The Open Session of the 86th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, May 20, 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 – 3:00 PM on May 20, 2019. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 8:30 AM – 10:00 AM on May 20, 2019 and from 8:30 AM until adjournment on May 21, 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)
Joshua Denny (Vanderbilt University)
Harry Dietz (Johns Hopkins University)
Stephen Fodor (13.8, Inc.)
Rafael Irizarry (Dana-Farber Cancer Institute)
Stephen Rich (University of Virginia)

*indicates participation by phone

STAFF PRESENT FROM NHGRI
Sarah Bates, DPCE
Gabriel Batzli, ERP
Vence Bonham, IOD
Joy Boyer, ERP
Comfort Browne, ERP
Devon Bumbray-Quarles, DEO
Alicia Caffi, DM
Eileen Cahill, ERP
Linda Chadwick, ERP
Christine Chang, ERP
Joanna Chau, ERP
Monika Christman, ERP
Michelle Cohen, ERP
Christina Daulton, DPCE
Jyoti Dayal, ERP
Valentina Di Francesco, ERP
Adam Felsenfeld, ERP
Ann Fitzpatrick, DM
Robert Fullem, ERP
Prabarna Ganguly, DPCE
Sylvia Garvey, ERP
Tina Gatlin, ERP
Elena Ghanaim, DPCE
Bette Graham, ERP
Linda Hall, ERP
Lucia Hindorff, ERP
Carolyn Hutter, ERP
Rongling Li, ERP
Nicole Lockhart, ERP
Ebony Madden, ERP
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: [link]

APPROVAL OF MINUTES
The Council approved the February 2019 Minutes by a unanimous vote.

FUTURE NACHGR MEETING DATES
- Aug. 13, 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/event-calendar/86th-Meeting-of-National-Advisory-Council-for-Human-Genome-Research

REPORT – ENCODE Project Update – Dr. Brenton Graveley
Dr. Graveley reviewed the progress of the Encyclopedia of DNA Elements (ENCODE) Project. ENCODE aims to catalogue the functional non-coding elements of the genome and is currently in its fourth phase, which will be completed in 2021. In previous phases of ENCODE, the functional elements of model organisms were also catalogued. All data generated by ENCODE is processed and analyzed using standardized pipelines, and is publicly available through the ENCODE portal. The ENCODE Consortium is actively
engaged in community outreach and training and follows a policy of rapid pre-publication release of data. Since its inception, ENCODE has been cited in over 2,200 publications by the research community. This spring, ENCODE was announced as being newly available as an Amazon Web Services (AWS) public data set to enable cloud-based analyses.

A video of Dr. Graveley’s presentation can be found at: [link]

**CONCEPT CLEARANCE – Diversity Action Plan PAR Renewal – Dr. Tina Gatlin**

Dr. Gatlin presented a concept clearance on the Diversity Action Plan (DAP) PAR renewal. Currently, there are twelve NHGRI-funded DAP awards covering the fields of genome sciences, genomic medicine, and ELSI research. There are no major changes in the proposed DAP PAR compared to the previous version. Previously, the DAP PAR limited institutions to one DAP award. In the next phase, institutions will instead be limited to one DAP award per scientific area.

Council approved the concept, with one member abstaining. A video of Dr. Gatlin’s presentation can be found at: [link]

**REPORT – NHGRI Strategic Planning Workshop: From Genome to Phenotype – Genomic Variation Identification, Association, and Function in Human Health and Disease – Dr. Sharon Plon**

Dr. Plon provided a summary of the January 22-24, 2019 NHGRI Strategic Planning Workshop. Details of this workshop are available online, including links to a workshop report and recordings of all presentations. The workshop was attended by over 150 individuals and focused on the link between the genome and phenotype. Discussions centered on seeking bold ideas for future projects, the landscape of current knowledge and capabilities, and key opportunities and collaborations that NHGRI should pursue. Four central themes emerged during the workshop: variant discovery and interpretation, basic research predicting clinicals needs, functional consequences of genomic variation, and technology and data resources supporting research. A number of short-term and long-term goals were recommended for NHGRI to consider.

A video of Dr. Plon’s presentation can be found at: [link]

**COUNCIL-INITIATED DISCUSSION**

Council members inquired about the status of cloud-based data commons being established by several NIH institutes in a trans-NIH fashion. Specifically, there was interest in understanding interactions among these projects to ensure interoperability and non-duplicative efforts. NHGRI is active in trans-institute working groups to ensure that NHGRI’s Analysis, Visualization, and Informatics Lab-space (AnVIL) is interoperable with other institutes’ data commons. Council noted the likelihood that commercially produced data from healthcare settings and direct-to-consumer testing companies will soon exceed the amount of genomic data produced by NIH-funded research. As such, exploring ways to allow access to this data for public research purposes will be key. NHGRI has actively supported the Global Alliance for Genomics and Health (GA4GH), which is a policy-framing and technical standards-setting organization, seeking to enable responsible genomic data sharing. Additionally, NHGRI-funded programs [such as the Clinical
Genome Resource (ClinGen) have had success partnering with clinical testing labs to promote public data sharing. GA4GH and ClinGen are also both working to address another issue raised by Council which concerns the lack of consistent canonical transcript identification. Lastly, Council members voiced their concerns with the lack of NIH standards for metadata formatting and sharing. NHGRI has recently established a Core Expectations for Metadata/Phenotypic Data Working Group that is assessing the current landscape of metadata standards and sharing practices. All of Us Research Project Director Eric Dishman was suggested as a guest speaker for a future NHGRI Council meeting.

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

CONFIDENTIALITY AND CONFLICT OF INTEREST

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

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 238 applications, requesting $81,715,785 (direct costs). The applications included: 181 research project applications (R01, R03, R15, R21 or R35); 11 cooperative agreement applications (U13 or U24); 8 career development or career transition applications (K99/R00, K01, or K08); 3 conference applications (R13); 3 education (R25) applications; 23 SBIR Phase I applications (R43); 6 SBIR Phase II applications (R44); 2 STTR Phase 1 applications (R41) and 1 STTR Phase 2 application (R42). A total of 143 applications totaling $48,384,895 were recommended for funding by the NACHGR.

This Council Meeting Summary document was prepared by Robert Fullem, NHGRI Scientific Program Analyst.

9/17/2019
Date

Rudy Pozzatti

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

9/17/2019
Date

Eric D. Green

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

This report was approved by the NACHGR on September 16, 2019.

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.