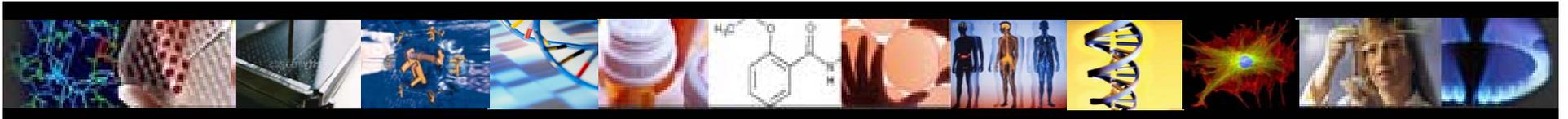


SACGHS

Proposed Policy for Sharing of Data Obtained in NIH Supported or Conducted Genome-Wide Association Studies (GWAS)



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What are GWAS?

- **A genome-wide association study (GWAS) is defined as any study of genetic variation across the entire human genome that is designed to identify genetic associations with observable traits (such as blood pressure or weight), or the presence or absence of a disease or condition.**

Sharing of Data

- **NIH is the steward of the American public's investment in global health.**
- **Information which is not shared represents lost opportunity to improve the health of the public – including negative studies.**
- **NIH has been encouraging wide sharing of information, including of published papers, for several years.**

The Information Glut

- **Scientists are now generating far more data than they can analyze.**
- **Genetic data, especially related to association with disease states, represents an exquisitely valuable resource.**
- **The participants in such research deserve to have their privacy protected, and their contribution maximized.**

NIH Policy

- **The cost of extensive genotyping has fallen rapidly, and continues to fall, making studies feasible which would not have been possible even 4 years ago.**
- **NIH is receiving many applications for GWAS, representing many millions of dollars of research investment.**
- **NIH has required submission of data sharing plans as part of all large grant applications for several years.**

Guiding Principle:

The greatest public benefit will be realized if data from GWAS are made available, under terms and conditions consistent with the informed consent provided by individual participants, in a timely manner to the largest possible number of investigators.

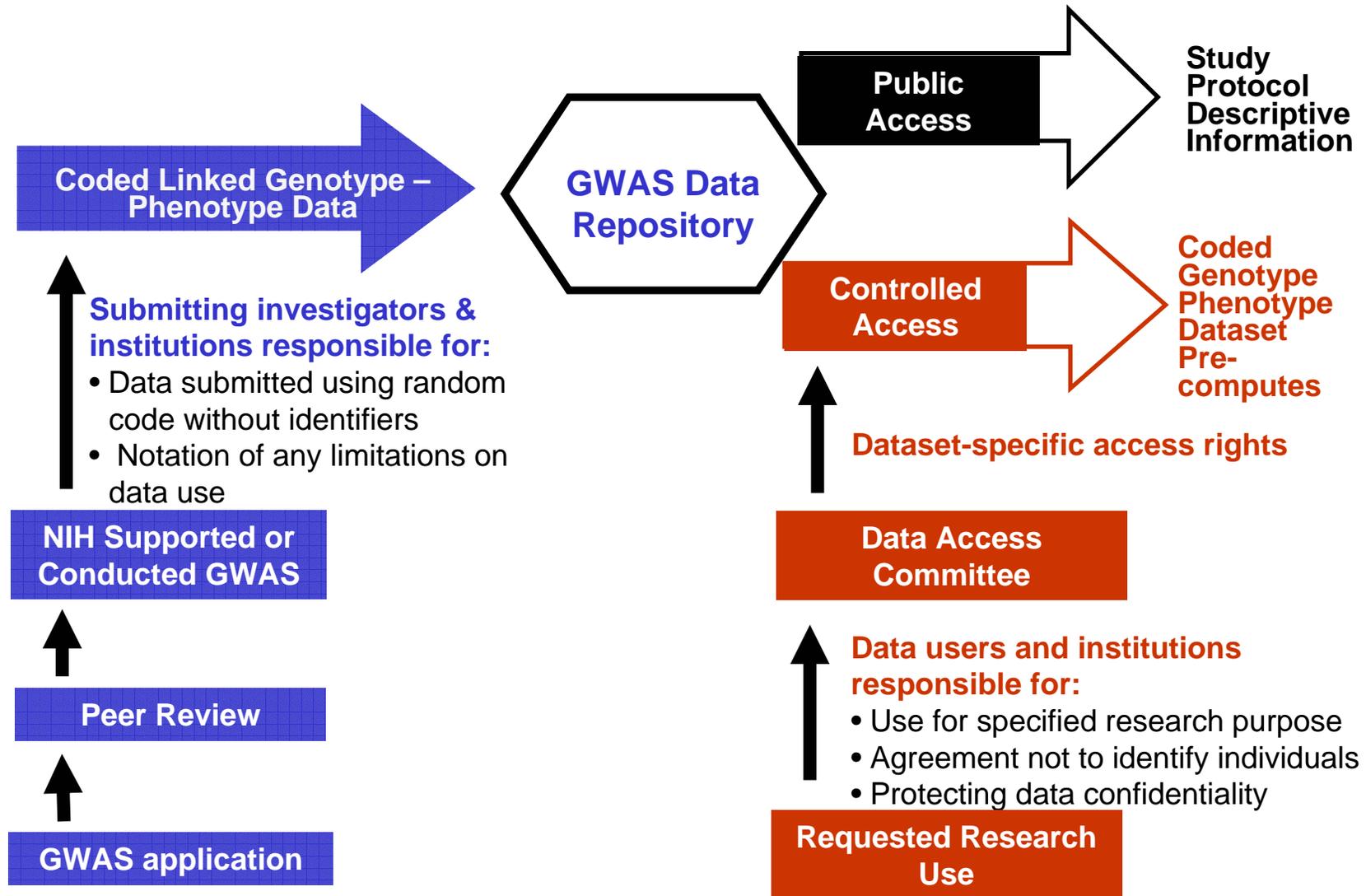
Elements of Proposed Policy

- **Data Management** – outlines expectations for the submission of data to the proposed GWAS data repository and the processes for secondary investigators to access the data from NIH;
- **Publication** – defines a period of publication exclusivity for investigators submitting data;
- **Intellectual Property** – encourages patenting approaches that enable both research and the downstream discoveries necessary to develop public health related products.

Benefits of the Proposed GWAS Data Repository

- **Improve Health:** Better understand the health needs of the public and facilitate the development of improved diagnostic tools and design of new, safe and highly effective treatments.
- **Maximize Public Investment:** Accelerate the discovery of associations between genetic data and disease, while minimizing research costs.

NIH GWAS Research Overview



Expectations of Investigators

- **Investigators who submit GWAS data provide:**
 - Description of studies
 - Coded genotypic and phenotypic data
 - Certification of IRB approval for submission
 - Assurance of compliance with applicable laws
- **Investigators who request GWAS data provide:**
 - Description of proposed research projects
 - Data Use Certification agreeing to defined period during which manuscripts will not be submitted for publication
 - Agreement to protect confidentiality of data
 - Annual progress reports

Is Secondary Use Human Subjects Research?

- **Removed from original participants.**
- **Stripped of identifiers.**
- **By current definitions, not HSR, per OHRP.**
- **Because it is genetic data, it is potentially identifiable. With current technology, this is difficult and requires a comparison sample.**
- **Extremely sensitive and complex, requires oversight and management.**

Issues for Participants

- **Consent for sharing: prospective, retrospective**
- **Security of data**
- **Inherent risks**
 - **Identifiability**
 - **Genetic discrimination**
 - **Family and community implications**

Protection of Data

- **Coded submission**

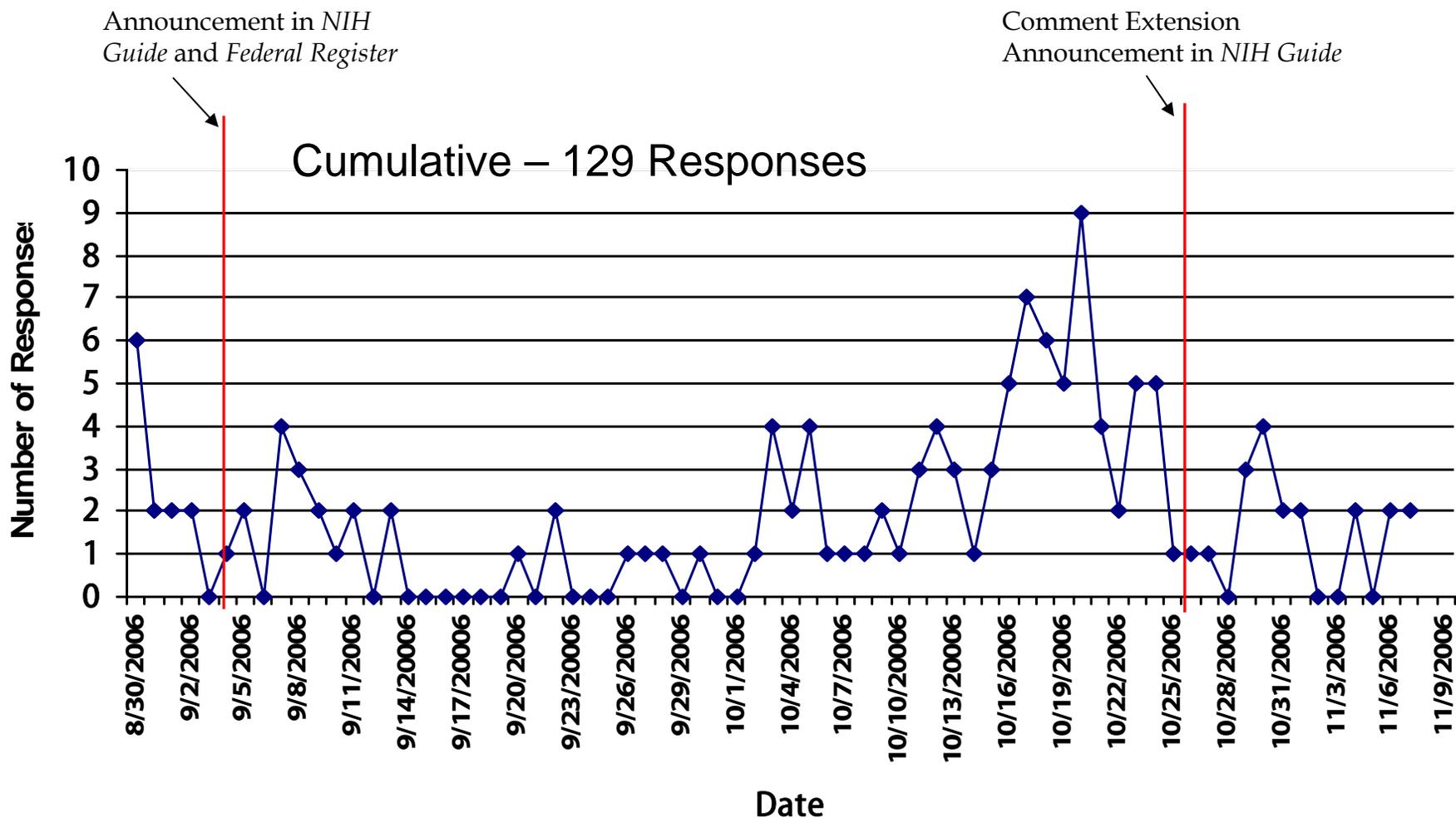
- Long-term responsibility for the code
- Updating, return of results

- **Some will be de-identified**

- **Computer security**

- Access
- Downloading data versus working on central server

GWAS Number of Responses Received Daily August 30 – November 9, 2006



This represents all the comments received during the commenting period but we are still verifying through our quality control process that there are no duplicate records, and that all the responses mailed, e-mailed or faxed have been entered into the database.

Questions for Public Consultation

1. What are **potential benefits and risks to research participants** of sharing phenotypic and genotypic data without information that would identify research participants?
2. Are **additional protections needed** to minimize risks to research participants whose personal identifying information has been removed?
3. What are the **advantages and disadvantages** of:
 - a. A centralized NIH data repository?
 - b. The proposed approach to data submission?
 - c. A grace period for scientific publication?
 - d. The approach to intellectual property?
4. What **specific resources may investigators and institutions need** to meet the goals of this proposed policy?

NIH Public Consultation on Sharing Genetic Data

Genome-Wide Association Studies (GWAS)

- [OER Home](#)
- [Funding Opportunities](#)
- [Applications & Forms](#)
- [Awarded Grants](#)
- [Grants Policy](#)
- [eRA](#)
- [About OER](#)

The NIH is interested in advancing Genome-Wide Association Studies (GWAS) to identify common genetic factors that influence health and disease because the information derived from such studies will be essential for developing new approaches to reduce disease burden and promote health. GWAS are currently being conducted to identify regions of genetic variation across the entire human genome that is designed to identify genetic associations with observable traits (such as blood pressure or weight) and susceptibility to a disease or condition. The goal of the proposed policy is to advance science for the benefit of the public through the creation of a centralized NIH GWAS data repository. This Website is to support the public consultation process to inform policy development activities.

The "Overview" section of this site presents the essential background and responses to frequently asked questions. The remaining sections on this page focus on the notices released to date which will result in a request for information through which the National Institutes of Health will receive comments and responses. For more information, please see the "Notices and Announcements" section.

Overview

- [Background](#)

Notices and Announcements

- [Submit a Comment](#) - Comments may be submitted through this website until 05/15/2006.
- [NIH Press Release](#) (08/30/2006) - NIH Establishes a Centralized Repository for Genetic Information
- [Federal Register Notice](#) (08/30/2006) - Request for Information (RFI): Proposed Policy for Sharing of Data Obtained in NIH Supported or Conducted Genome-Wide Association Studies (GWAS)
- [NIH Guide Notice NOT-OD-06-09](#) (08/30/2006) - Request for Information (RFI): Proposed Policy for Sharing of Data obtained in NIH supported or conducted Genome-Wide Association Studies (GWAS)
- [NIH Guide Notice NOT-OD-06-071](#) (05/15/2006) - Notice to Applicants for NIH Genome-Wide Association Studies

Comments or Questions?

- Please send email to GWAS@nih.gov.

Google: "GWAS policy"

For More Information

- <http://grants.nih.gov/grants/gwas/index.htm>
- **GWAS@nih.gov**