

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
February 11-12, 2019

The Open Session of the 85th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, February 11, 2019 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 – 5:00 PM on February 11, 2019. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 8:00 AM – 10:00 AM on February 11, 2019; and from 8:30 AM until adjournment on February 12, 2019 for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT:

Jeffrey Botkin (University of Utah)
Brenton Graveley (University of Connecticut)
Jonathan Haines (Case Western Reserve University)
Gail Henderson (University of North Carolina)
Trey Ideker (University of California, San Diego)
Mark Johnston (University of Colorado)
Sharon Plon (Baylor College of Medicine)
Jonathan Pritchard (Stanford University)

AD HOC ADVISORS PRESENT:

Wendy Chung (Columbia University)
Mark Craven (University of Wisconsin)
Patricia Deverka (Geisinger Health System)
Stephen Fodor* (13.8, Inc.)
Rafael Irizarry (Dana-Farber Cancer Institute)
E. Albert Reece (University of Maryland)
Aviv Regev (Broad Institute of Harvard & MIT)
Chuan He (University of Chicago)
James Neaton (University of Minnesota)
Stephen Wisniewski (University of Pittsburgh)

**indicates participation by phone*

STAFF PRESENT FROM THE NATIONAL INSTITUTES OF HEALTH

Gabriel Batzli, ERP	Alvaro Encinas, DPCE
Joy Boyer, ERP	Elise Feingold, ERP
Comfort Browne, ERP	Adam Felsenfeld, ERP
Eileen Cahill, ERP	Ann Fitzpatrick, DM
Linda Chadwick, ERP	Robert Fullem, ERP
Christine Chang, ERP	Prabarna Ganguly, DPCE
Joanna Chau, ERP	Sylvia Garvey, ERP
Michelle Cohen, ERP	Tina Gatlin, ERP
Christina Daulton, DPCE	Elena Ghanaim, DPCE
Jyoti Dayal, ERP	Bettie Graham, ERP
Valentina Di Francesco, ERP	Linda Hall, ERP
Carla Easter, DPCE	Lucia Hindorff, ERP

Rebecca Hong, DPCE
Carolyn Hutter, ERP
Sonya Jooma, DPCE
Britny Kish, DPCE
Rongling Li, ERP
Nicole Lockhart, ERP
Ebony Madden, ERP
Allison Mandich, IOD
Teri Manolio, ERP
Mukul Nerurkar, DPCE
Michael Pagan, ERP
Kiara Palmer, DPCE
Mike Pazin, ERP
Natalie Pino, ERP

Erin Ramos, ERP
Jessica Reusch, DPCE
Laura Rodriguez, DPCE
Ellen Rolfes, DM
Jill Saletta, ERP
Michael Smith, ERP
Taylorlyn Stephan, ERP
Jeff Struewing, ERP
Michelle Tallman, ERP
Jennifer Troyer, ERP
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP

OTHERS PRESENT FOR ALL OR A PORTION OF THE MEETING:

Ellen Giarelli, International Society of Nurses in Genetics (remote participation)
Mona Miller, American Society for Human Genetics
Rhonda Schonberg, National Society of Genetic Counselors
Michael Watson, American College of Medical Genetics and Genomics

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 September 2018 Minutes by a unanimous vote.

FUTURE NACHGR MEETING DATES

- 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/27572638/february-2019-council-agenda-and-documents/>

PRESENTATION – *A Clinical Imperative: Genomics, Population Health, and Precision Health at Geisinger – Huntington Willard*

Dr. Huntington Willard gave a presentation on the precision health program at Geisinger Health System. Building off their success with the [MyCode Community Health Initiative](#),

Geisinger now offers clinical exome sequencing/screening to all patients in their health system. In the future, they plan to implement their program on a national scale in collaboration with other health systems that are committed to the vision of genome-based population health screening to improve patients' health and healthcare over the lifespan.

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

<https://www.youtube.com/watch?v=G84m3YSuRkQ>

CONCEPT CLEARANCE – *Electronic Medical Records and Genomics (eMERGE): Comprehensive Genomic Risk Assessment and Management – Teri Manolio*

Dr. Teri Manolio presented the eMERGE renewal concept. This concept proposes to use biorepositories linked to electronic medical records (EMR) to develop, implement, and disseminate genomic risk assessment and management tools for clinical use. Recent advances, such as polygenic risk scores (PRS) for multiple conditions, professional guidelines for clinical use of actionable variants in high-risk individuals, and automated tools for patient-driven collection of family history information, make a study like this feasible. The study will retrospectively validate and adapt PRS to EMR-defined phenotypes. It will also recruit 20,000 participants and use a single network-wide protocol to assess risk, make clinical management recommendations, and ascertain outcomes. Three funding opportunity announcements are proposed: one for a Coordinating Center, one for Clinical Sites that would recruit a minimum of 35% participants from underserved or underrepresented minorities, and a third for Clinical Sites of enhanced diversity that would recruit a minimum of 75% participants from underserved or underrepresented minorities.

Council approved the concept, with one member abstaining. A video of Dr. Manolio's presentation can be found at:

<https://www.youtube.com/watch?v=mnySRB-8qhM>

CONCEPT CLEARANCE – *Centers of Excellence in ELSI Research (CEER)– Joy Boyer*

Joy Boyer presented the concept for a limited competition re-issue of the CEER Request for Applications (RFA). The CEER program funds projects that use interdisciplinary approaches to investigate ELSI research questions related to genetics and genomics. This RFA will limit the growth of the CEER program and thereby allow flexibility for NHGRI to develop new initiatives based on the outcome of the 2020 strategic planning process.

Council approved the concept, with one member abstaining. A video of Joy Boyer's presentation can be found at:

<https://www.youtube.com/watch?v=v1PESxGYyj0>

REPORT – *NACHGR Genomic Data Science Working Group– Trey Ideker*

Dr. Trey Ideker gave an update from the NACHGR Genomic Data Science Working Group. The group facilitates a deeper engagement of NHGRI in issues at the interface between genomics and data science. It also advises the NHGRI Director and other NIH leaders about trans-NIH issues related to data science. The group played a key role in the release of three informatics PARs in 2018 and in the formation and launch of NHGRI's Analysis, Visualization and Informatics Labspace (AnVIL) cloud-based resource. Currently, the working group is helping to guide NHGRI's planning efforts for

data science and the ongoing NHGRI 2020 strategic planning process.

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

<https://www.youtube.com/watch?v=AjNGTXH-Wfc>

REPORT – NACHGR Community Engagement in Genomics Working Group (CEGWG) – Gwen Darien

Gwen Darien reported on the progress of the NACHGR CEGWG. The group is made up of community liaisons and health advocates representing diverse populations. It has worked to create dialogue and intersections between NHGRI and communities to ensure that genomics research benefits all. NIH programs such as All of Us and the 1000 Genomes Project have sought advice from the CEGWG. In the future, the group will continue to identify barriers to the broad and equitable adoption of genomic technologies among all communities through collaborative partnerships.

A video of Gwen Darien's presentation can be found at:

<https://www.youtube.com/watch?v=b34dE-i8klw>

REPORT – Clinical Genomics (ClinGen) Resource– Erin Ramos and Sharon Plon

Dr. Erin Ramos and Dr. Sharon Plon gave a joint presentation on the progress with the [ClinGen](#) resource. It was launched in 2013 with the goal of increasing data sharing and building an authoritative resource to define the clinical relevance of genes and genomic variants for use in medicine and research. Phase II began in 2017 and established gene curation expert panels that have published reclassified variants for multiple diseases.

A video of Dr. Ramos' and Dr. Plon's presentation can be found at:

<https://www.youtube.com/watch?v=VZghJ95Qfzo>

REPORT – NHGRI Triennial Inclusion Report– Christine Chang

Christine Chang gave the NHGRI Triennial Inclusion Report. All Institutes and Centers at NIH are mandated to ensure the inclusion of women and minority groups as participants in research projects as appropriate to the scientific question under study. Data from NHGRI-sponsored projects in 2016, 2017, and 2018 were presented, along with comparisons to data from earlier reporting periods. The Council voted unanimously to accept the report.

A video of Christine Chang's presentation can be found at:

<https://www.youtube.com/watch?v=NodGtPtFXh4>

COUNCIL-INITIATED DISCUSSION

Council members expressed concern about how Great Britain's exit from the European Union would affect the European Union's General Data Protection Regulation and genomics research. Council also raised the European Research Council's plan for an open access for publication initiative called "Plan S." NIH is monitoring these issues and NHGRI will provide information to the Council as developments occur. Other topics suggested by Council members for future reports included: how to handle functional re-interpretation of genomic data; examples of other precision health programs similar to the

Geisinger Health System, but with a more diverse population of patients; a review of the landscape of laboratory and clinical genomic testing; and a discussion about the field of implementation science.

A video of the Council-initiated discussion can be found at:
<https://www.youtube.com/watch?v=ZkhFjt3faGI>

CONFIDENTIALITY AND CONFLICT OF INTEREST¹

Dr. Rudy Pozzatti, NACHGR Executive Secretary 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 162 applications, requesting \$64,745,579 (direct costs). The applications included: 115 research project applications (R01, R03, R15, R21 or RM1); 6 cooperative agreement applications (U24); 9 career development or career transition applications (K99/R00, K01, or K08); 2 conference applications (R13); 2 institutional training applications (T32); 21 SBIR Phase I applications (R43); 5 SBIR Phase II applications (R44); 1 STTR Phase 1 application (R41) and 1 STTR Phase 2 application. A total of 104 applications totaling \$41,981,210 were recommended for funding by the NACHGR.

This Council Meeting Summary document was prepared by Gabriel Batzli, NHGRI Scientific Program Analyst.

5/28/2019
Date

Rudy Pozzatti
Rudy Pozzatti, Ph.D.
Executive Secretary
National Advisory Council for Human Genome Research

5/28/2019
Date

Eric Green
Eric Green, M.D, Ph.D.
Chairman
National Advisory Council for Human Genome Research

This report was approved by the NACHGR on May 20, 2019.

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.