

1-1-2016

Introduction to Bioethics Special Supplement V: Ethical Issues in Genomic Testing of Children.

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Recommended Citation

Lantos, John D., "Introduction to Bioethics Special Supplement V: Ethical Issues in Genomic Testing of Children." (2016).
Manuscripts, Articles, Book Chapters and Other Papers. 389.
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Introduction to Bioethics Special Supplement V: Ethical Issues in Genomic Testing of Children

John D. Lantos, MD

abstract

Next-generation genome sequencing of children is one of the most promising and most challenging new technologies in pediatrics. On the one hand, it offers the hope that we will be able to diagnose rare conditions that were previously impossible to diagnose, which, in turn, might lead to new treatments. On the other hand, the technology for sequencing presents daunting problems of interpretation. It is problematic to conduct the research necessary to characterize the pathogenicity of those variants at the same time that we are using them to guide the clinical care of children who have complex medical problems. It is difficult to know how parents will deal with predictive genomic information about their children. It is also difficult to know whether genome sequencing will complement or replace more traditional methods of newborn screening. This special supplement to *Pediatrics* presents some early reports from researchers who are exploring both the technical issues as well as the ethical, legal, and social issues that arise when we perform genomic sequencing on newborns.

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DOI: 10.1542/peds.2015-3731B

Accepted for publication Nov 10, 2015

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PEDIATRICS (ISSN Numbers: Print, 0031-4005; Online, 1098-4275).

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FINANCIAL DISCLOSURE: The author has indicated he has no financial relationships relevant to this article to disclose.

FUNDING: No funding.

POTENTIAL CONFLICT OF INTEREST: The author has indicated he has no potential conflicts of interest to disclose.

To cite: Lantos JD. Introduction to Bioethics Special Supplement V: Ethical Issues in Genomic Testing of Children. *Pediatrics*. 2016;137(s1):e20153731B

Whole-genome sequencing of children is one of the most promising and one of the most disturbing new technologies in pediatrics. On the one hand, it offers the hope that we will be able to diagnose rare conditions that were previously impossible to diagnose, which, in turn, might lead to new treatments. On the other hand, the technology for sequencing presents daunting problems of interpretation. Among our 6 billion base pairs, there are millions of variants of unknown significance. It is difficult to conduct the research necessary to characterize the pathogenicity of those variants at the same time that we are using them to guide the clinical care of children who have complex medical problems.

In August 2012, the Eunice Kennedy Shriver National Institute of Child Health and Human Development and the National Human Genome Research Institute issued a call for project proposals that would be designed "...to explore the implications, challenges and opportunities associated with the possible use of genomic sequence information in the newborn period." They favored projects that looked at the analysis of genomic data sets; the use of DNA-based analyses for newborn screening; and research related to ethical, legal, and social implications of the possible implementation of genomic sequencing of newborns.

Eventually, they funded projects at 4 sites: Children's Mercy Hospital (CMH) in Kansas City, University of California–San Francisco (UCSF), University of North Carolina (UNC), and a collaborative project between Harvard University and Baylor College of Medicine (Harvard/Baylor).

Each of the projects has a different focus. The project at CMH focuses on the use of next-generation sequencing to diagnose critically ill newborns in the NICU. The Harvard/Baylor project is enrolling critically ill newborns but also includes a control group of healthy newborns. UCSF is comparing the use of next-generation sequencing with the traditional methods used for newborn metabolic screening and exploring the attitudes of parents and physicians regarding this form of testing. The UNC project focuses on helping parents understand the type of results such testing might produce and assisting physicians to decide which results should be returned to parents. Each project has technical, clinical, and bioethical components.

This special supplement to *Pediatrics* is a first look at some of the ethical issues the investigators have encountered in these projects. We begin with an article by Thiffault and Lantos that explains the technology and some practical aspects of interpreting the massive amount of data that are generated by any

analysis of a whole-genome or a whole exome. The second article, by King and Smith, analyzes the legal framework for newborn screening programs and speculates about the implications of genome sequencing for those programs. Lewis and colleagues from UNC discuss the development of a decision aid tool that is designed to help parents consider the type of information that they would like to receive about their infants. Frankel, Pereira, and McGuire of the Harvard/Baylor project discuss what we know—and what we hope to learn—about the potential psychosocial risks of genome screening of newborns. In a related article, Waisbren and colleagues from Harvard present data on psychosocial factors that influence parents' interest in genomic screening of their children. Joseph and colleagues from UCSF present the preliminary results of focus groups that they have conducted with diverse sets of pregnant women in the San Francisco Bay area about their knowledge, attitudes, and beliefs regarding genomic testing. Finally, Deem, from CMH, analyzes some implications of genomic screening and our attitudes about children with disabilities.

Taken together, these articles represent an early report from programs that are starting to explore the clinical uses of genome sequencing and the ethical, legal, and psychosocial issues that have arisen.

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Pediatrics 2016;137;S1

DOI: 10.1542/peds.2015-3731B

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The online version of this article, along with updated information and services, is located on the World Wide Web at:

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