Collection and Use of Genetic Information in the National Children's Study

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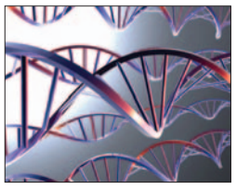
Importance of Genetic Information for the National Children's Study



- Unique opportunity exists in the National Children's Study to determine how genes interact with the environment to influence the health and development of children
- Requires consideration of potential uses of genetic information, collection and storage of biologic materials in an appropriate manner, and awareness of the ethical, legal, and social implications of the use of genetic information
- A workshop held September 8, 2004 in Washington, DC brought together experts in the federal government (NIH, EPA, CDC, FDA) to explore opportunities and challenges for *Collection and Use of Genetic Information in the National Children's Study*

Workshop Objective

To consider available methods to ensure that biologic samples are appropriately collected and stored to provide sufficient quality and quantity of genetic information to study health outcomes over time.

Potential Uses of Genetic Information in the National Children's Study

Identify disease risk factors

- DNA variation and relation to disease and health including disease susceptibility, severity, prognosis; interaction with other risk factors; response to therapeutics

Assess exposures to environmental agents

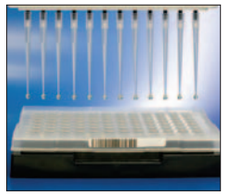
- Changes in gene expression in response to exposure or changes in DNA structure as a result of exposure to environmental agents

Characterize disease outcomes

- Biomarkers used to stratify Study populations into more homogeneous groups

Issues to Consider in Planning for Collection of Genetic Information

Types of specimens (essential, optional) from child and family members



- Peripheral blood, cord blood, buccal cells, placenta (all considered)
- Sampling of parents (essential), grandparents (optional)
- Sampling of subsets of participants for specific studies (essential)

Timing of specimen collection

- One or multiple collection points (depends on the use of the information, e.g., genetic variation, gene expression)
- Integration with collection for other biologic studies

Ensuring sufficient quantity of genetic material

- For testing Core Hypotheses
- For testing in multiple studies over time (options include DNA amplification, immortalized cell lines)

Technology for genetic studies

- Rapidly evolving technology
- Increasing use of genome-wide scans
- Potential for whole genome sequencing at reasonable cost in near future

Challenges

- Limited availability of biologic materials especially at young ages; instability of some types of genetic information such as RNA
- Effects often time-limited (e.g., gene expression) and may not be time-specific (e.g., DNA adducts)
- Effects may be limited to certain tissues, some of which not readily accessible and surrogate tissues may or may not be known

Conclusions and Recommendations from Genetic Information Workshop

- Essential to collect biologic materials to study both genetic variation and gene expression
- Collection, storage, and analytic approaches need to be reconsidered as studies are developed and new technologies become available
- Planning should focus on collection of high quality biologic specimens for genetic studies and storage of sample aliquots for genomic DNA, RNA, protein; whole genome amplification; and cryopreservation

- Specimens to be collected from the child
  - Cord blood collection (key)
  - Peripheral blood sampling in early and late childhood (key)
  - Blood spot in infancy (desirable)
- Family members to be sampled include mother, father, and, if possible, siblings enrolled in study
- Flexibility desired to add other collections in certain situations such as acute, unpredicted exposures (e.g., natural disasters, disease outbreaks)

Implementing the National Children's Study:
Scientific Progress, Challenges, and Opportunities

Study Assembly Meeting

November 29–30, 2005 • Washington, DC

THE NATIONAL CHILDREN'S STUDY

HEALTH GROWTH ENVIRONMENT