NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH MEETING SUMMARY

February 12-13, 2018

The Open Session of the 82nd meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, February 12, 2018, at the Fishers Lane Terrace Level Conference Center in Rockville, 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 until 5:00 PM on February 12, 2018. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 8:00 AM to 10:00 AM and 5:00 PM to 5:30 PM on February 12, 2018, and from 8:30 AM until adjournment on February 13, 2018, for the review, discussion, and evaluation of grant applications.

Council Members and Ad Hoc Advisors Present:

Eric Boerwinkle

Jeffrey Botkin

Carol Bult

Wendy Chung

Stephen Fodor

Brenton Graveley

Gail Henderson (by phone)

Chanita Hughes Halbert

Trey Ideker

Mark Johnston

Sharon Plon

Jonathan Pritchard

Aviv Regev

Dan Roden

Val Sheffield

Jav Shendure

Staff Present from the National Institutes of Health:

Gabriel Batzli, ERP

Vence Bonham, IOD and IRP

Joy Boyer, ERP

Comfort Browne, ERP

Christine Chang, ERP Monika Christman, ERP

Heather Colley, ERP

Erin Currey, ERP

Jyoti Dayal, ERP

Valentina di Francesco, ERP

Carla Easter, DPCE

Sheena Faherty, DPCE

Adam Felsenfeld, ERP

Robert Fullem, ERP

Tina Gatlin, ERP

Bettie Graham, ERP

Sylvia Garvey, ERP

Margaret Ginoza, ERP

Linda Hall, ERP

Lucia Hindorff, ERP

Rebecca Hong, DPCE

Carolyn Hutter, ERP

Deanna Ingersoll, ERP

Sonya Jooma, DPCE

Sheethal Jose, ERP

Melpi Kasapi, ERP

David Kaufman, ERP

Jimmie Keel, CC

Britny Kish, ERP

Jennifer Krupp, NIH-ACMG

Rongling Li, ERP

Nicole Lockhart, ERP

Allison Mandich, IOD
Ebony Madden, ERP
Donna Messersmith, ERP
Jenn Montooth, DPCE
Sam Moore, ERP
Ken Nakamura, ERP
Vivian Ota Wang, ERP
Michael Pagan, ERP
Kiara Palmer, DPCE
Mike Pazin, ERP
Lisa Proctor, ERP
Erin Ramos, ERP
Robb Rowley, DIR
Laura Rodriquez, DPCE

Ben Ryan, IOD
Laura Skow, ERP
Mike Smith, ERP
Heidi Sofia, ERP
Taylorlyn Stephan, ERP
Jeff Struewing, ERP
Barbara Thomas, ERP
Elizabeth Tuck, DPCE
Lu Wang, ERP
Cara Weismann, DPCE
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP

Rosann Wise, ERP

Others Present for All or a Portion of the Meeting:

Michael Lauer, NIH OD James Ostell, NCBI/NLM

Materials, including the agenda and PowerPoint presentations for the Open Session of the 82nd NACHGR meeting are available at: https://www.genome.gov/27570550/

A recorded video of the entire Open Session is available at: https://www.youtube.com/playlist?list=PL1ay9ko4A8sm6xyTXFC7ZsjCbG4EJHdm2.

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

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

https://www.youtube.com/playlist?list=PL1ay9ko4A8sm6xyTXFC7ZsjCbG4EJHdm2.

APPROVAL OF MINUTES FOR THE SEPTEMBER 2017 NACHGR MEETING

The Council approved the Minutes by unanimous vote without any edits to either content or format.

FUTURE MEETING DATES

- May 21-22, 2018
- Sept. 24-25, 2018
- Feb. 11-12, 2019
- May 20-21, 2019
- Sept. 16-17, 2019

DIRECTOR'S REPORT

Dr. Eric Green gave his Director's Report, which included a series of updates about recent activities at NHGRI, NIH, and in the broader genomics community. A video of his presentation can be found at: https://www.youtube.com/watch?v=a_UAktr1F70.

PRESENTATION – En Route to a "2020 Vision for Genomics": The Next Round of NHGRI Strategic Planning

Dr. Eric Green introduced two initiatives: an examination of NHGRI's current role in the genomics ecosystem and a plan for revising the current NHGRI strategic vision. A video of this presentation can be found at: https://www.youtube.com/watch?v=cCazVxD2isI. Strategic planning updates can be found at www.genome.gov/genomics2020.

PRESENTATION – Updates from Building 1: Next Generation Researchers Initiative and Clinical Trial Reforms

Dr. Michael Lauer presented on the status of the Next Generation Researchers Initiative (NGRI) and the Clinical Trials Reforms. Dr. Lauer shared a portfolio analysis of NIH grants that explored how decreasing success rates have affected early career stage investigators. He defined NIH's goal of identifying and supporting young researchers who are at risk of a lapse in funding. With respect to the Clinical Trials Reforms, Dr. Lauer reviewed a revision of the NIH Clinical Trials Requirements for Grants and Contracts, which aims to better identify NIH-funded clinical trials, register those clinical trials with clinicaltrials.gov, and track the publication history of those trials. A video of this presentation can be found at:

https://www.youtube.com/watch?v=Cg3Ocm_65LA&list=PL1ay9ko4A8sm6xyTXFC7ZsjCbG4EJHdm2&index=4.

CONCEPT CLEARANCE - Novel Nucleic Acid Sequencing Technology Development

Dr. Michael Smith made a presentation about the Novel Nucleic Acid Sequencing Technology Development Program. Dr. Smith proposed renewing the current Request for Applications (RFA) with modest revisions. The RFA will remain focused on the development of new DNA and RNA sequencing technologies, and supporting research that will result in substantial improvements to current sequencing technologies. Council unanimously voted to approve the concept. A video of this presentation can be found at: https://www.youtube.com/watch?v=H7pG64aUqtc.

REPORT – The Human Microbiome Project

Dr. Lita Proctor reported on the goals and outcomes of the Human Microbiome Project's (HMP) from FY2007-2016. Dr. Proctor discussed the major findings of the HMP, which identified the host compositions of microbiomes across a diverse set of body locations and disease cohorts, analyzed multi-omic traits across cohort studies, and made these data available to the scientific community. A video of this presentation can be found at: https://www.youtube.com/watch?v=ldjZFSyGK-Q.

REPORT – Recommendations from the 2017 eMERGE and Beyond Workshop

Dr. Sharon Plon discussed the recommendations from the 2017 eMERGE and Beyond Workshop. Dr. Plon summarized the goals of eMERGE III and the discussions at the eMERGE

and Beyond Workshop, which focused on identifying emerging issues and opportunities for using electronic phenotyping in genomics research. A video of this presentation can be found at: https://www.youtube.com/watch?v=l8cyRzU1zA4. Additionally, the workshop video and executive summary can be found here: https://www.genome.gov/27569445/emerge--beyond-the-future-of-electronic-medical-records-and-genomics/.

PRESENTATION – NCBI in a Data-Enabled World

Dr. James Ostell reported on the current activities and future plans of the National Center for Biotechnology Information (NCBI). Dr. Ostell highlighted efforts to improve the design and user interface of NCBI tools, as well as a commitment to integrate cloud technologies. Dr. Ostell also noted that NCBI is critically studying the question of whether all data should be preserved indefinitely, or whether there comes a point when some data should be stored in lower-cost formats or removed from a database entirely. A video of this presentation can be found at: https://www.youtube.com/watch?v=iQh9pPpvkrw.

REVIEW OF THE STATEMENT OF UNDERSTANDING BETWEEN NACHGR AND NHGRI

Dr. Graham reviewed the Statement of Understanding (SOU), which is reviewed annually at the February NACHGR meeting. This document details the responsibilities and authorities of the NACHGR and the NHGRI staff, including the types of applications and actions that must be brought to the Council for review and approval. Dr. Graham noted three changes to the SOU. The Statement of Understanding was unanimously approved by the Council. A video of this discussion can be found at: https://www.youtube.com/watch?v=jw8zU1axAAw

COUNCIL-INITIATED DISCUSSION

A video of the Council-initiated discussion can be found at: https://www.youtube.com/watch?v=2hMCcd0n6N8

ANNOUNCEMENTS AND ITEMS OF INTEREST

Three reports from NHGRI's national liaison societies were provided this round:

National Society of Genetic Counselors Quarterly Update International Society of Nurses in Genetic 2017 Annual Report American Society of Human Genetics Report to Council

CONFIDENTIALITY AND CONFLICT OF INTEREST

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

REVIEW OF APPLICATIONS₁

In the Closed Session, the NACHGR reviewed 126 applications, requesting \$69,802,847 (total cost). The applications included: 68 research project applications (R01, R03, R21 or RM1); 4 cooperative agreement applications (U24); 10 ELSI applications (R01, R03, R21, K01, or K99/R00); 3 conference application (R13); 2 career transition award applications (K99/R00); 2 institution training application (T32); 16 SBIR Phase I applications (R43); 9 SBIR Phase II applications (R44); 5 STTR Phase 1 applications (R41); 6 Research Education applications (R25), and 1 clinical investigator application (K08). A total of 91 applications totaling \$40,533,006 were recommended by the NACHGR.

This Council Meeting Summary document was prepared by Erin Currey, Scientific Program Analyst at NHGRI.

05/23/2018	<u>Rudy</u>
<u>Pozzatti</u>	
Date	Rudy Pozzatti, Ph.D. Executive Secretary National Advisory Council for Human Genome Research
05/23/2018 Green	<u>Eric D.</u>
Date	Eric Green, M.D, Ph.D. Chairman National Advisory Council for Human Genome Research

This report was approved by the NACHGR on May 21, 2018.

¹ For the record, it is noted that to avoid a conflict of interest, Council members absent themselves from the meeting when the Council discusses applications from their respective institutions or in which a conflict of interest may occur. Members are asked to sign a statement to this effect. This does not apply to "en bloc" votes.