FORDHAM UNIVERSITY
THE JESUIT UNIVERSITY OF NEW YORK

THE CENTER FOR ETHICS EDUCATION
CELIA B. FISHER, PH.D., DIRECTOR
www.fordham.edu/ethics

# Informational Risk in Research Involving Genetic Testing

---

**Celia B. Fisher**
Director, Center for Ethics Education
Fisher@Fordham.edu
www.fordham.edu/ethics

---

Society for Research in Child Development

Philadelphia, March, 21, 2015

# Research Involving Genetic Testing Enhances Understanding of the:

- Role of genetic factors in the etiology, onset and escalation of biosocial and behavioral problems
- Interplay between genetic and non-genetic influences
- Sensitivities that may increase prevention success
- **However, understanding of ethical challenges has not caught up with the rapid increase and constantly evolving science**

# Genetic Determinism & Informational Risks to Participants & Populations

- Self identification, stigmatization, informational burden
- Asymptomatic individuals may be treated differently by schools, families, healthcare providers
- New "genetized" disorders may be created
- Misuse of genetic findings for medicalizing behavior, criminal justice decisions, employment barriers, insurance costs
- Shift from health or social policy solutions to individual or family responsibility

# Genetic Determinism & Limitations of Research Designs

Control: Quasi Experimental versus Random Assignment

Generalizability:

- Variability in school, family, peer, neighborhood environments
- Disparities in access to and use of education enhancing activities, health care, psychological services

Temporality: Disorders or behaviors linked to genetic/ polygenetic susceptibility may manifest at different points in the lifespan

# Limitations of Clinical Relevance and Prediction

- Gene-environment interactions are probabilistic.
- Genetic variants may be neither sufficient nor necessary to influence onset
- Myriad pathways and individual variation in expression of disorder
- Most phenotypes associated with developmental problems involve the interplay between multiple genetic and non-genetic factors

# Ethical Risks and Remedies

- Genetic Literacy, Informed Consent and Child Assent
- Data Protection & Disclosure of Genetic Information to Participants & Families
- Genetic Essentialism and Social Justice

# "Genetic Research Literacy"

The degree to which prospective participants are familiar with and can apply information about the use of genetic data to make appropriate research participation decisions.

Developing informed consent procedures can be a teaching moment for participants, guardians *and* researchers

# Enhancing Genetic Research Literacy
# Purpose & Limitations of Research

- Why are we studying genetic influences on development?
- Why child is selected for participation?
- Multifactorial and probabilistic nature of genetic risk
- Analytic validity and clinical utility of results
- Individual, scientific or social value of research?

# Informed Consent: Confidentiality Protections

- How and for how long genetic material will be stored
- If, when, and how materials will be destroyed
- Confidentiality protections including de-identification and risks of identity linkage when appropriate
- Security of laboratories conducting genetic tests
- Linking multiple participant data sets (e.g. school records, health records—FERPA, HIPAA)
- Data protection for multisite studies

# Informed Consent: Research Rights

- What genetic information will or will not be disclosed to participants; with rationale
- Opportunities for and limitations on the right to withdraw data
- Permission to contact participants in the future and share de-identified genetic information with other researchers

# Informed Consent: Biobanks and Future Use of Genetic Data

- Stored data may be used by other investigators for new purposes
- Pleiotropy: Many genes are related to more than one trait
- Unanticipated genetic risk may be discovered
- Environmental and social information stored along with tissue samples

# Informed Consent: Longitudinal Pediatric Research

- Evidence of disease/disorder or protective factors may not emerge for several years following initial data collection
- Or, may emerge in different forms at various points along the lifespan.
- Genetic data stored and re-analyzed ➔ unanticipated information ➔ re-consent

# Child Assent

- Consent ability matures over time
- Maturation may lead to different participation preferences
- Privacy needs change over time
- Assent information needs to be fitted to these capacities and needs
- Re-consent is required, including realistic options to withdraw data

# Informed Consent: Family Privacy Challenges

- When research identifies potential participants through familial characteristics (e.g. substance use, ADHD)
- Inferences about family members' genotype and potential susceptibility to disease/disorder
- Misattributed paternity discovered
- Familial behaviors are identified as mediators or moderators of genetic risk

# Informing Family Members
## Points to Consider

- Does research involve data that can explicitly or implicitly be linked to a family member's genetic profile, pedigree, or behavior
- Are there financial, social, legal or other risks? If so how can they be eliminated or minimized?
- Have you designed procedures to inform family members of risk?

# When Should Researchers Disclose a Child's Genetic Information to Parents?

# Disclosure: Points to Consider

- Is there evidence of a reliable genetic association between behavior or response to prevention?
- Is design sufficient to identify relative contribution of genes and environment?
- How high is the predictive validity?
- Is there a low probability of false positives?
- What is the risk of personal stigmatization or financial burden of increased health costs?

# Disclosure: Points to Consider

- Does the disorder present a major risk to the child's immediate health or wellbeing? Are remedies possible?
- Is this an adolescent or adult onset disorder? Are there preventive interventions available prior to the child's age of majority
- Are evidence-based treatments available?
- Will adequate genetic counseling be provided?
- What is the participant's preference?

# Predictive Genetic Testing: Children's Rights

- Parents/investigators have access to private information which the participant him/herself is unaware
- What informational rights do children have if parents refuse to permit results of genetic testing to be shared with children?
- Do children and families have a right not to know?

# Pediatric Notification Options

- Children given option: (a) Not to know (b) parent tells (c) research staff tells
- All children are notified at age 18 of the availability of their data and their right to know or not know
- Parents and children are informed at the outset of this policy and can refuse participation in the research

# Social Justice and Research Involving Genetic Testing

- Gene-context risk influences societal definitions of normal and abnormal and health and disease.
- New "genetized" diseases may be created
- Public tendency toward genetic essentialism
- Potential for group genetic stereotyping
- Genetic versus social definitions of race and gender

# The Rights of "Genetic Citizenship"

- Federal funding for genomic research is often driven by economic and political concerns (e.g., urban crime, educational costs, mental health disorders, health care costs)
- These may have little to do with or are antithetical to the concerns and social circumstances of participant groups
- Ethical decision-making cannot be isolated from knowledge of how participant's view the informational goods and harms of genomic research

# Doing Good Well: Community Engagement

- Inclusion of participant perspectives and heightened sensitivity to the real world implications of research ethics decisions will…
- Minimize informational risk
- Optimize participant' informed choice
- Consider social justice implications

# Questions/further discussion

# Full Text & References Available from Fisher@Fordham.edu

- Fisher, C. B., & McCarthy, E. L. (2013) Informational Risk and Ethics for Prevention Science Involving Genetic Testing. *Prevention Science.*
- Fisher, C. B. (2006). Privacy and ethics in pediatric environmental health research—Part I: Genetic and prenatal testing. *Environmental Health Perspectives*, *114*, 1617–1621. PMID: 17035153.
- Fisher, C. B. (2006). Privacy and ethics in pediatric environmental health research—Part II: Protecting families and communities. *Environmental Health Perspectives*, *114*, 1622–1625. PMID: 17035154.