The NIH Genome-Wide Association Studies Policy:

A common approach to data sharing

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National Human Genome Research Institute

GAIN Analysis Workshop II
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GENETIC ASSOCIATION INFORMATION NETWORK

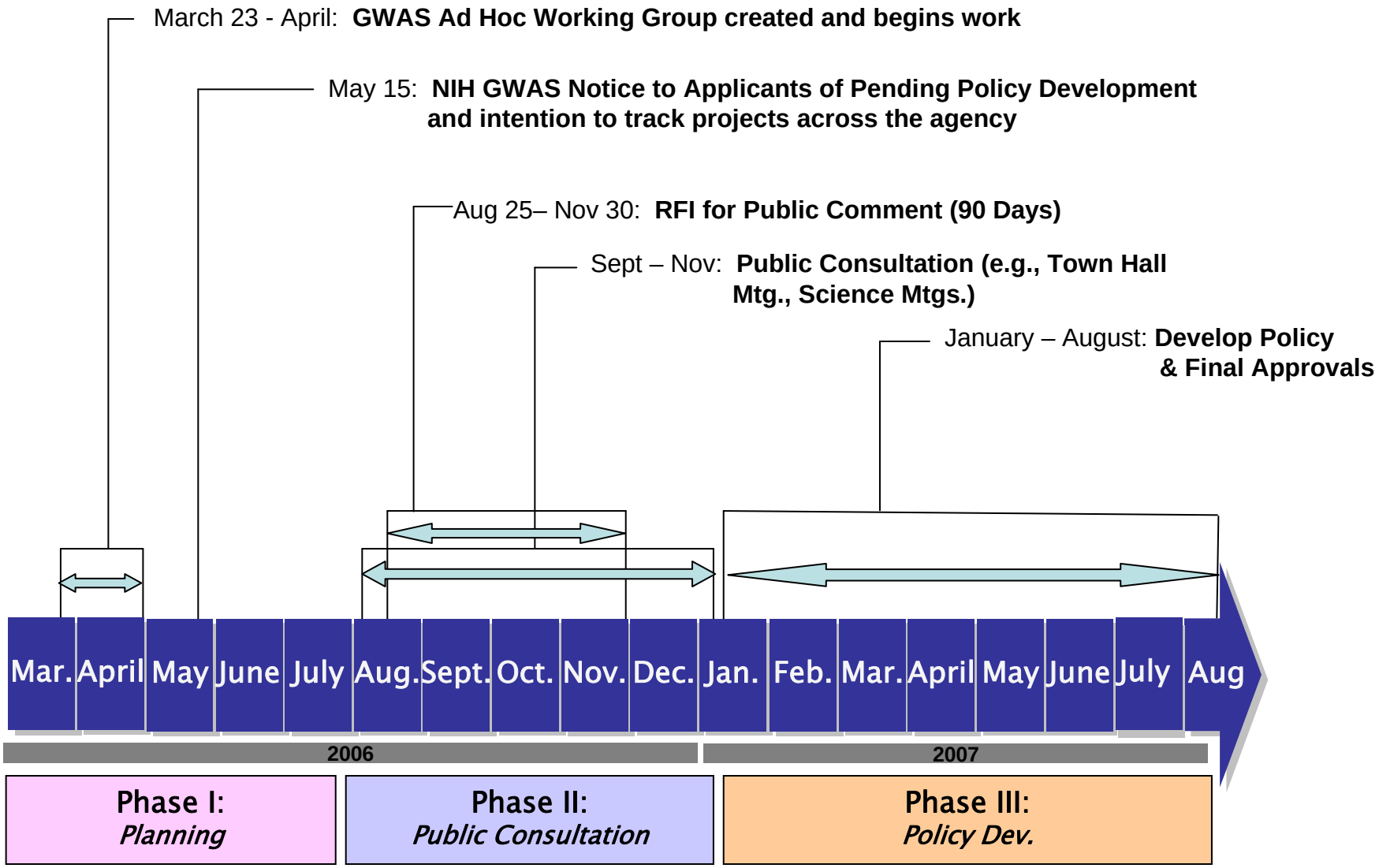
Guiding Principle

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.

Participating Institutes, Centers, & Offices

NHLBI
NHGRI
NLM
NCI
NCMHD
NCRR
NEI
NIA
NIAAA
NIAID
NIAMS
NICHD
NIDA
NIDCD
NIDCR
NIDDK
NIEHS
NIMH
NINDS
NINR
OER
OIR
OSP
OGC
CSR
OBSSR
OLPA

GWAS Timeline (Policy Development)



GWAS Policy Elements

- Data Management
  - Data Submission Procedures
  - Data Access Principles
  - Protection of Research Participants
- Scientific Publication
- Intellectual Property

GWAS Data Management Overview

Data Submission

Data Collection

Submission & Management of Data

Distribution & Secondary Use of Data

Research Participants

Informed consent

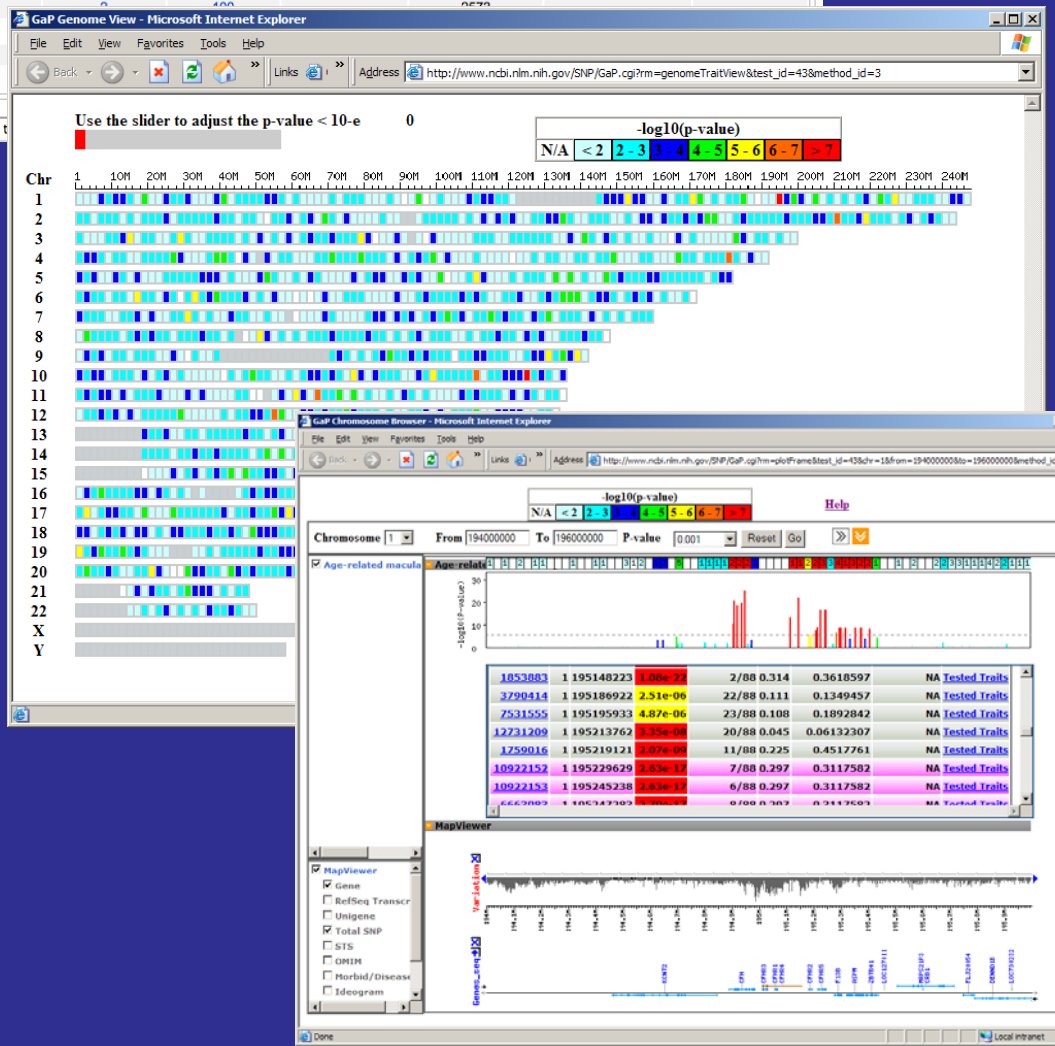
Submitting Investigators

De-identified, Coded Data

GWAS Data Repository

Data Access Request

Recipient Investigators



AREDS: Age Related Eye Disease Study

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NCBI WGA Document
Age Related Eye Disease Study

Chapter 7 EXAMINATION PROCEDURES

7.1 INTRODUCTION

The procedures for carrying out the examinations required in the study are described in this chapter. Required ocular examinations include refraction and visual acuity measurements, intraocular pressure measurement, and ophthalmoscopic examination. General characteristic assessments include measurement of height, weight, and blood pressure and determination of past medical history. Risk factor assessments will require the administration of the food frequency and sunlight exposure questionnaires as well as collection of blood specimens. Procedures for participant identification, masking, distribution and management of the supplementation, adherence assessment, and home visit examination are also described. Procedures for taking photographs of the lens and fundus are described in detail in Chapter 8. The schedule and description of participant visits in Chapter 6 outline the examinations required during each visit.

7.2 REFRACTION AND VISUAL ACUITY

A manifest refraction and visual acuity measurement according to the detailed study protocol must be performed during (a) the Qualifying Visit when the visual acuity score using Chart R is 73 letters or less in at least one eye, (b) the Randomization Visit, (c) Annual Visits, and (d) any Nonannual Visit when the visual acuity score using Chart R has dropped by 10 letters or more compared to the Randomization Visit score for the first time. Participants' pupils should not be dilated at the time of visual acuity testing at any study visit; except they may be dilated during the Qualifying Visit. Pinhole acuity will not be tested as part of AREDS. At the Qualifying Visit, visual acuity may be initially assessed utilizing the participant's current distance glasses. At the Nonannual Visits, visual acuity is initially assessed utilizing the previously obtained manifest refraction. Participants will be asked to read the letters on Chart R only (not Charts 1 or 2), using the equipment described in Section 7.2.1. They will start reading from the top left-most letters--first with the right eye and then with the left eye. A visual acuity score will be calculated as described in Section 7.2.3.3. If at the Qualifying Visit the visual acuity is 74 letters or more in each eye or if at a Nonannual Visit the visual acuity is within nine letters of the Randomization Visit score in each eye, or a vision drop has already been documented in each eye, the visual acuities measured will be entered on the study form. For these participants, a manifest refraction and measurement of best-corrected visual acuity, using the detailed protocol (Sections 7.2.1 - 7.2.3), will not be required.

7.2.1 Visual Acuity Equipment and Facilities

7.2.1.1 Introduction.—The visual acuity of participants will be measured according to the standard procedure developed for the Early Treatment diabetic Retinopathy Study (ETDRS) and adapted for AREDS. The procedure is described in this section. The following equipment is used in AREDS: a set of three Lighthouse Distance Visual Acuity Test charts (second edition), which are modified ETDRS Charts 1, 2, and R, 1 and a retroilluminated box providing standardized chart illumination, as modified from the design by Ferris and Sperduto. 2 The charts and boxes are manufactured by:

Lighthouse Low Vision Products

36-02 Northern Boulevard

Long Island, New York 11101

Data Submission: GAIN/GWAS Similarities

- Approval for submission to the central repository rests with the local institution.
- Investigators and home institutions are responsible for compliance with relevant laws and policies
- Data is de-identified and coded with a random, unique identifier.
- Information regarding any data use limitations is provided at the time of data submission
- Investigators and institutional officials formally acknowledge the scientific publication and intellectual property policies

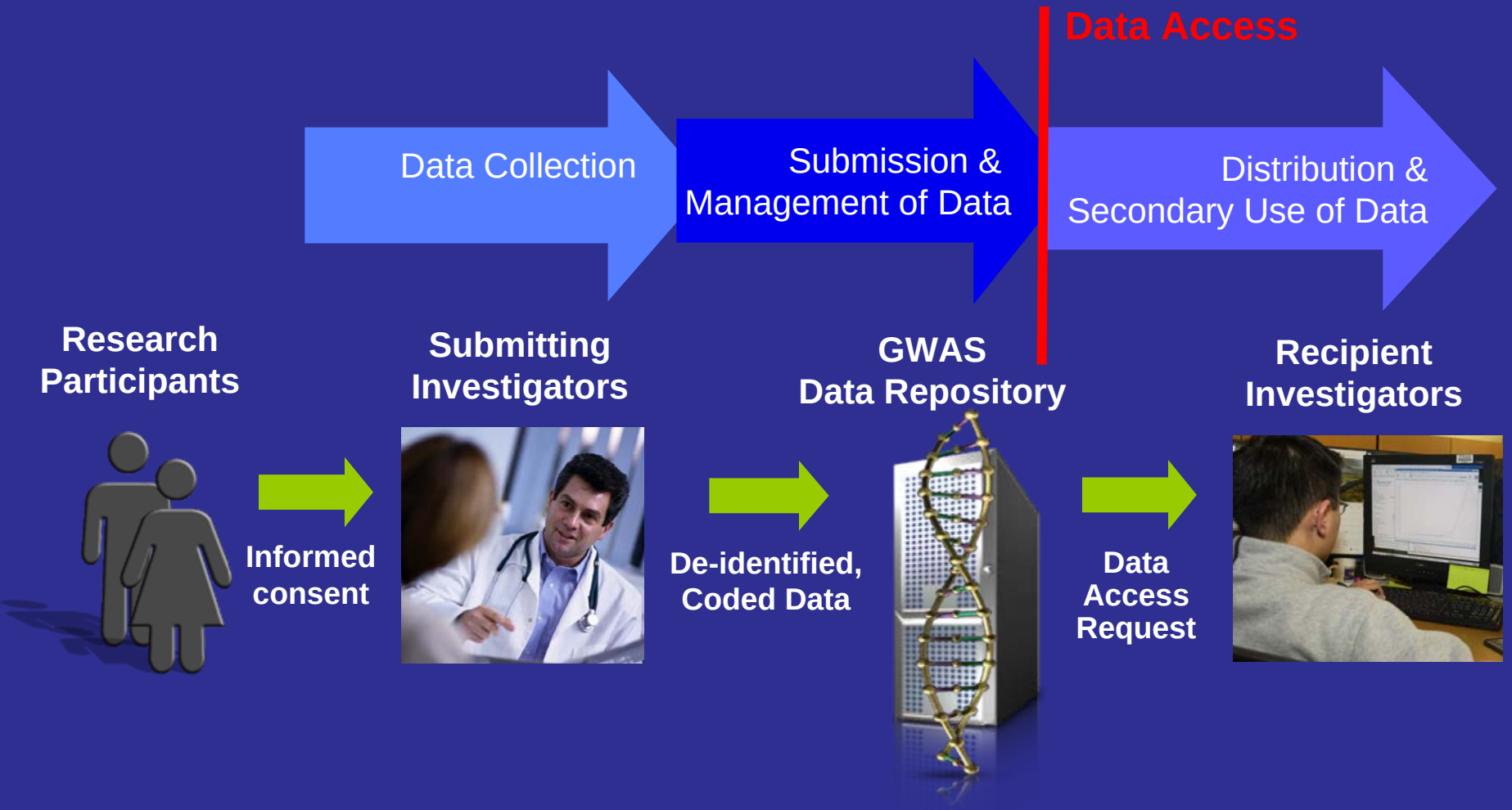
Data Submission: GAIN/GWAS Distinctions

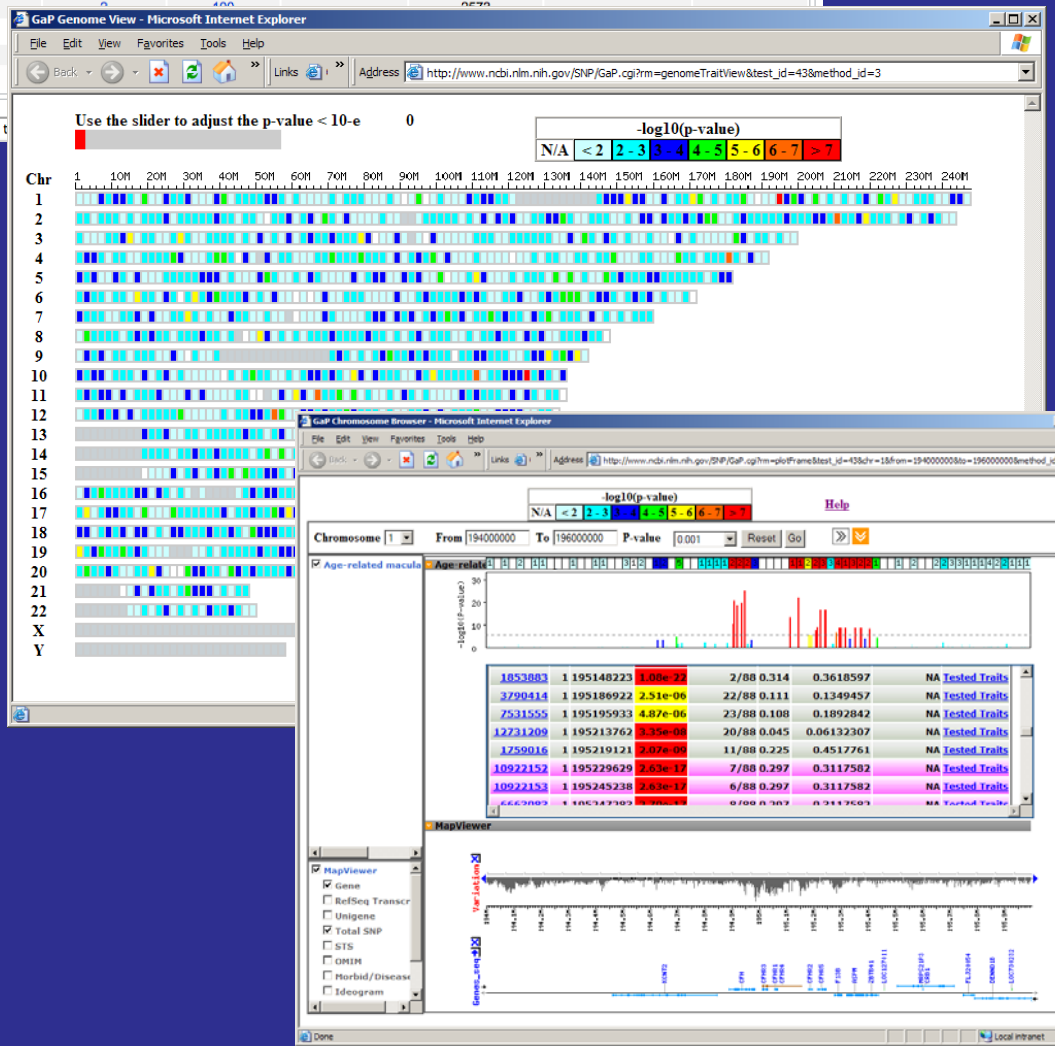
- GAIN obtained agreement to policies and procedures through an “Applicant Agreement” signed by investigators and an Institutional Official
- The NIH GWAS Policy seeks approval for data deposition and agreement to policies through an “institutional certification” that includes review by an IRB or Privacy Board:
  - to assess the de-identification plans for a given dataset
  - to assess the consistency of the informed consent with inclusion in the central repository
- GWAS will not accept datasets that include any of the identifiers listed in the HIPAA Privacy Rule.

Data Submission - Points to Consider

- Intent is to provide investigators and IRBs reviewing datasets for GWAS submission with information on important participant protection considerations
- Topics include:
  - Background on the scientific opportunities presented by GWAS
  - Discussion of the ethical issues relevant to the review of submission plans for GWAS datasets
  - Specific points to consider in the evaluation of informed consent documents

GWAS Data Management Overview





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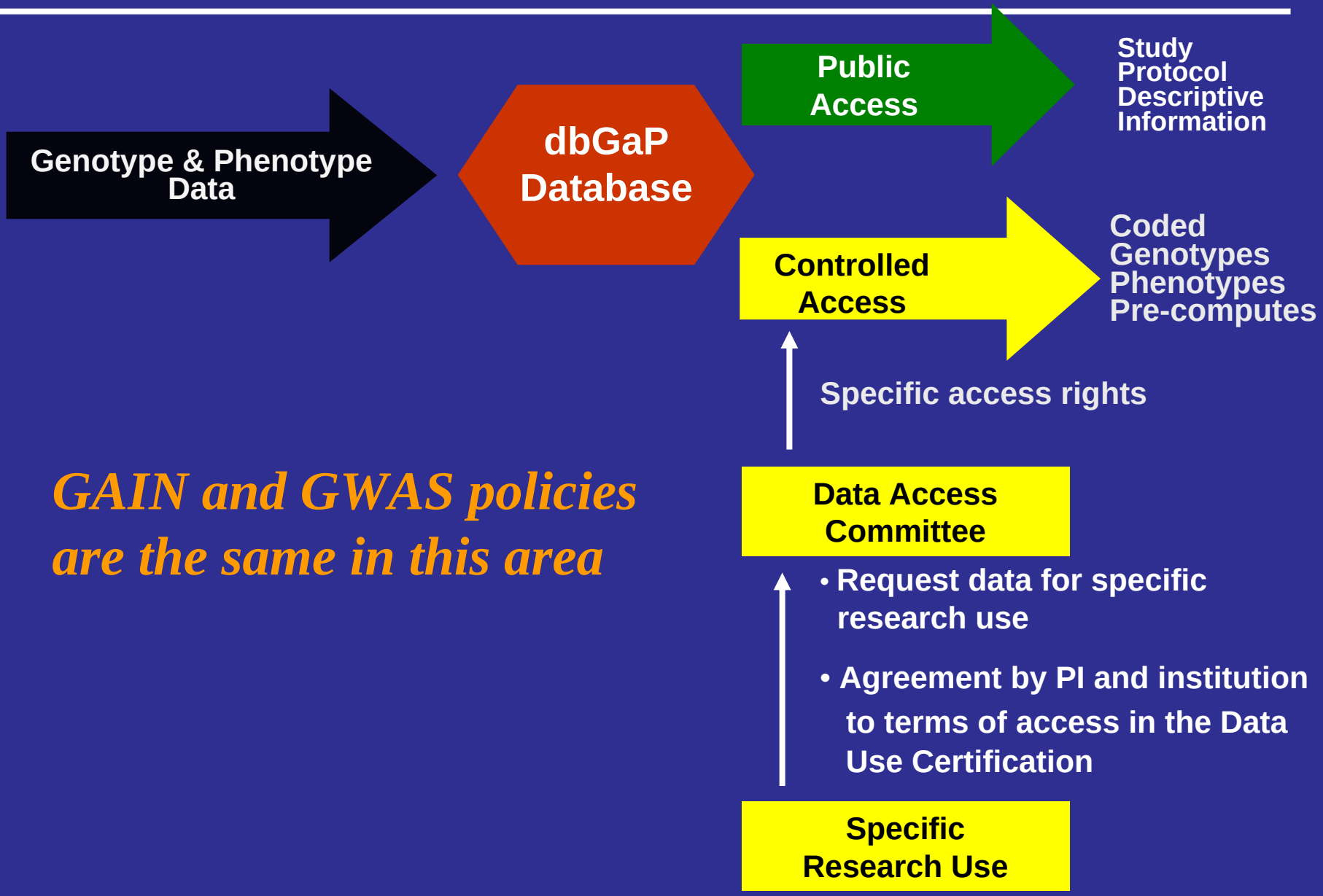
Open Access (summary information)

- Search for studies, review protocols and questionnaires
- View summary phenotype and genotype data
- View pre-computed or published genetic associations (after embargo)
- Identify studies of interest, view their consent conditions, and review terms for data access
- Locate potential collaborators for follow up studies
- **No individual data**

Controlled Access (individual-level)



GAIN and GWAS policies are the same in this area

Data Use Certification Agreement

- The NIH is not currently planning to draft a single Data Use Certification, but the intent is to produce a common framework with similar elements across programs.
- Within the GAIN DUC: Requestors and home institutions will certify that they :
  - are responsible for compliance with federal, state, and local policies
  - will only use the data for the specified research use
  - will disseminate research results broadly and acknowledge GAIN & Contributing Investigators in published or presented work
  - acknowledge GAIN policies on Publication and Intellectual Property
  - will submit brief annual updates on research progress and publications
  - will immediately notify the DAC if a security breach occurs
  - will not identify study participants
  - will not transfer data
  - will be identified within the dbGaP as an Approved User of GAIN data and their approved research use will be posted

Public Disclosure under FOIA

- dbGaP GWAS data will be coded and deidentified.
- Policy concern remains that the extensive genotype data in dbGaP is intrinsically unique.
- NIH officials have agreed that FOIA requests for individual-level GWAS data will be denied.

GWAS Policy Elements

- Data Management
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- Intellectual Property

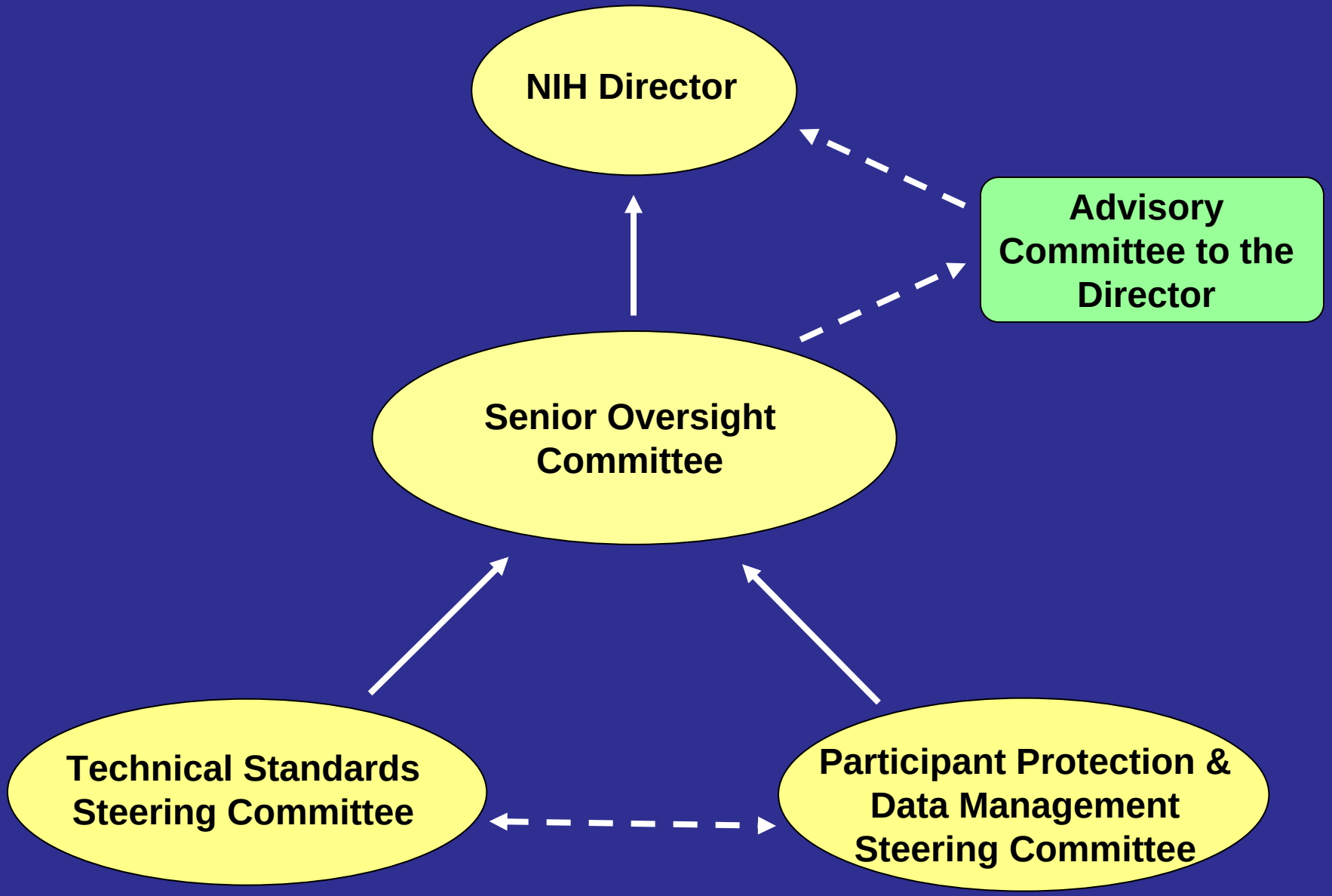
Scientific Publication

- Period of exclusivity for Primary Investigators
  - Maximum period of 12 months for PIs to publish
  - Exclusivity to apply to any public dissemination of the data or analyses
- Acknowledgement of contributing investigators and funding organization

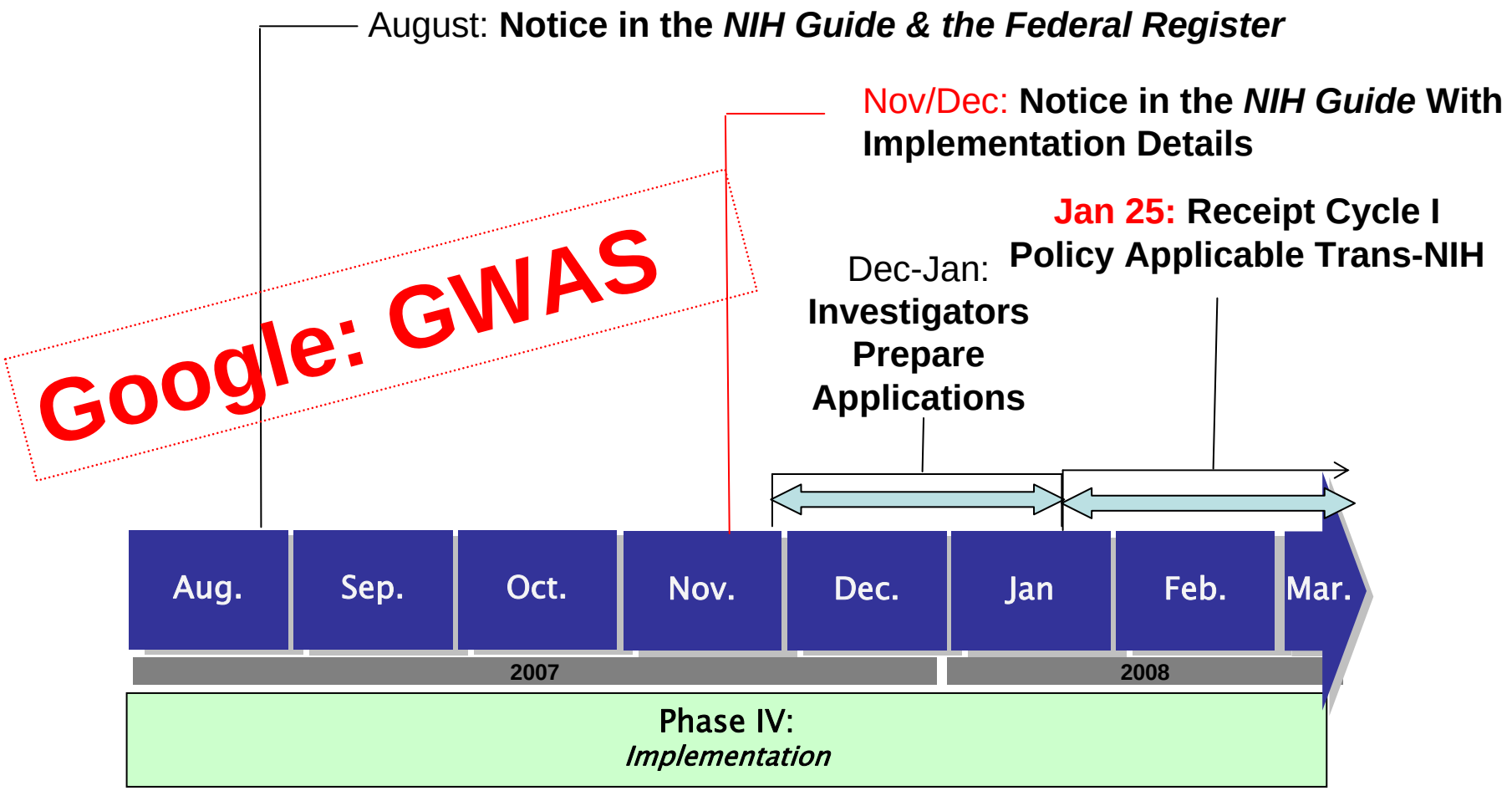
Intellectual Property

- NIH urges that genotype-phenotype associations remain available to all investigators, unencumbered by IP claims.
- NIH discourages premature claims on pre-competitive information.
- NIH encourages broad use of NIH supported genotype-phenotype data consistent with NIH's Best Practices for Licensing with Genomic Inventions.

GWAS Oversight Structure



Implementation Timeline



***Watch Here*: http://grants.nih.gov/grants/gwas/index.htm**

Acknowledgements – GWAS AdHoc Working Group

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Teri Manolio
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Laura Rodriguez
Lisa Brooks
Jean McEwen
Elizabeth Thomson
Jerome Wilson
Katrina Gwinn-Hardy
Anthony Hayward
Elaine Collier
Hemin Chin
James Battey
Jerome Wilson
Katrina Gwinn-Hardy
Anthony Hayward
Elaine Collier
Zhaoxia Ren
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Vivian Ota Wang
David Lipman
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Steve Sherry
Alan Graeff
Lana Skirboll
Marianna Bledsoe
Amy Patterson
Sarah Carr
Norka Ruiz-Bravo
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