PARTNERS HUMAN RESEARCH COMMITTEE
POLICIES AND PROCEDURES

Summary of Partners Implementation of the NIH GWAS
(Genome-Wide Association Study) Policy

This policy is applicable if:

- You have obtained NIH funds on or after January 25, 2008 to conduct genome wide analysis studies (GWAS);
- You plan to voluntarily submit any genotype/phenotype data to the NIH GWAS Registry, or
- You plan to access data from the NIH GWAS Repository.

1.0 GENERAL INFORMATION

What is the NIH GWAS Policy?
This is a data sharing policy for Genome Wide Association studies or GWAS.
The policy establishes a NIH GWAS Repository that has two components:

- An open access portion that will be freely available to the public and will include:
  - The protocol
  - Questionnaires
  - Variables measured, and
  - 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

What is a Genome Wide Association Study?
A genome-wide association study is one in which 100,000 or more SNP markers are tested in individual DNA samples.

NIH also offers the following definition:

> "To meet the definition of a GWAS, the density of genetic markers and the extent of linkage disequilibrium should be sufficient to capture (by the r2 parameter) a large proportion of the common variation in the genome of the population under study, and the number of samples (in a case-control or trio design) should provide sufficient power to detect variants of modest effect."

When did the policy go into effect?
The GWAS Policy went into effect for competing applications and new proposals submitted on or after January 25, 2008.

To whom does it apply?
The policy applies to investigators who receive any NIH money for genome-wide analysis of tissue specimens (GWAS). Of note, unlike the NIH data sharing policy that has a threshold of $500,000, the GWAS policy has no threshold – therefore, any NIH funding for GWAS triggers the policy.

When is the policy NOT triggered?
The policy is not triggered if:

GWAS conducted without NIH funds

- Even if the GWAS use specimens that were collected and/or stored with NIH money

2.0 DATA SUBMISSIONS TO THE GWAS REPOSITORY

Is it possible to voluntarily submit data to the NIH GWAS Repository?
Yes.
The NIH is hoping that even when the policy is not triggered, investigators will voluntarily share their data. If an investigator voluntarily submits data to GWAS – the submission must be in compliance with the GWAS Policy.

How do you submit materials for the open-access portion of the Repository?
Unless otherwise described in the grant announcement, these materials (The protocol, questionnaires, variables measured, and other supporting documentation) should be submitted at the same time the genotype/phenotype information is submitted to the controlled access portion. Specific instructions will be part of the grant instructions.

Who is responsible for submitting data to the NIH GWAS Repository?
The person/entity who is receiving the NIH funds for the GWAS analyses is responsible for submitting the data.

N.b., For multi-institution research, the single submitting person/center is responsible for ascertaining the compliance of all centers.

How do you submit coded genotype-phenotype data to the NIH GWAS Repository?
Each submission must include a letter of certification signed by the Institutional Official. This letter certifies that the IRB has reviewed and verified the following:

- That the submission of the data is consistent with the informed consent form of study participants from whom the data was initially obtained
- That the de-identification of the data sets is consistent with the GWAS standards (HIPAA standards)
- That the risks to individuals, their families and groups or populations associated with the data submitted has been considered and
- That the genotype and phenotype data to be submitted were collected in a manner consistent with the Common Rule (federal law re: human research protections)

Therefore, the submitting investigator must provide the following documents to the IRB:

- Description of what genotype/phenotype data is being submitted to the NIH GWAS Repository
- Copy of the consent form/s used to collect the initial data/samples
    - N.b., this must include forms from all sites
- Description of the method/s used for coding the data

- Description of how the link will be maintained with assertion that it will never be shared with the NIH.

Is there a specific 'certification' form that you should use for submission of genotype/phenotype data into the NIH GWAS Repository?
Yes.
Partners has developed language for a certification letter to be signed by the appropriate Institutional Official (BWH – Barbara Bierer; MGH – Richard Bringhurst; McLean – Scott Rausch or their designees). The IRB will forward the certification form to the appropriate Institutional Official after appropriate information has been reviewed.

What happens if the IRB determines that the initial consent forms are not consistent with submission of data to the NIH GWAS Repository?
The data cannot be submitted.
The only way to submit the data would be if study participants were re-contacted and re-consented.
Re-consent requires submission of an amendment to the IRB. The IRB would have to consider whether or not re-contact and re-consent is feasible and/or appropriate.
NIH has said that investigators can request funds to cover the expense of re-consenting.

What if study participants cannot be re-contacted for a new consent?
At this time, the NIH will not accept data without an IRB approved consent form.

What if the initial consent form included language consistent with sharing genetic data with the NIH, but limited the research to a single disease/condition?
The IRB can determine that the data can be submitted – but that the NIH GWAS Repository can only make the data available for that restricted use.

What if the original data and tissue were collected under a waiver of consent?
At this time, the NIH will not accept data that was obtained without informed consent and a form documenting that consent.

3.0 OBTAINING DATA FROM THE OPEN- AND CONTROLLED-ACCESS PORTIONS OF THE REPOSITORY

How can you access data from the NIH GWAS Repository?
Information in the open access portion of the Repository can be accessed from any computer.
Remember that this includes general information and does NOT include any genotype/phenotype data.

Information in the controlled access portion of the Repository requires submission of a data request to the GWAS Repository and approval by a Data Access Committee (DAC).
The request must include a certification signed by your Institutional Official that includes a number of conditions.
In general, these conditions address:

- That the data will only be used for the approved research
- Data confidentiality and security protections are in place
- There will be no attempt to identify any individual

- No data will be sold
- You will not share the data with anyone not listed on the data request
- You will post a summary of the research on the NIH GWAS site
- You will provide annual reports on research using GWAS data set/s
- You will acknowledge the NIH GWAS Repository on any publications or IP requests

The NIH will provide specific forms that must be submitted to the Data Access Committee.

Do you need IRB review for accessing data from the NIH GWAS Repository?
Generally no
The NIH Data Access Committees (DACs), that must approve access to data in the Repository, may in some uncommon situations require local IRB review.