National Human Genome Research Institute

Genomic Data and the NIH Genomic Data Sharing Policy

Laura Lyman Rodriguez, Ph.D.
Division of Policy, Communications, and Education
National Human Genome Research Institute

NIH Tribal Advisory Committee Meeting
September 29, 2015

Values Intrinsic to NIH

- Stimulate research to improve human health
- Respect research participant interests
- Promote (maximize) public benefit
- Responsible stewardship of public investment (not just the dollars)

Features of Genetic/Genomic Data

- Stability
- Unique
- Probabilistic and complex information
- Familial implications
- Effect on reproductive decision-making
- Group/community implications
- Special cultural meaning

What Is Different with Genome Sequence?

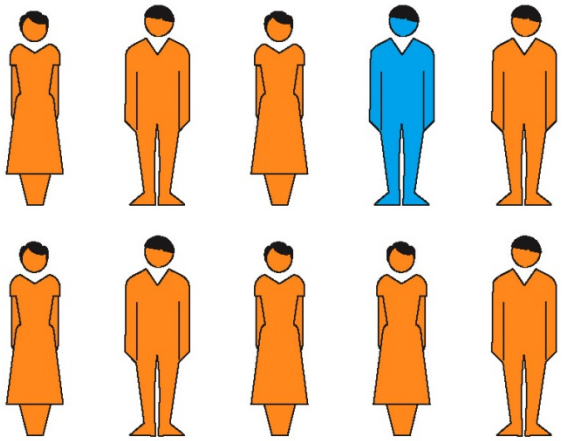
- Not the issues per se
  - — Privacy
  - — Potentially sensitive data
  - — Uncertain risks
  - — Incidental findings
  - — Relevance to relatives, groups, communities
  - — Complex and rapidly evolving science
- Issues/risks more “concrete”
- Potential for benefit (generalized) coming into focus

Variant A

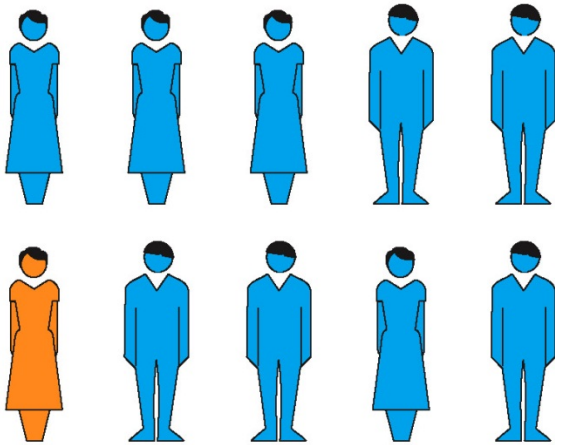
Affected

Unaffected

Variant B



Affected



Unaffected

Alzheimer's

Cancer

Aging

Autism

Drug Response

Diabetes

Challenges to Address

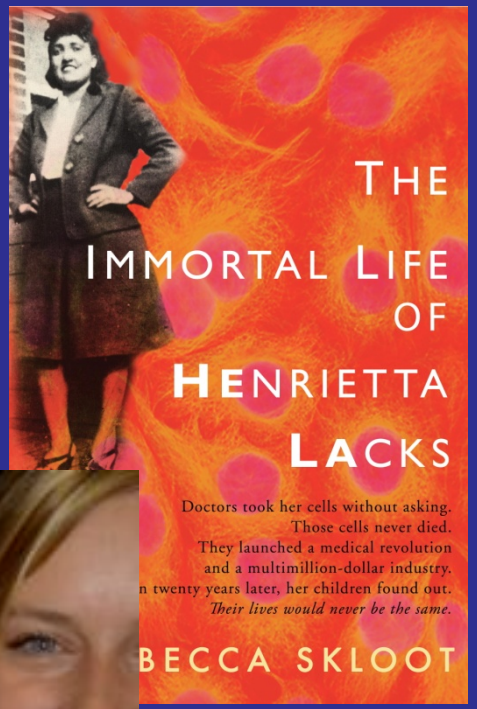
- 40% of population of non-European descent, but 96% studies conducted with European populations
- Recruitment of participants from underserved populations can be harder
- Approaches will need to reach beyond genomics

Addressing Our History….

Indian Tribe Wins Fight to Limit Research of Its DNA

Edmond Tilousi, 56, who can climb the eight miles to the rim of the Grand

By AMY HARMON
Published: April 21, 2010



Informed Consent

- Proposes new requirement for informed consent for the use of biospecimens and information sharing in research
  - Not related to “identifiability” but autonomy of participan
- Broad consent using to-be-developed HHS template
- On-going data collection after 10 years would require new consent

Informed Consent

Comment Period through 12/7/15

http://www.hhs.gov/ohrp/humansubjects/regulations/nprmhome.html

Understanding & Working with Communities

NHGRI or ity

Outreach en mics

Putting the Pieces Together

- **Scientific Design**
  - Research aims and objectives
  - Program priorities
- **Policies and Procedures**
  - Ethical principles and values
  - Applicable laws and regulations
- **Governance & Oversight**
  - Shared responsibilities
  - Accountability
  - Transparency & Trust

The NIH Genomic Data Sharing Policy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NEWS & EVENTS

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NIH News in Health
NIH Research Matters
NIH Record

For Immediate Release: Wednesday, August 27, 2014, 1:00 p.m. EDT

NIH issues finalized policy on genomic data sharing

Policy's Implementation is key to accelerating biomedical discoveries

Institute/Center
NIH Office of the Director (OD)

Contact
NIH Office of Communications
301-496-5787

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The National Institutes of Health has issued a final NIH Genomic Data Sharing (GDS) policy to promote data sharing as a way to speed the translation of data into knowledge, products and procedures that improve health while protecting the privacy of research participants. The final policy was posted in the Federal Register Aug. 26, 2014 and published in the NIH Guide for Grants and Contracts Aug. 27, 2014.

Starting with funding applications submitted for a Jan. 25, 2015, receipt date, the policy will apply to all NIH-funded, large-scale human and non-human projects that generate genomic data. This includes research conducted with the support of NIH grants and contracts and within the NIH Intramural Research Program. NIH officials finalized the policy after reviewing public comments on a draft released in September 2013.

Genomic Data Sharing Policy

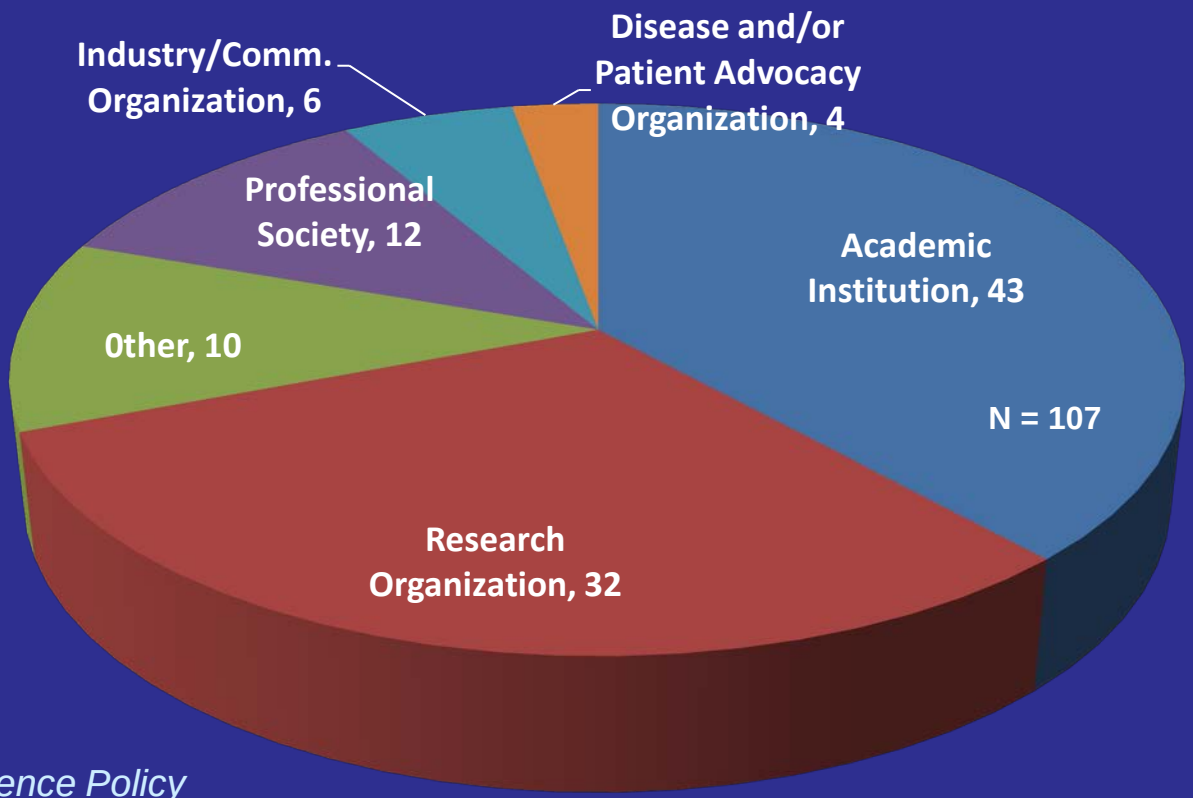
The GDS policy can be traced to the Human Genome Project, completed in 2003, which required rapid and broad data release during its mapping and sequencing of the human genome. The GDS policy is an extension of and replaces the Genome-Wide Association Studies (GWAS) data sharing policy. Since 2007, the GWAS policy has governed biomedical researchers' submission and access to human data through the NIH database for Genotypes and Phenotypes (dbGaP). Its two-tiered data distribution system has made some information and data available to the public without restrictions. Access to other data has been controlled and made available only for research purposes consistent with the consent provided by participants in the original study.

Announced: August 27, 2014
Effective: January 25, 2015

- Establish community resource to promote maximum public benefit
  - Rapid and broad data sharing for all human genomic data
  - Consistent participant protections and data sharing expectations beyond GWAS
- Establish overarching framework
  - Build on GWAS Policy to bring human and non-human genomic data under a single policy
  - Enable more rapid and efficient revision/updating process

Public Comment on GDS

- 60 day comment period including “Town Hall” webinar
- 107 public comments received
- 575 specific points made
- All support general principle of data sharing
- Nearly all general support for policy



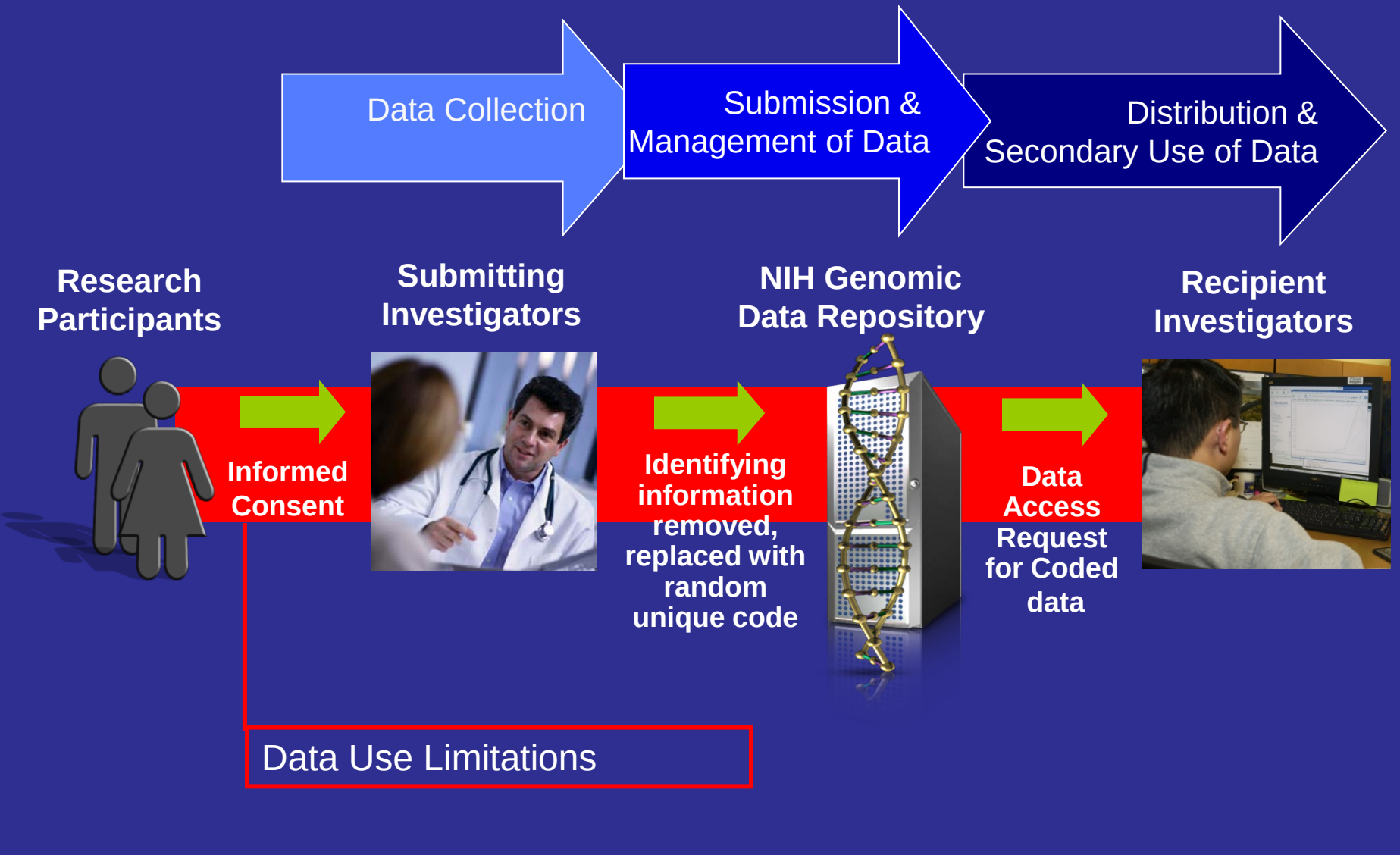
Analysis conducted by NIH Office of Science Policy

Guiding Principle for Genomic Data Sharing

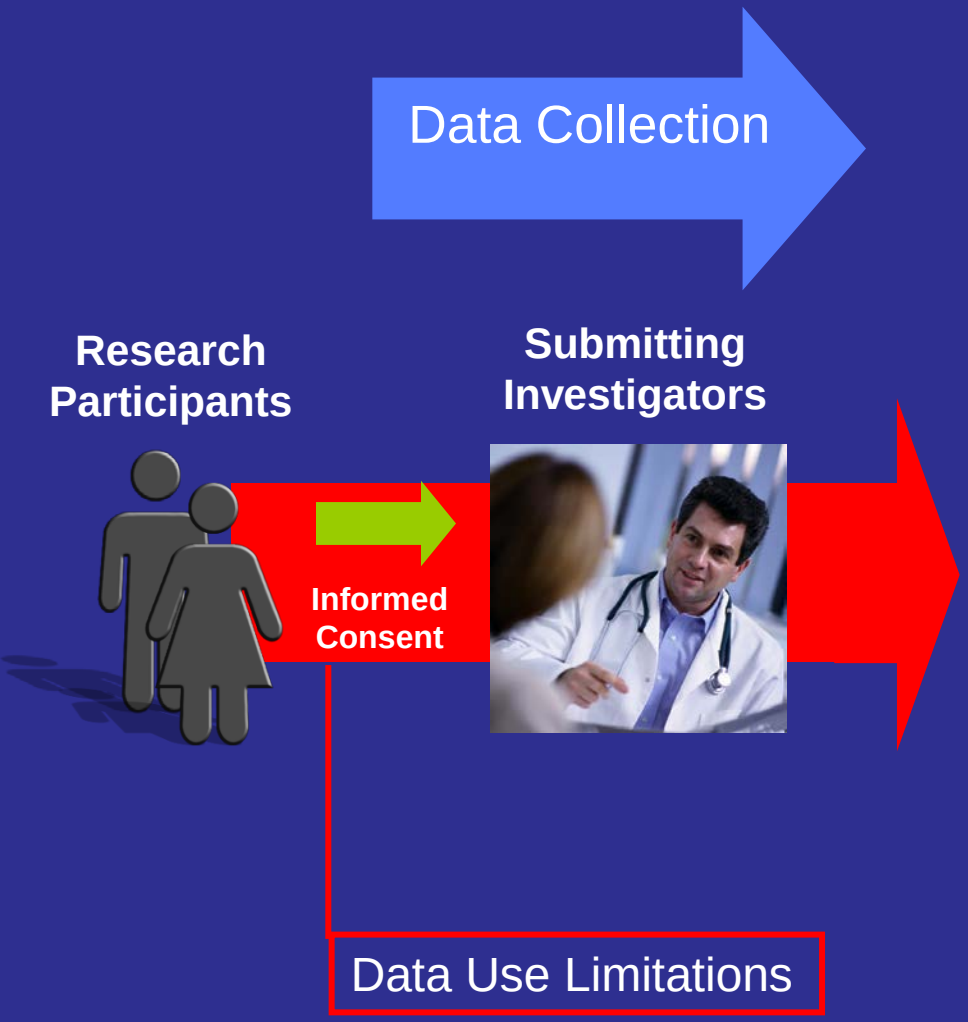
The greatest public benefit will be realized if data from genomic studies are made available, under terms and conditions consistent with the informed consent provided by individual participants, in a timely manner to the largest possible number of investigators.

- Respect for Participants
- Data Sharing
- Freedom to Operate

Genomic Data Management Overview



Informed Consent

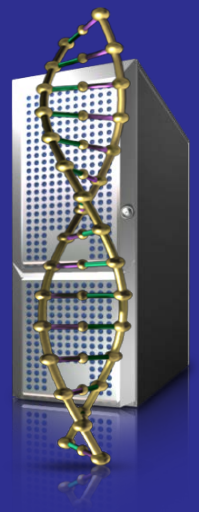


- Original standard for data submission: data use “not inconsistent with consent”
- GDS Policy has explicit standard of consent for research purposes and broad sharing (note access)
- Grandfather clause
- Allows for exceptions for compelling scientific circumstances

Data Access is Two-Tiered

NIH-Designated Genomic Data Repository

Genomic & Phenotype Data



Open Access

Study Protocol
Descriptive Information

Controlled Access

Coded Genotypes
Phenotypes
Pre-computes

Approve based upon Data Use Limitations

Data Access Request

Co-signed by institution
Agree to terms of use
PI agrees to Code of Conduct

Data Access Committee

Institutional Certification

- The Institutional Certification assures that
  - The data submission is consistent with applicable national, tribal, and state laws and regulations
  - Any limitations on the research use of the data are delineated (“Data Use Limitations”)
  - The identities of research participants will not be disclosed to NIH

Institutional Certification (continued)

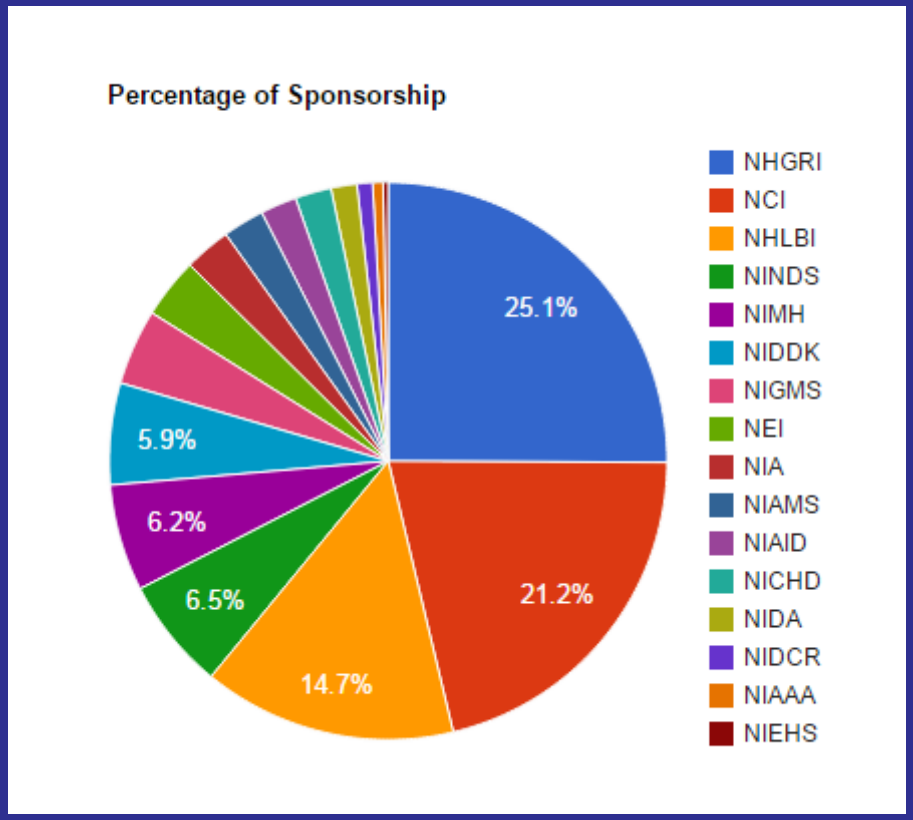
- The IRB has certified that:
  - The protocol for collection of genomic and phenotypic data is consistent with regulations
  - Data submission and sharing are consistent with informed consent
  - Considered risks to individuals and their families
  - Considered risks to groups or populations
  - The plan for de-identifying data is consistent with the Policy

Exceptions to Data Deposition

- Policy notes that there will be cases where data deposition may not be appropriate
- Institution requests exceptions within the application's Data Sharing Plan
- To date exceptions granted due to:
  - Limited consent
  - Legal restrictions
  - Localized geographic representation

Data Submission, Access, and Use Statistics

NIH ICs Sponsoring dbGaP Studies (currently 607)



Data Access and Use Snapshot
(since 2007)

32962= Requests submitted

21973= Requests approved

1200+ = Secondary use research publications

3843 = PIs requesting data

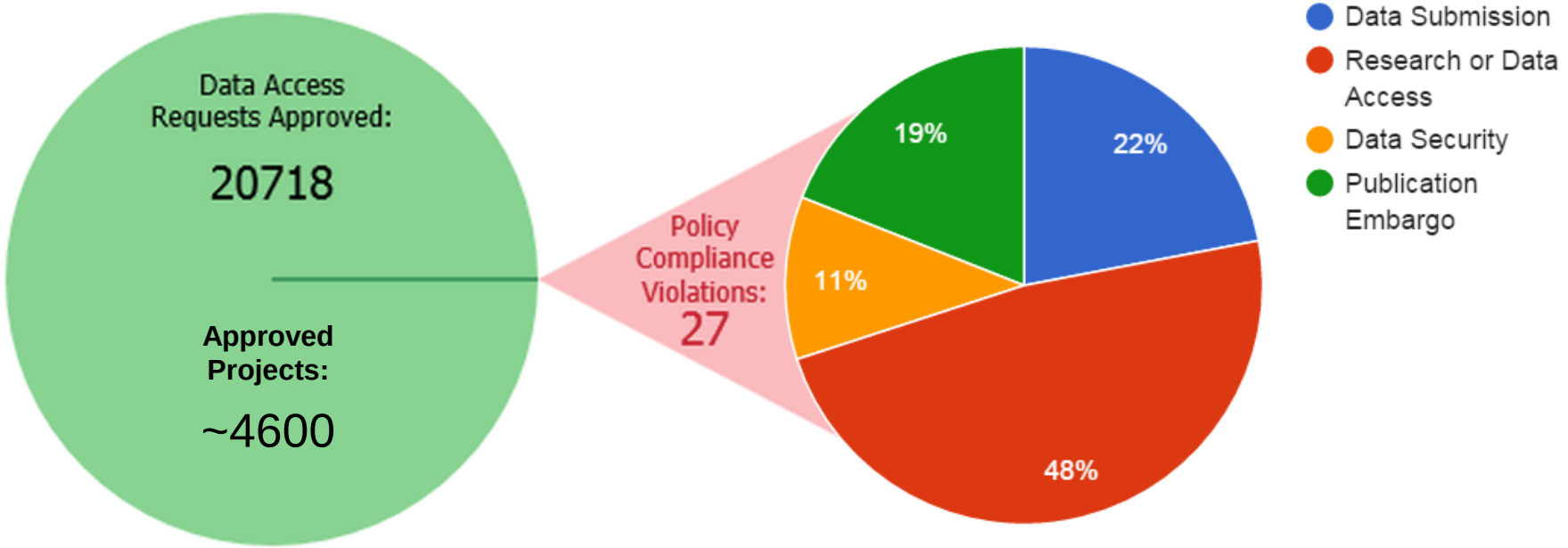
42 = Countries with approved users

1,000,000+ = Participants represented

https://gds.nih.gov/17summary_dbGaP_statistics.html

Slide adapted from Dina Paltoo, Ph.D.

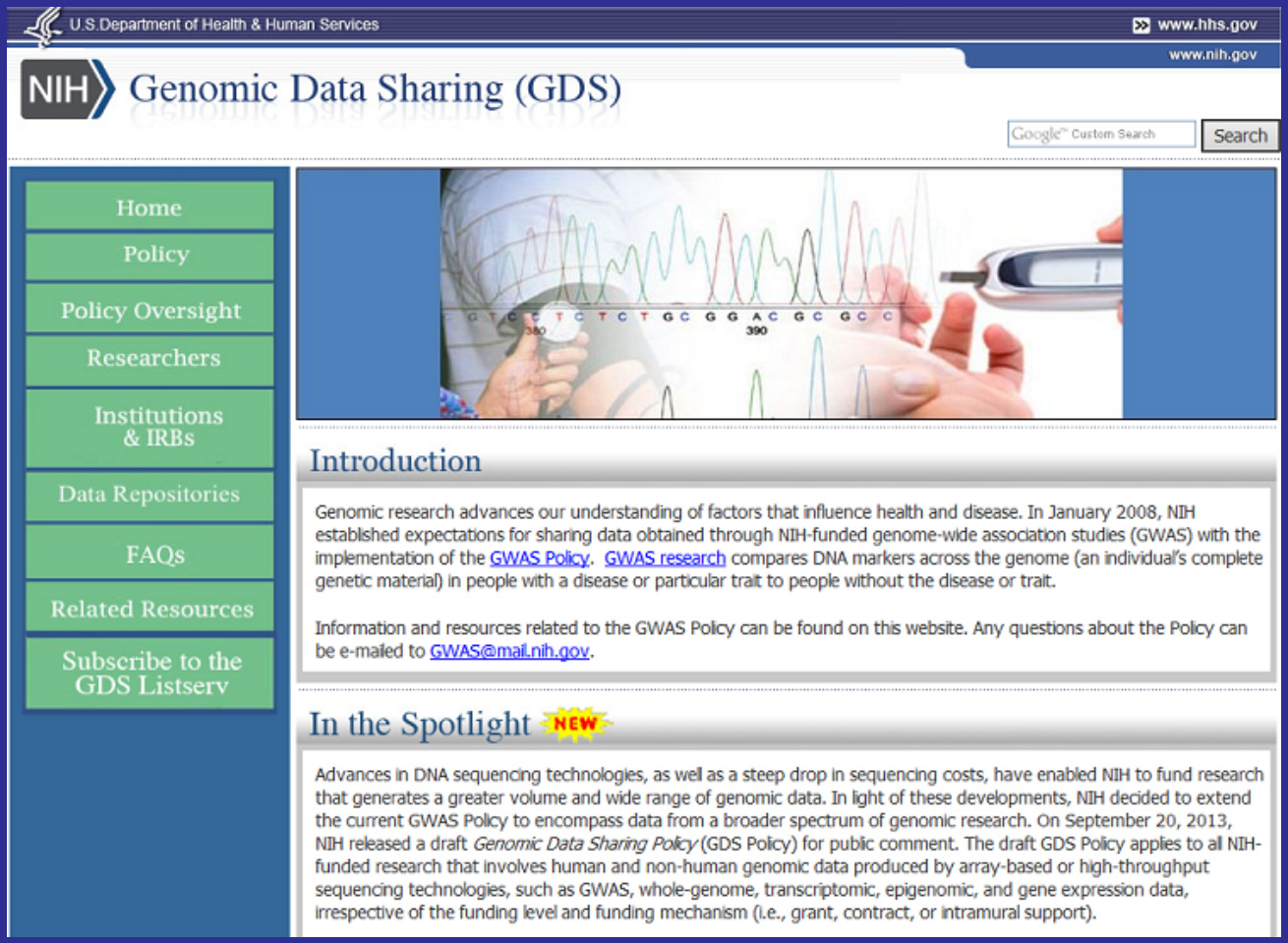
Data Management Experience to Date



- Issues identified by DACs, users, others
- Response and penalties managed through DACs and coordinated centrally

http://gds.nih.gov/20ComplianceStatistics_dbGap.html

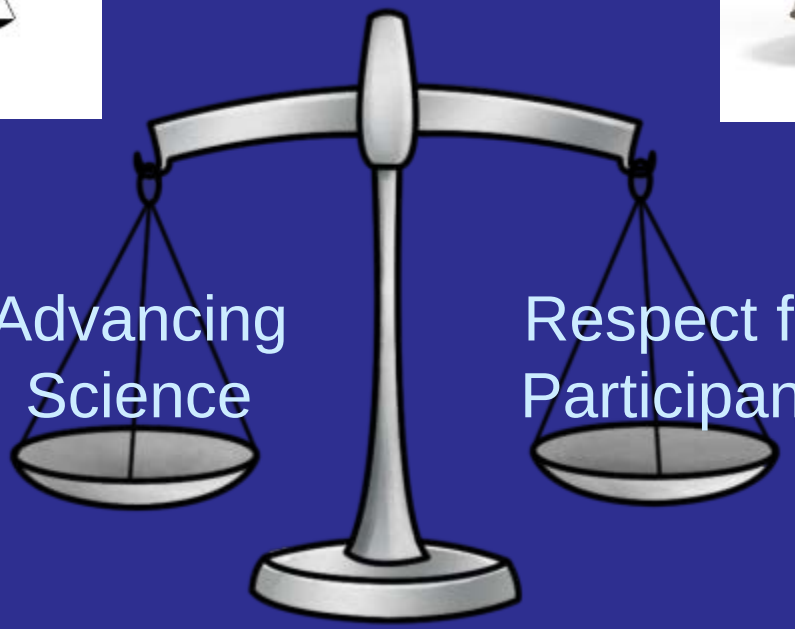
For More Information



http://gds.nih.gov

gds@nih.gov

Goal: Finding the balance …



Prudent Vigilance: Assessing, learning, adjusting …

Merging Values to Achieve Common Goal

NATIONAL HUMAN GENOME RESEARCH INSTITUTE

Thank you!

laura.rodriguez@nih.gov

Advancing human health through genomics research