GWAS - Submission of De-Identified Specimens and Data to NIH Repositories for GenomeWide Association Studies (GWAS)

Modified: January 2017

Background:

Q: What is the National Institutes of Health (NIH) GWAS Policy?

This is a data sharing policy implemented in 2008 for NIH-funded Genome-Wide Association studies, or GWAS. (See GWAS Policy). The policy establishes a NIH Database of Genotypes and Phenotypes (dbGAP), which has two components.

• An open access portion that will be freely available to the public and will include:
  o The protocol;
  o Questionnaires;
  o Variables measured; and,
  o Other supporting documentation
• A controlled access portion that will only be available to researchers who have been approved by an NIH Data Access Committee (DAC). This portion will include coded phenotype, exposure, genotype and pedigree data, and summary statistics.

Q: What is a Genome Wide Association Study?

• A GWAS study is defined in the NIH policy as “any study of genetic variation across the entire human genome that is designed to identify genetic associations with observable traits or the presence of a disease or condition.”
• A GWAS study is typically one in which 100,000 or more SNP markers are tested in individual DNA samples, to produce a “high density” genomic profile.

Q: When did the policy go into effect?

The GWAS Policy went into effect for competing funding applications and new funding proposals submitted to NIH on or after January 25, 2008.

Q: To whom does it apply?

The policy applies to investigators who receive any NIH funding for genome-wide analysis of specimens. Of note, unlike the general NIH data sharing policy that has a threshold of $500,000, the GWAS policy has no threshold; therefore, any NIH funding for GWAS triggers requirements under the policy.

Q: What is needed from the IRB in order for a local investigator to apply for an NIH grant that involves GWAS?
NIH requires a “Certification for Data Submission” letter. This is a template letter that indicates the Institution is certifying that the research will be conducted according to the Common Rule, and that resultant data will be shared. To obtain this letter, the investigator must complete and submit a paper application “GWAS Data Sharing Plan” located on the IRB website. Assuming it is approved, the following will occur:

- The IRB will place the study information on the template letter.
- The template letter will be sent to the VP for Research, who places on his/her letterhead, and signs it.
- It is then scanned, and sent to the IRB Chairperson; who will also sign.
- It is then scanned and sent to the PI, who signs it.
- The letter can then accompany the NIH grant submission.

Assuming the grant is awarded, when the investigator submits the protocol to the IRB via myIRB, the signed letter must be uploaded in the miscellaneous section of the submission.

Q: Does a study subject have the option to agree to the study, but not have their data shared with NIH?

Yes. To that end, all UF GWAS studies must include a GWAS consent addendum which explains the risks to the subject when their data is shared with NIH. Subjects have to agree to the sharing of their data, and investigators must develop a process to keep track of this consent when it is time to share the data with NIH.

Q: What if the IRB determines that the original consent forms are not consistent with the requirements for sharing, that no research consent forms were signed, or the risk of re-identification seems too high?

If the research consent was not consistent with GWAS sharing requirements or there are no research consent forms (e.g., samples were obtained under a surgical consent or waiver of consent) subjects must be contacted and consented before the PI may submit their data to the NIH GWAS repository. The IRB will allow the use of the GWAS consent addendum to be used to re-contact subjects to seek their re-consent.

If the consent forms impose use restrictions that are inconsistent with broad data sharing, the IRB may impose use restrictions upon the data submitted to the NIH, (e.g., a restriction to specify that the data may be disclosed by NIH only for the study of a particular disease or only for non-commercial research).

To reduce the risk of re-identification, the IRB may limit the types or fields of phenotype data that the PI may submit to the GWAS repository.