

# **Session Overview and Report from the SACGHS Large Population Studies Task Force**

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# Large Population Studies Task Force

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# Issue Description

- Large population studies are one approach to learning more about the relationship among genes, the environment and common disease
- The goals of such studies include:
  - Determining the mechanisms underlying common, complex disease
  - Targeting treatment and prevention strategies
  - Improving health

# Presentation Overview

- Background of full Committee progress
- Task Force review of NIH's Work Group Report
  - *Design Considerations for a Potential United States Population-Based Cohort to Determine the Relationships among Genes, Environment, and Health: Recommendations of an Expert Panel*
- Overview of today's session

# Background

- June 2003: NIH requested that the Committee weigh in on the value of a LPS
- March 2004: SACGHS undertook a priority setting process and determined that the issue of large population studies warranted *in-depth study*
- October 2004: Formed the Large Population Studies Task Force to lead Committee work in this area

# Background

- March 2005: Full day fact-finding session
- Goals included:
  - To inform the Committee about different approaches to large population studies of the relationships between genetic variation, the environment and common disease
  - To facilitate a discussion of the various scientific, logistical, ethical, legal, and social issues around such studies

# Background

- June 2005: TF sought full Committee approval for development of a report
- The Committee decided that a report should be sent to the Secretary that:
  - Identifies the key policy issues around a potential LPS in the U.S.
  - Outlines mechanisms that could be used to address the identified issues

# Background

Major action items from June meeting:

- TF to review NIH's Design Considerations Work Group report and provide an update in October
- TF to coordinate a meeting(s) to gather input from the scientific and ethics communities and the public



# Review of NIH Work Group Report

*Design Considerations for a Potential United States  
Population-Based Cohort to Determine the  
Relationships among Genes, Environment, and Health:  
Recommendations of an Expert Panel*

# Overview of Report Review

- Goals of a potential study as outlined in the report
- Key characteristics of a potential study as outlined in the report
- Key policy issues addressed in the report

# NIH Work Group

- Established to examine the scientific foundations and broad logistical outlines of a hypothetical U.S. large population study
- Comprised of national experts in a variety of fields

# Design Considerations Report: Goals of a Potential Study

The goals of a U.S. large population study would be:

- To ascertain and quantify all of the major environmental and genetic causes of common illnesses
- To set the stage for a future of better preventive medicine and more effective therapy

# Design Considerations Report: Key Potential Study Characteristics

- Representative samples of the U.S. population followed prospectively for development of specific endpoints
  - 500,000 to 1,000,000 participants
  - Sampled from defined census tracts
  - Recruited primarily door-to-door over a four-year period

# Design Considerations Report: Key Potential Study Characteristics

- Data collection at entry would include a wide breadth of phenotypes and environmental factors needed to predict outcomes, balanced by cost and participant burden
  - A core group of baseline variables should be collected in all or nearly all participants

# Design Considerations Report: Key Potential Study Characteristics

- Disease outcomes would be assessed using hospital records, outpatient records, and other data sources such as CMS data and registries

# Design Considerations Report: Key Policy Issues Addressed

- Public engagement
- Representativeness of the cohort
- Need for collaboration (international and national)
- Access to data
- Notification of genetic results and counseling



# Design Considerations Report: Key Policy Issues Addressed

- Intellectual property
- Confidentiality and privacy
- Informed consent
- Central IRB
- Importance of electronic medical records

# Policy Issues Identified by the Large Population Studies TF

# Issues in Need of Further Review

These issues can be divided into four categories:

- BROAD SOCIAL ISSUES
- ENGAGEMENT
- ACCESS AND HEALTH CARE SYSTEM ISSUES
- RESEARCH ISSUES

# Broad Social Issues

- VALUE: Do data exist that indicate the information generated from such a study will be useful?
- NEED: Is a large cohort study the best way to attain information about the genetic and environmental influences on common disease?
- COST: How much will such a study cost?

# Broad Social Issues

- RESOURCE ALLOCATION: What trade-offs would be made if this study were funded?
- RACE AND GENETICS: Would such a study increase or decrease stigmatization? Would it reinforce or help dismantle social constructs of race?
- BENEFIT DISTRIBUTION: Will the benefits of such a study be distributed evenly to all groups within society?

# Engagement

- PUBLIC TRUST, ENGAGEMENT, AND WELFARE: How can public trust of science, genetics, and the government be increased? How should such a study engage the public?
- ENGAGEMENT OF BROAD SCIENTIFIC COMMUNITY: How can input from the broader scientific community be gathered?

# Access and Health Care System Issues

- HEALTH DISPARITIES: Would this study reduce or exacerbate health disparities? Will the results benefit people with limited access to care?
- DIAGNOSIS VS. TREATMENT: How will the ethical dilemma created by widening the gap between what can be diagnosed (or predicted) and what can be treated (or prevented) be addressed?

# Access and Health Care System Issues

- COST TO PARTICIPANTS/ COST-SHARING: What is the cost burden to study participants? How will this affect access to study participation?
- EASE OF ACCESS TO UNDERSERVED AND MINORITY COMMUNITIES: How should minority communities be accessed? If the uninsured are part of the study, how will they be accessed?



# Research Issues

- LEVERAGING EXISTING HHS COHORTS: Have previous HHS cohort studies been effective at answering our research questions?
- STORAGE: How will collected samples be secured, stored and disposed of?
- FAMILY MEMBER NOTIFICATION: Should family members be notified of participants' results that are also relevant to them?

# Research Issues

- DETAILED RECRUITMENT PLAN: What steps will be taken to ethically achieve a cohort of between 500,000 and 1,000,000?
- APPLICATION OF RESEARCH FINDINGS: Guidelines are needed for the application of research findings and anticipated technological developments, with special attention to avoiding discrimination and stigmatization.

# Research Issues

- DEFINITION OF ENVIRONMENT: What does the term “environment” encompass? How should environmental, socioeconomic and behavioral variables be measured?
- NON-COERCIVE RECRUITMENT: How will protocols for recruitment, enrollment and withdrawal be kept free of incentives that are not coercive?

# Overview of Large Population Studies Session

# Session Goals

- Gather input on key policy issues and how to address them from members of the bioethics and scientific communities
- Gather input from public engagement experts on mechanisms and best practices

# Session Purpose

- To inform a report to the Secretary that:
  - Identifies the key policy issues around a potential LPS in the U.S.
  - Outlines mechanisms that could be used to address these issues
- Importantly, the Committee will not endeavor to:
  - Assess the scientific need for such a study
  - Assess the scientific aspects of study or research design

# Session Structure

- The day will consist of three panels:
  - Science
  - Public Engagement
  - Bioethics
- Each panel will be followed by Q&A and discussion with the panelists
- The session will end with full Committee discussion to determine next steps