



# NHANES Genetics Program Recap

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# Benefits of NHANES DNA Analysis

- Unique genetic variant prevalence resource due to the representative nature of the sample
- Valuable genotype/phenotype resource due to the thousands of variables in NHANES

# NHANES DNA Bank

- Started in 1991
- Specimens available for use from:
  - 91-94: n=7,000
  - 99-02: n=8,000
  - 07-12: n=13,800
  - 13: suspended collection

# Evolution of NHANES Genetics Program

- **Genetics Program has evolved**
  - **Anonymized testing →**
  - **RDC testing**
    - **Candidate genes →**
    - **Limited GWAS**
- **Genetic technology advances and analytic changes from candidate gene approaches to that of large scale assays**
  - **increased potential for incidental clinically relevant findings**

## Examples of consent statements related to DNA

- NHANES 1999-2002. Consent form states we do not plan to contact you or your family with individual results from these studies
- NHANES 2009-2010 Consent language changed to say that we will not contact you with results. We also say we will describe completed studies on our web site

# Disclosure of individual genetic data to research participants: the debate reconsidered

Annelien L. Bredenoord<sup>1</sup>, Hester Y. Kroes<sup>2</sup>, Edwin Cuppen<sup>2</sup>, Michael Parker<sup>3</sup> and Johannes J.M. van Delden<sup>1</sup>

## *Argument against:*

- Promotes therapeutic misconception
- Rests on mistaken interpretation of autonomy
- Poses untenable burden on research infrastructure
- Not feasible
- Harmful consequences

## *Arguments for:*

- Beneficence requires disclosure
- Autonomy requires disclosure
- Reciprocity requires disclosure
- Blurring research/ clinical care not bad
- Improves public understanding of genetics

## Relevant Advances in Genetics

- With genetic technology advances, and analytic changes from candidate gene approaches to multiple SNP arrays, there is an increased potential for identifying incidental clinically relevant findings.
- These advances have led to changes in medical ethics guidance on reporting results of genetic tests on bio-banked specimens (blanket non-disclosure is not appropriate).

# May 2011 NHANES Genetics Program Workshop Highlights

- Panel of experts
  - intra/extra mural experts
  - geneticists/bioethicists
- **What** results should be reported back – are standards or guidelines available?
- **How** to determine and operationalize criteria for clinically relevant genetic findings with a dire duty to warn threshold?
- **Who** determines ROF threshold?
- **How/When** to report back?



## Proposed Solution

- BSC oversight of a standing panel of experts to determine notification threshold for clinically actionable results
- One time recontact of previously consented individuals
- Future consent that includes possibility of providing genetic results

## BSC Concerns

- Recontacting participants regarding genetic results
  - Cost especially given low likelihood of reportable condition
  - Unknown reaction of participants upon recontact—possible harm
- Change in current consent form to report genetic results requires pretesting
- Outreach to other organizations given broad impact for other surveys

# Federal Register Notice 6/11/12

- The National Health and Nutrition Examination Survey (NHANES) will not be receiving DNA proposals in the near future. NHANES is changing its plan for making DNA available for genetic research and its proposal guidelines. NHANES will announce when it will reopen its repository for use of DNA specimens for research protocols once it has developed its new plan of operation.

# Next steps

Guidelines for Returning Individual Results from Genome Research Using Population-Banked Specimens: A Workshop

## National Research Council

- Committee on National Statistics
- Committee on Population

## Institute of Medicine

- Roundtable on Translating Genomic-Based Research for Health

# Guidelines for Returning Individual Results from Genome Research Using Population-Banked Specimens: A Workshop

- Topics
  - What should be returned?
  - Who should determine that?
  - Consent
    - **Banked specimens**
    - **Future collection**
- Steering Committee formed
- Javier Nieto -- BSC representative