

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY**

September 24-25, 2018

The Open Session of the 84th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, September 24, 2018 at the 6700B Rockledge Conference Center in Bethesda, Maryland. Dr. Eric Green, Director of the National Human Genome Research Institute (NHGRI), called the meeting to order.

The meeting was open to the public from 10:00 AM – 4:00 PM on September 24, 2018. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 8:00 AM – 10:00 AM and 4:00 PM – 6:00 PM on September 24, 2018; and from 8:30AM until adjournment on September 25, 2018 for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT:

Jeffrey Botkin (University of Utah)
Carol Bult (Jackson Laboratory)
Brenton Graveley (University of Connecticut)
Gail Henderson* (University of North Carolina)
Mark Johnston (University of Colorado)
Jonathan Pritchard (Stanford University)
Dan Roden (Vanderbilt University)
Val Sheffield (University of Iowa)
Jay Shendure (University of Washington)

AD HOC ADVISORS PRESENT:

Wendy Chung (Columbia University)
Nancy Cox (Vanderbilt University)
Mark Craven (University of Wisconsin)
Patricia Deverka (American Institutes for Research)
Stephen Fodor* (13.8, Inc.)
Jonathan Haines (Case Western Reserve University)
Trey Ideker (University of California, San Diego)
Rafael Irizarry (Dana-Farber Cancer Institute)
Sharon Plon (Baylor College of Medicine)
Albert Reece (University of Maryland)
Stephen Rich (University of Virginia)

**indicates participation by phone*

STAFF PRESENT FROM THE NATIONAL INSTITUTES OF HEALTH:

Vence Bonham, IOD and IRP	Jyoti Dayal, ERP
Joy Boyer, ERP	Chris Donahue, DPCE
Larry Brody, ERP	Carla Easter, DPCE
Comfort Browne, ERP	Elise Feingold, ERP
Eileen Cahill, ERP	Adam Felsenfeld, ERP
Lisa Chadwick, ERP	Valentina di Francesco, ERP
Christine Chang, ERP	Robert Fullem, ERP
Joanna Chau, ERP	Sylvia Garvey, ERP
Michelle Cohen, ERP	Elena Ghanaim, DPCE
Heather Colley, ERP	Tina Gatlin, ERP
Christina Daulton, DPCE	Bettie Graham, ERP

Linda Hall, ERP
Joseph Henke, DOM
Lucia Hindorff, ERP
Rebecca Hong, DPCE
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Sonya Jooma, DPCE
Sheethal Jose, ERP
David Kaufman, ERP
Rongling Li, ERP
Britny Kish, DPCE
Rongling Li, ERP
Nicole Lockhart, ERP
Allison Mandich, IOD
Ebony Madden, ERP
Donna Messersmith, DPCE
Emily Neveux, DOM
Ken Nakamura, ERP
Vivian Ota Wang, ERP
Michael Pagan, ERP
Kiara Palmer, DPCE

Erin Ramos, ERP
Mike Pazin, ERP
Natalie Pino, ERP
Lita Proctor, ERP
Jessica Reusch, DPCE
Robb Rowley, ERP
Laura Rodriguez, DPCE
Ellen Rolfes, DOM
Ben Ryan, IOD
Mike Smith, ERP
Heidi Sofia, ERP
Taylorlyn Stephan, ERP
Tasha Stewart, ERP
Jeff Struewing, ERP
Michelle Tallman, ERP
Cecelia Tamburro, ERP
Elizabeth Tuck, DPCE
Harry Wedel, ERP
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP

OTHERS PRESENT FOR ALL OR A PORTION OF THE MEETING:

Anne Berry, Association of American Medical Colleges (AAMC)
Mona Miller, American Society for Human Genetics (ASHG)
Michael Watson, American College of Medical Genetics (ACMG)

INTRODUCTION OF NEW COUNCIL MEMBERS, NHGRI STAFF, LIAISONS, AND GUESTS

Dr. Rudy Pozzatti began the Open Session with the introduction of new members of the NHGRI staff, liaisons, and guests, which can be found at:

<https://www.youtube.com/watch?v=rYIMufrYHY>.

APPROVAL OF MINUTES

The Council approved the May 2018 Minutes by unanimous vote.

FUTURE MEETING DATES

- Feb. 11-12, 2019
- May 20-21, 2019
- Sept. 16-17, 2019
- Feb. 10-11, 2020
- May 18-19, 2020
- Sept. 14-15, 2020

DIRECTOR'S REPORT

Dr. Eric Green gave his Director's Report, which included a series of updates on recent activities at NHGRI, NIH, and in the broader genomics research community. A [video of his presentation](#) and the slides can be found at:

https://www.genome.gov/Pages/About/NACHGR/September2018AgendaDocuments/Directors_Report_September_2018.pdf.

REPORT – Genomics and Society Working Group Annual Report

Dr. Jeffrey Botkin provided an update on the Genomics and Society Working Group (GSWG) of NACHGR. The Working Group provides advice about the ELSI Research Program and activities of NHGRI's Division of Genomics and Society. The May 2018 meeting report did not generate any specific recommendations for NACHGR, but other Council members suggested evaluating the ethical, legal, and social implications of polygenic risk scores at future GSWG meetings.

GSWG will host a strategic planning session for NHGRI at the [American Society for Bioethics and Humanities Meeting](#) on October 19, 2018.

A video of Dr. Botkin's presentation can be found at:

<https://www.youtube.com/watch?v=homjCgNvBJQ>.

REPORT – National Academies of Sciences, Engineering and Medicine: *Returning Individual Research Results to Participants: Guidance for a New Research Paradigm*

Dr. Jeffrey Botkin gave a summary of the NIH co-sponsored report by the National Academies on returning individual research results to participants. Two Health and Human Services regulations – Clinical Laboratory Improvement Amendments (CLIA) and Health Insurance Portability Accountability Act (HIPAA) – provide contradictory guidelines. The charge to the committee generating the report was to determine if/when it is appropriate to return individual research results to research participants by reviewing current practices, examining the evidence of benefits, risks and cost, and considering the ethical, social and regulatory aspects associated with returning results.

A video of Dr. Botkin's presentation can be found at: https://www.youtube.com/watch?v=12-ZZ_7sKOW.

CONCEPT CLEARANCE – *Human Genome Reference Center and Related Concepts*

Dr. Adam Felsenfeld presented five concepts that would constitute a proposed Human Genome Reference Program. The Program would provide new levels of support for the [Human Genome Reference](#) by: updating reference builds and representations of the human genome; adding new high-quality human genome sequences; developing bioinformatic tools for the clinical and basic science communities; and developing education and outreach activities.

The Council unanimously approved the concepts. A video of Dr. Felsenfeld's presentation can be found at: https://www.youtube.com/watch?v=iD9H9_rY-4c.

REPORT – *Genomic Medicine Working Group Activities in 2018*

Dr. Teri Manolio gave the annual report of NACHGR's Genomic Medicine Working Group (GMWG). The GMWG advises NHGRI on the research needed to evaluate and implement genomic medicine. GMWG is now planning the 11th Genomic Medicine meeting; this one will focus on the role of employers as the primary payers for genomic medicine.

A video of Dr. Manolio's presentation can be found at:

<https://www.youtube.com/watch?v=94Lk5GeQYDI>.

COUNCIL-INITIATED DISCUSSION

A video of the Council-initiated discussion can be found at:

<https://www.youtube.com/watch?v=hSwkHWMV0MI>.

Council Members expressed interest in NHGRI's implementation of NIH's Strategic Plan for Data Science. Dr. Plon requested an update on the Variation, Function, and Disease supplements (approved at the May 2018 NACHGR meeting) at either the May or September

2019 Council meetings. Dr. Chung requested an update on the wrap-up and lessons learned from the Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT) program at a future Council meeting. Council suggested two topics for scientific presentations at a future Council meeting: the value and utility of polygenic risk scores and future approaches for high-throughput functional genomics in the context of NHGRI's new Strategic Plan.

ANNOUNCEMENTS AND ITEMS OF INTEREST

Four retiring Council members were honored for their service: Drs. Carol Bult, Dan Roden, Val Sheffield and Jay Shendure.

Supporting Documents:

- [The Genomics and Society Working Group Report](#)
- [The National Academies of Sciences, Engineering, and Medicine Report](#)
- [Human Genome Reference Program Concept Clearance Presentation](#)
- [Genomic Medicine Working Group Activities in 2018](#)

CONFIDENTIALITY AND CONFLICT OF INTEREST¹

Dr. Rudy Pozzatti read the Confidentiality and Conflict of Interest Policy to Council and asked the members to sign the forms provided to them.

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 146 applications, requesting \$47,168,491 (direct costs). The applications included: 96 research project applications (R01, R03, R15, or R21); 1 research center application (U54); 8 cooperative agreement applications (U01 or U24); 6 career development or career transition applications (K99/R00, K01, or K08); 6 research education applications (R25); 3 conference applications (R13); 15 SBIR Phase I applications (R43); 9 SBIR Phase II applications (R44); and 2 STTR Phase 1 applications (R41). A total of 146 applications totaling \$47,168,491 were recommended by the NACHGR.

This Council Meeting Summary document was prepared by Taylorlyn Stephan, NHGRI Scientific Program Analyst.

2/12/2019

Date

Rudy O. Pozzatti

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

2/12/2019

Date

Eric D. Green

Eric Green, M.D., Ph.D.

Chairman

National Advisory Council for Human Genome Research

¹ Council Members absented themselves from the meeting when the Council discussed applications from their institutions or in which a conflict of interest may occur. Members signed a statement to this effect. This does not apply to "en block" votes.