g. the study team, co-investigators, etc.). State who will disclose the results (if applicable) and what method will be used to disclose results (e.g. written notification, telephone contact, etc.).
- 2. Study Procedures, Genetic Counseling:** Please describe what provisions have been made to answer questions (PI, genetic counselor, etc.) that may arise as a result of disclosure (e.g., who will respond to such questions, what hours will they be available, long the services will be available to subjects, etc.).
- 3. Study Procedures, Follow-Up:** Please indicate whether or not there are plans for post-disclosure contact or follow-up with participants, and what risks may be borne by the participants in conjunction with disclosure.
- 4. Risks, Employability and Insurability:** State that the results of genetic testing will NOT be made available to employers, insurance companies, etc., **OR** please disclose the possibility of social risks by including the following disclosure, "Social risks related to genetic testing include the possibility of employment or insurance discrimination. It includes the possibility of increased cost of your current insurance and/or the inability to obtain insurance in the future. Insurance companies may ask you about medical tests you have had if you are applying for a new policy. Insurance companies may be able to get genetic information about you if you give it to your doctor. We make every effort to protect your data, and to the best of our ability we keep all results as part of this research study out of your medical record. There is a small chance that your information would unintentionally end up in your medical record. If clinical results (procedures or genetic tests) are put into your medical record and are obtained by employers and/or insurance providers, employment or insurance discrimination may occur. If it is important for your medical care for us to reveal specific findings to your personal physician, this can be done at your request. However, please remember that you may have given insurance companies permission to access your physician's records."
- 5. Confidentiality:** Please state that how results of genetic testing will be stored (e.g. in locked cabinets, on password-protected computers, etc.), and state that they will be accessible only to the investigator and other authorized people.
- 6. Costs and Compensation:** Please indicate how genetic counseling services will be paid for (if applicable).

References & Links

*National Society of
Genetic Counselors*

<http://www.nsgc.org/>

*DHHS Statement of
Genetic Testing*

<http://www.hhs.gov/asl/testify/t990421c.html>

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