**GUIDANCE 12**

**HUMAN GENETIC RESEARCH GUIDANCE**

1. **Genetic research**

Genetic research on human subjects can raise a number of distinctive problems. One common problem is that researchers (and sometimes experimental subjects) may acquire genetic information that also pertains to relatives who have not consented to any investigation and need not be made aware of its results.

1. **Applicability**

Rowan University defines genetic research as research that involves the analysis of DNA, RNA, chromosomes, proteins, or certain metabolites which might act as or identify markers associated with a known or suspected predisposition to disease or behavior. Usually genetic research involves the collection of human biological material such as blood, skin or other tissues, nail clippings or hair. Genetic research also may include the construction of pedigrees (maps of the distribution of a particular trait or condition among related individuals or family medical histories). Although gene transfer is another form of genetic research, this guidance document does not apply to gene transfer research.

1. **IRB requirements**

Federal guidelines strongly advise IRBs to consider specific issues when reviewing clinical genetic research and to alert investigators engaged in such research to address these issues in their application for IRB approval. Unlike the risks presented by biomedical research, the primary risks of genetic research are risks of social and psychological harm rather than risks of physical injury. Genetic studies that generate information about subjects' personal health risks can provoke anxiety and confusion, damage familial relationships, and compromise subjects' insurability and employment opportunities. Although these genetic studies may be limited to a collection of family histories or blood draws, the IRB does not necessarily consider them to be minimal risk.

1. **What qualifies as genetic research?**

Genetic research does not mean only research that involves looking for mutations in DNA. Research that involves looking at the differences between proteins in individuals with or without a certain disease can also qualify as genetic research. Records research involving information that was derived from a previous genetic test can also qualify as genetic research. See definitions below.

1. **Genetic research:** Research using human DNA samples, genetic testing or genetic information.
2. **Genetic information:** Information about an individual or the individual's blood relatives obtained from a genetic test.
3. **Genetic test:** A test for determining the presence or absence of genetic characteristics in a human individual or the individual's blood relatives, including tests of nucleic acids, such as DNA, RNA, and mitochondrial DNA, chromosomes or proteins in order to diagnose or determine a genetic characteristic.
4. **Genetic characteristic:** A gene, chromosome or alteration thereof that may be tested to determine the existence of or risk for acquiring a disease, disorder, trait, propensity or syndrome, or to identify an individual or a blood relative. "Genetic characteristic" does not include family history or a genetically transmitted characteristic whose existence or identity is determined by means other than through a genetic test. IRB approval is required before disclosure of research results can occur. In the event these conditions are met, the results may only be released to the subject or any other party with the subject's permission, and appropriate medical advice and referral must be provided.
5. **Disclosure of research results to subjects**

Disclosure of genetic research findings to a research subject or the subject's physician through use of personal identifiers should not occur unless:

1. The research findings are scientifically valid and confirmed (done in a CLIA approved lab);
2. The findings have significant implications for the subject's or the public's health; and
3. A course of action to ameliorate or treat the subject's or the public's health concerns is readily available.
4. **Re-contact**

Re-contact of a research subject or a patient from whom samples or information was obtained originally for clinical purposes should not occur unless the subject was informed during the initial treatment or research consent and authorization process, that re-contact may occur under specified circumstances. Reasons for re-contacting research subjects can include re-contact for release of clinically relevant research results. If this is desired, precautions must be taken both to minimize the potential harm to subjects of receiving bad news and to guard against the unintended release of the information. The precautions needed in conveying genetic research results depend on the age at onset of the disorder, the burden of illness, and the availability of treatment or prevention. The communication of genetic information carries with it the responsibility to interpret the results and provide care for the individual; and, thus, it is ideally done in the setting of a clinical rather than research relationship with the subject. Because of the complexity of the results of most genetic tests, subjects cannot be required to inform relatives of the results of the research**.**

1. **FAQs**

Please go to the following link to learn more about genetic research: <https://www.genome.gov/For-Patients-and-Families/Health-FAQ>.