Genetic Testing and Screening in Children—Ethics and the Future

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What’s Changing in Genetics and Health Care?

- Far greater uses for genetic information
- Far cheaper to genetic create information
- Far more genetic information about individuals
- Far greater access to genetic information once it is created
  - Not only within health care contexts, but by individuals themselves
    - DTC testing
    - DIY “testing”
Who is Interested in Genetic Information and Why?

- Physicians and patients/parents
  - care decisions
- Insurers
  - info. for health, life and disability risk rating
    - eligibility, premium setting, limitations on coverage
- State and local health departments
  - population info, helps identify disease and allocate resources
- Researchers
  - samples for research studies
    - genetic markers and specific genetic defects
    - epidemiological assessments
- Employers (eventually?)
  - decisions on hiring, benefits and workplace placement
A Range of Issues

- Increasing amounts of genetic information are being collected
  - Personalized medicine is arriving; full genome sequencing is not far off
  - Care and access decisions will be informed if not determined
  - Storing, sharing, and research on genetic data
  - But how useful will it be
    - For children
    - For anyone

- Third party Interests may be in direct conflict with individual interests
  - Not special in or unique to genetics

- Potential for genetic discrimination
  - Concerns that disclosure of test results may affect life and disability insurance of individuals and families
    - GINA prevents some discriminatory uses in health care and employment
  - But other health info can be used in these ways
    - eg, family history

- Legal protections vary
  - GINA, ADA (unclear)

- Informed consent process must address issues in creation and collection of genetic information

- Frontier issues
Bioinformatics challenges are immense
- Bottleneck in the process
- Computing power and storage required
  • How often to “test” (query?) patients’ data?

Access to data
- By whom and for what purposes?
  • How much control by individuals?
    - Should individuals be able to access their own genomic information? What’s different for children?
      • DIY testing
  • What are the research rules
    - Proposed revision of federal regulations
The Challenges of Personalized Medicine

- Tailoring but also limiting therapies?
  - Better therapeutic outcomes? We hope . . .
  - Cost savings? Maybe . . .
  - Limited access to some therapies?
    • What likelihood of effect will be required to prescribe, reimburse?
- Incidental findings/collateral information
  • How to handle; when and what to disclose, and to whom?
    – Actionable clinical information
    – What about information such as paternity, other?
Full Genome Sequencing in Children

Plumetting cost is encouraging considerations around implementation. Allows potential to screen for many more conditions than is currently in the NBS. But expands screening far beyond what has been the goal of NBS - identify risks, early and later in life. Obesity, diabetes, cancer, etc. in addition to traditional genetic diseases. Genome data will become part of their lives. “Structured” health care and lifestyle around genomic info.

But how informative is it likely to be? Much will be highly tenuous; other will be late-onset, etc. Easy to envision overreaction. Ethics perspective has been to postpone testing or screening unless medically important/informative. Allows individuals to decide for themselves. Difficult to accomplish in context of full genome sequencing very early in life.

NIH seeks proposals to study genomic sequencing in newborn period

Funding intended to spur information on implications, potential of new technology.

The National Institutes of Health is seeking proposals for research projects on the implications of information obtained from sequencing the genome to identify diseases in newborns. The intent of funding such projects is to further the understanding of disorders that appear during the newborn period and to improve treatments for these diseases.

Whole genome sequencing is a laboratory method that can determine the entire DNA sequence of a person's genetic material, or genome, from a DNA sample. In recent years, genomic technologies have advanced sufficiently so that it may be possible to use individuals' whole genome information to enhance medical care, including the care and treatment of newborns.

According to the funding announcement, the NIH is seeking proposals “to explore the implications, challenges, and opportunities associated with the possible use of genomic sequence information in the newborn period.”
Privacy and Confidentiality

- Privacy: Individual’s right to control who can gain access to health care information.
- Confidentiality: Information provided with expectation that it will not be disclosed to others or beyond what is agreed upon.

Identifying Personal Genomes by Surname Inference

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Sharing sequencing data sets without identifiers has become a common practice in genomics. Here, we report that surnames can be recovered from personal genomes by profiling short tandem repeats on the Y chromosome (Y-STRs) and querying recreational genetic genealogy databases. We show that a combination of a surname with other types of metadata, such as age and state, can be used to triangulate the identity of the target. A key feature of this technique is that it entirely relies on free, publicly accessible Internet resources. We quantitatively analyze the probability of identification for U.S. males. We further demonstrate the feasibility of this technique by tracing back with high probability the identities of multiple participants in public sequencing projects.

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But the Topography is Changing

- Evolving privacy standards and practices
  - Are expectations different?
    - Good evidence from other contexts that:
      - Most users don’t read privacy policies
      - Majority of users assume that existence of privacy policies mean their data is protected
    - But,
      - Expectations skew with age, depending on who you ask . . .
      - In fact, much more difficult to control access to information than had been the case in the past
      - Move towards control rather than privacy
        - Open data movement
        - Portable Legal Consent; “Citizen Science
  - International standards are evolving
    - EU vs. US; the most stringent will win out since borders matter much less
Late in May, the direct-to-consumer gene-testing company 23andMe proudly announced the impending award of its first patent. The firm’s research on Parkinson’s disease, which used data from several thousand customers, had led to a patent on gene sequences that contribute to risk for the disease and might be used to predict its course. Anne Wojcicki, co-founder of the company, says, “As researchers find more uses for data, informed consent has become a source of confusion. Something has to change.”
Looking Forward

□ Many more uses of genetic technologies and information
  - Pharmacogenetics and other “personalized” medicine
  - Management and oversight of biorepositories and large datasets

□ Frontier issues
  - Genetics and infectious disease
  - Genetic modification as therapy
    • Gene therapy is inching closer
      – eg, Leber Congenital Amaurosis (LCA)