Future Opportunities for Genome Sequencing and Beyond

Recommendations on Clinical Sequencing at Scale

NHGRI Workshop, July 28-29, 2014

Carolyn M. Hutter, PhD

Integrating Genomic Sequencing into Clinical Care: CSER and Beyond

September 28, 2015

Genome Sequencing Program Workshop

National Human Genome Research Institute
Advancing human health through genomics research

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Charting genome sequencing's future: NHGRI workshop offers potential paths to discovery and the clinic

By Steven Benowitz
Associate Director of Communications, Extramural Research Program

On Other Sites:

YouTube Future Opportunities for Genome Sequencing and Beyond Workshop Report
Adam Felsenfeld, Ph.D.
Presentation to Council
Sept. 8, 2014

On July 28-29, 2014, the National Human Genome Research Institute (NHGRI) convened geneticists, biologists, clinicians, bioinformatics specialists and others to provide expert advice on the future of its flagship Genome Sequencing Program (GSP).

The two-day meeting, titled, *Future Opportunities for Genome Sequencing and Beyond: A Planning workshop for the National Human Genome Research Institute*, was the third formal review of the GSP, the previous discussions taking place in 2005 and 2009. After the last review, new core programs were added, including efforts to find causes of rare diseases and move genome sequence information into the clinic. In addition to the long-established Large-Scale Sequencing and Analysis Center (LSAC) program, the GSP now includes the Centers for Mendelian Genomics program, the Clinical Sequencing Exploratory Research (CSER) program and the Genome Sequencing Informatics Tools (GS-IT) program.

NHGRI should continue its role as a "genomics trailblazer," said NHGRI Director Eric Green, M.D., Ph.D. This means supporting research into new DNA sequencing methods and data analysis techniques and solving difficult problems in genome sequencing, he said.

The success of NHGRI's efforts in this area will lie in its ability to partner with other institutes and organizations, he

http://www.genome.gov/27558232

July Workshop Topics

I
- Genetic architecture of health and disease at scale

II
- Integrating genomic variant discovery with function

III
- Clinical genome sequencing at scale

IV
- Comparative and Evolutionary Genomics

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Clinical Genome Sequencing at Scale

Break Out Group Members

- Katrina Armstrong
- David Bentley
- Les Bieseker
- Stephen Chanock
- Rex Chisholm
- Mildred Cho
- Mark DePristo
- James Evans
- Stephen Fodor
- Richard Gibbs
- Robert Green
- Lucia Hindorff
- Carolyn Hutter
- Gail Jarvik
- Hanlee Ji
- Steve Joffe
- Dan Kastner
- Bruce Korf
- David Ledbetter
- Dan Masys
- Amy McGuire
- Patrice Milos
- Cynthia Morton
- Lucila Ohno-Machado
- Sharon Plon
- Heidi Rehm
- Dan Roden
- Pamela Sankar
- Xun Xu

Recommendations for Clinical Genome Sequencing

1. Define clinical contexts in which genome sequencing improves patient outcomes
2. Enable rapid, robust detection of all clinically relevant variation in a single test
3. Leverage clinical sequencing data for research use
4. Improve approaches for determining pathogenicity
5. Identify effective methods for implementing sequencing in medical practice

Define clinical contexts in which genome sequencing improves patient outcomes

- Support catalytic research
- Demonstrate whether/ when genome-scale testing improves health
- Develop evidence based paradigms
- Address role of sequencing in public health

Enable rapid, robust detection of all clinically relevant variation in a single test

- Improve accuracy
- Decrease cost and turn-around time
- Expand to RNA/ transcriptome variation when merited
- Increase the spectrum of tissues undergoing clinical sequencing

Leverage clinical sequencing data for research use

- Foster a “virtuous cycle”
- Use exemplar studies to influence key areas:
  - clinical utility
  - clinical implementation
  - data sharing
  - tool development

Improve approaches for determining pathogenicity

- Incorporate genetic, functional and computational data
- Expand beyond manual curation
- Facilitate the development of standards

Identify effective methods for implementing sequencing in medical practice

- Develop novel clinical decision support tools
- Broaden the population impact
- Connect with professional societies

July Workshop Resources Online

http://www.genome.gov/27558042

Acknowledgements

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Adam Felsenfeld, Shannon Biello, Deborah Colantuoni, Lucia Hindorff, Carolyn Hutter, Lu Wang, Elise Feingold, Mike Pazin, Mike Smith, Heidi Sofia, Chris Wellington, Kris Wetterstrand

Workshop Attendees