

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

May 16, 2016

The Open Session of the 77<sup>th</sup> meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, May 16, 2016 at the Fishers Lane Terrace Level Conference Center in Rockville, MD. 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 2:00 PM on May 16, 2016. 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 2:00 PM to 6:00 PM on May 16, 2016 for the review, discussion, and evaluation of grant applications.

Council members present:

Eric Boerwinkle  
Lon Cardon  
Joseph Ecker  
Chanita Hughes-Halbert  
Howard Jacob  
Robert Nussbaum  
Lucila Ohno-Machado  
Arti Rai  
Carol Bult, ad hoc  
Brenton Graveley, ad hoc  
Gail Henderson, ad hoc  
Mark Johnston, ad hoc  
Jonathan Pritchard, ad hoc  
Dan Roden, ad hoc  
Val Sheffield, ad hoc  
David Walt, ad hoc  
Jay Shendure, ad hoc (by webcast and telephone)

Staff from the National Human Genome Research Institute:

Ronit Abramson, DPCE	Kim Ferguson, ERP
Vence Bonham, IOD and IRP	Ann Fitzpatrick, DM
Joy Boyer, ERP	Colette Fletcher-Hoppe, ERP
Larry Brody, ERP and IRP	Tina Gatlin, ERP
Comfort Browne, ERP	Peter Good, ERP
Christine Chang, ERP	Bettie Graham, ERP
Monika Christman, ERP	Jyoti Gupta, ERP
Julie Coursen, ERP	Linda Hall, ERP
Valentina di Francesco, ERP	Lucia Hindorff, ERP
Cecilia Dupecher, ERP	Rebecca Hong, DPCE
Carla Easter, DPCE	Carolyn Hutter, ERP
Brenda Iglesias, ERP	Sonya Jooma, DPCE
Alex Lee, ERP	Ashley Lewis, DPCE
Kevin Lee, ERP	Rongling Li, ERP
Jon Lotempio, Jr., ERP	Nicole Lockhart, ERP
Elise Feingold, ERP	Ebony Madden, ERP
Adam Felsenfeld, ERP	Allison Mandich, IOD

Casey Martin, ERP  
Jean McEwen, ERP  
Keith Mckenney, ERP  
Donna Messersmith, DPCE  
Hannah Naughton, ERP  
Annie Niehaus, ERP  
John Ohab, DPCE  
Teri Manolio, ERP  
Mike Pazin, ERP  
Ajay Pillai, ERP  
Lita Proctor, ERP  
Erin Ramos, ERP  
Jeffery Schloss, ERP

Elle Silverman, ERP  
Michael Smith, ERP  
Heidi Sofia, ERP  
Jeffery Struewing, ERP  
Simona Volpi, ERP  
Vivian Ota Wang, ERP  
Chris Wellington, ERP  
Kris Wetterstrand, IOD  
Bob Wildin, DPCE  
Rosann Wise, DPCE  
Lu Wang, ERP  
Kira Wong, ERP

Others present for all or a portion of the Open Session:

Siobhan Addie, National Academy of Sciences, Engineering, and Medicine  
Adam Fagan, Genetics Society of America  
Alice Kuaban, American Society of Hematology  
James O'Leary, Genetic Alliance  
Emily Poston, Genetics Society of America  
Rhonda Schonberg, National Society of Genetic Counselors  
Michael Watson, American College of Medical Genetics

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

**APPROVAL OF MINUTES FOR THE FEBRUARY 8-9, 2016 MEETING**

The Council members unanimously approved and accepted the Minutes from the February 8-9, 2016 Advisory Council meeting.

**FUTURE MEETING DATES**

Sept. 12-13, 2016	Sept. 11-12, 2017
Feb. 6-7, 2017	February 12-13, 2018
May 8-9, 2017	May 21-22, 2018

**DIRECTOR'S REPORT**

Dr. Eric Green presented the Director's Report to Council.

Council inquired about the relationship between the Precision Medicine Initiative (PMI) and the Million Veteran Program (MVP). Dr. Carolyn Hutter, a member of ERP staff, commented that the PMI and MVP are in collaboration and have been working with the Veteran's Administration to re-consent individuals. Robert Nussbaum, NHGRI Council member, is on an advisory committee for the MVP and suggested that someone from the program come speak at a future Council meeting to discuss the program.

**PRESENTATION FROM NEW NIMHD DIRECTOR**

Dr. Eliseo Perez-Stable, the new Director of the National Institute on Minority Health and Health Disparities (NIMHD), gave a presentation on the goals and objectives of NIMHD-sponsored research and areas of common interest with NHGRI. The overarching mission of NIMHD is to

lead and support scientific research that advances our understanding of minority health and health disparities.

Council inquired about outreach to potential research subjects, and if NIMHD has found effective ways to encourage minority participation in research projects. Dr. Perez-Stable clarified that research involving human subjects should always include minorities, regardless of the topic of research. Research that is focused on a minority health question may be within one minority group or among several groups. Health disparity research additionally includes individuals with a socio-economic disadvantage. Dr. Perez-Stable has presented this perspective at several other Institute/Centers (IC) Council meetings, and there have been no fundamental disagreements with these principles of inclusion in research. He noted that several ICs have supported research on recruitment science and improving minority participation in research for a number of years, and it might be a good time to revisit some of the best practices that were developed from those studies.

Council commented on the cost of applying new technologies to genomic medicine, which will expand as the Precision Medicine Initiative develops. Council expressed the concern that access to new genomic medicine technologies may be limited to individuals who are wealthy, or have access to high quality health insurance. Dr. Perez-Stable Eliseo noted that when electronic health records were initially developed, usage by minorities lagged behind individuals of high socioeconomic status. However, over time that gap has narrowed. It's possible that access to genomic medicine technology may follow a similar pattern. But Dr. Perez-Stable agreed it is a legitimate concern that a technology divide could worsen health disparities for minorities in the domain of genomic medicine. If the cost of any new therapeutic is prohibitively high, then solutions will involve government regulation, negotiations with the provider, and knowledge learned during the research that led to the new therapy.

Council was excited about the vision for NIMHD-sponsored research, and asked if investigators would be able to pursue research questions by receiving R01 grants. Dr. Perez-Stable noted there is a community of investigators who study minority health research questions, but they have not viewed NIMHD as a funding source because R01 awards have not been made in the past. He is hoping to change this, and NIMHD is now putting out program announcements to support R01-based research. Council also wanted to know if Community-Based Participatory Research (CBPR) Centers would continue to have a role in the future of NIMHD. Dr. Perez-Stable supports CBPR, and noted that NIMHD has been the place where CBPR has thrived at NIH. Overall, he would like to see a more balanced distribution of R01 grants and Centers where CBPR research can be conducted.

Council asked Dr. Perez-Stable to comment on the relationship between self-identified race/ethnicity and the use of genomic markers developed as determinants of population ancestry, and which is more appropriate to use when conducting genetic/genomic research. Some research grants like the Clinical Sequencing Exploratory Research (CSER) program will focus on under-represented populations, and they will need to use objective measures to recruit and define these groups. Dr. Perez-Stable said that NIMHD is partnering with Vence Bonham at NHGRI to develop an agenda for a workshop that will be held this fall to explore this issue, as well as other topics related to minority health and minority participation in research. In the early years of the Human Genome Project, race was defined by social scientists as a social construct. Since that time, ancestral markers have been developed that can be used as a tool to define population ancestry and to study admixture within specific populations. For some investigators, this has clouded the discussion about race as a social construct. Dr. Perez-Stable noted that he will always view race to be a social construct, and he sees the ancestral markers as useful tools

that can be used to guide research. More research, and more discussion is needed to help resolve the question of how racial/ethnic groups should be defined for assigning individuals to different groups in the context of NIH-sponsored research projects.

Vence Bonham concluded the discussion by announcing that he and Larry Brody are leading the planning of an NIMHD-NHGRI meeting on the topic, "The Use of Race and Ethnicity in Genomic and Biomedical Research." Comments or questions about this meeting should be referred to Vence and Larry.

### **COUNCIL-INITIATED DISCUSSION**

Council suggested having representatives from the FDA come back and present at a future Council meeting. They noted that a specific issue to address would be regulatory hurdles as potential barriers to the implementation of genomic medicine. Council suggested asking the FDA to discuss the Precision Medicine Initiative (PMI) and how their views may/may not change as PMI is implemented.

Cristina Kapustij, the NHGRI Policy and Program Analysis Branch Chief, commented on NHGRI's ongoing discussions with the FDA. The Policy and Program Analysis Branch and the Division of Policy, Communications, and Education at NHGRI have a regularly scheduled monthly meeting with the PMI team. On June 10<sup>th</sup>, there is a workshop scheduled on Investigational Device Exemptions (IDEs) to help grantees work through the process of whether or not their research includes a significant risk device and how to work through the process. They continue to collect input from NHGRI and its grantees. They are also working on regulations for Laboratory Developed Tests (LDTs), but nothing is set in place at the moment. NHGRI will see how this plays out in the next few months and have this on the list for discussion.

Lastly, Council members commented on the parallel and converging activities between NHGRI and the National Heart, Lung, and Blood Institute (NHLBI). It was suggested to invite either Dr. Gary Gibbons, or a senior staff member from NHLBI's Trans-Omics for Precision Medicine (TOPMed) program to speak at a future Council meeting, and to encourage more communication between the two Councils.

### **ANNOUNCEMENTS AND ITEMS OF INTEREST**

Dr. Pozzatti referred Council to two reports: the American Society of Human Genetics Report and the National Society of Genetic Counselors Report. These are descriptions of the activities in which these societies have been engaged since the February 8-9, 2016 Council Meeting.

### **CONFIDENTIALITY AND CONFLICT OF INTEREST**

Dr. 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<sup>1</sup>

In the Closed Session, the Council reviewed 142 applications, requesting \$64,913,050 (total cost). The applications included: 79 research project applications, 10 cooperative agreement (U01 or UM1) applications, 13 ELSI applications (11 R-series and 2 career development), 2 research center applications (U41), 2 conference applications (R13), 4 career transition award applications (K99/R00), 22 SBIR Phase I applications (R43), 3 SBIR Phase II applications (R44), 2 STTR Phase 1 applications (R41), and 5 Research Education (R25) applications. A total of 91 applications totaling \$35,664,731 were recommended by the Council.

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Date

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

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Date

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

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<sup>1</sup> 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.