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Genomics and Society Working Group Update

February 10, 2020



National Human Genome
Research Institute

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Mission

- To provide to Council advice on short- and long-range planning and priority setting for Genomics and Society activities at the Institute, with particular emphasis on the ELSI Research Program in the NHGRI Division of Genomics and Society



Current Members



Jeffrey Botkin, M.D. (Chair)
Steven Joffe, M.D., M.P.H
Sandra Soo-Jin Lee, Ph.D.
Maxwell Mehlman, J.D.
Melanie Myers, Ph.D.
Vardit Ravitsky, Ph.D.
Consuelo Wilkins, M.D.,
M.S.C.I.

Incoming Members

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Malia Fullerton
University of
Washington



Alondra Nelson
Institute of
Advanced Studies



Lisa Parker
University of
Pittsburgh

Key Discussion Points from May 2019

- Lessons from Canada on GE3LS (Genomics and its Ethical, Environmental, Economic, Legal and Social aspects) Research – Vardit Ravitsky
 - All Genome Canada applications required to have an integrated GE3LS component
 - GE3LS research is broader than ELSI with explicit inclusion of environmental and economic aspects
- History of Genomics Program– Chris Donohue

Key Discussion Points from May 2019

- Broad discussion of how to consider the impact of ELSI research
 - We identified large number of possible stakeholders and metrics related to scholarship and non-scholarly impacts
 - Recommend tying any metrics to the goals and mission of ELSI research program
- Update from the NIH Office of Science Policy – Adam Berger
 - \$5M annual budget for bioethics research
 - Discussed strategies for fostering bioethics research across NIH

GSWG Strategic Planning Activities

- Devoted significant time & effort to discussing the focus area of society, education and engagement
- Key areas of input:
 - Distinguish between ELSI research, education and engagement
 - Emphasize *research* questions for ELSI
 - Integrate values throughout the strategic plan (e.g., diversity, privacy)
 - Ensure goals are concrete and aspirational
 - Discussed strategies for outreach and engagement for the ELSI research community

Highlights from January 2020 Meeting

- Training team update, including a preliminary analysis of ELSI trainee career paths
 - Highly productive training pipeline from original CEERs
- Update on the H3Africa program — Ebony Madden
 - Progress of ELSI research studies to date
 - Resources developed by H3A Ethics and Regulatory Working Group

Discussion Topics January 2020

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- Polygenic Risk Scores:
 - Uncertain clinical utility
 - Lack of reproducibility across populations/groups
 - Expansion of PRS into behavioral traits and prenatal testing
- Genetic genealogy and forensic use
 - Active area for policy development – from data producers and states
 - Data used in different contexts and purposes; unclear if people understand or support such uses
- Gene editing & gene therapy
 - Less attention to somatic editing, yet conceptual questions about concepts like “serious disease” or when to consider a therapy “safe”
 - Cost and access to resultant therapies is a huge concern

Discussion Topics January 2020

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- Long-term outcome data collection
 - Theme emerging from recent NHLBI workshop
 - Create an evidence base for clinical and personal utility
 - Normative questions, e.g. when clinical sequencing *should* be implemented
 - Consider both clinical and non-clinical endpoints
 - ELSI integral to genomic medicine implementation research questions
- Achieving diversity in genomic research participants
 - Identify and implement proven tools and resources
 - Continue efforts to develop new tools and workforce resources
 - Consideration of stipulations in RFAs of measures to enhance diversity



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