FACT SHEET

NATIONAL INSTITUTES OF HEALTH

Genome - Wide Association Studies (GWAS) Policy

What is GWAS?

A genome-wide association study (GWAS) is an approach used in genetics research to associate specific genetic variations with particular diseases. The method involves examining genetic variations (genotypes) across the complete sequences of DNA, or genomes, of many different people to find genetic variants associated with a disease or trait (phenotypes). Researchers can use the information to better understand how genetic variation affects the normal function of genes, in addition to helping develop better prevention and treatment strategies.

Why A NIH GWAS Repository?

- **Improve Health** – Sharing genome-based research, including both genotypic and phenotypic information, with a broad number of scientists, will enable medical science to better understand the health needs of the public and facilitate development of new technologies and approaches for the prevention, diagnosis and treatment of disease.
- **Maximize Public Investment** – Providing centralized access significantly increases the availability of data for researchers, which is predicted to accelerate the discovery of associations between genetic data and disease while reducing research costs.

About the Policy

The GWAS policy applies to investigators who are supported by NIH funding to perform genome-wide association studies.

Datasets from GWAS are stored in the database of Genotypes and Phenotypes (dbGaP) (http://www.ncbi.nlm.nih.gov/gap). dbGaP has two levels of access, open –(public) for summary information and controlled for individual information. Investigators submitting data to dbGaP provide a description of their study, which will be viewable to the public. The data submitted must be de-identified to protect the privacy of study participants according to criteria defined in the HIPAA Privacy Rule.

Publication Embargo

Any investigator contributing data to dbGaP has the exclusive right to publish analyses of that data up to 12 months after the data is released in dbGaP. Approved secondary users may access the data before this period expires but cannot make their analyses available to the public in any form until the period of exclusivity expires. The embargo release date is listed next to each study on dbGaP.

Intellectual Property

The NIH encourages patents for downstream discoveries that would be necessary to develop products to meet public health needs, while discouraging obtaining patents for early, pre-competitive information that may impede future research.

Based on the primary nature of GWAS, nearly all of the data within dbGaP could be considered pre-competitive information.

Governance

A governance system is in place to oversee the implementation of the GWAS policy. Data Access Committees (DACs) review project proposals submitted by secondary investigators. If issues arise, the DACs can consult with the Participant Protection & Data Management Steering Committee (PPDM). The PPDM also reviews policies and procedures for the implementation of the GWAS policy and advises and makes recommendations to the Senior Oversight Committee (SOC).

The Technical Standards Steering Committee (TSSC) ensures the rigorous standards for the GWAS data repository and the GWAS data. The TSSC provides analyses and makes recommendations to the SOC on policy and technical issues. The SOC makes both GWAS policy decisions and recommendations to the NIH Director.

TO READ MORE ABOUT GENOME-WIDE ASSOCIATION STUDIES:

http://gwas.nih.gov/index.html

http://grants.nih.gov/grants/guide/notice-files/NOT-OD-07-088.html

http://www.ncbi.nlm.nih.gov/gap

COMMENTS

Comments and questions about the policy should be sent to gwas@mail.nih.gov.