

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH**  
**MEETING SUMMARY**  
September 11-12, 2017

The Open Session of the 81<sup>st</sup> meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, September 11, 2017, 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 5:15 PM on September 11, 2017. 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 5:15 PM to 6:30 PM on September 11, 2017, and from 8:30 AM until adjournment on September 12, 2017, for the review, discussion, and evaluation of grant applications.

Council Members Present:

Eric Boerwinkle  
Carol Bult  
Wendy Chung  
Patricia Deverka  
Brenton Graveley  
Jonathan Haines  
Gail Henderson  
Trey Ideker  
Raphael Irizarry  
Mark Johnston  
Sharon Plon  
Aviv Regev  
Dan Roden  
Val Sheffield  
Jay Shendure

Staff Present from the National Human Genome Research Institute:

Gabriel Batzli, ERP	Linda Hall, ERP
Vence Bonham, IOD and IRP	Lucia Hindorff, ERP
Joy Boyer, ERP	Rebecca Hong, DPCE
Comfort Browne, ERP	Ellen Howerton, ERP
Christine Chang, ERP	Carolyn Hutter, ERP
Michelle Cohen, ERP	Sheethal Jose, ERP
Heather Colley, ERP	Melpi Kasapi, ERP
Erin Curry, ERP	Alexander Katz, DIR
Jyoti Dayal, ERP	Jeff Kim, ERP
Valentina di Francesco, ERP	Jennifer Krupp, NIH-ACMG
Carla Easter, DPCE	Rongling Li, ERP
Adam Felsenfeld, ERP	Nicole Lockhart, ERP
Robert Fullem, ERP	Allison Mandich, IOD

Tina Gatlin, ERP  
Bettie Graham, ERP  
Vivian Ota Wang, ERP  
Michael Pagan, ERP  
Kiara Palmer, DPCE  
Mike Pazin, ERP  
Robb Rowley, DIR  
Laura Skow, ERP  
Mike Smith, ERP  
Heidi Sofia, ERP

Jean McEwan, ERP  
Sam Moore, ERP  
Taylorlyn Stephan, ERP  
Barbara Thomas, ERP  
Elizabeth Tuck, DPCE  
Lu Wang, ERP  
Cara Weismann, DPCE  
Chris Wellington, ERP  
Kris Wetterstrand, IOD  
Ken Wiley, ERP

Others Present for All or a Portion of the Meeting:

Rex Chisholm, Northwestern University  
Ellen Giarelli, Council Liaison Member, International Society of Nurses in Genetics  
Shoshera Kahara, NIH/OD  
Rachel Levinson, Arizona State University  
Mona Miller, ASHG  
Derek Scholes, ASHG  
Rhonda Schonberg, NSGC  
Tara Schwetz, NIH/OD  
Lawrence Tabak, NIH/OD

Materials (e.g., agenda and presentations) for the Open Session of the 81<sup>st</sup> NACHGR meeting are available at: <https://www.genome.gov/27569449/september-2017-council-agenda-and-documents/>). A recorded video is available at: <https://www.youtube.com/playlist?list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ>.

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

Dr. Rudy Pozzatti began the Open Session with the introduction of new NHGRI staff, liaisons, and guests, which can be found at: <https://www.youtube.com/watch?v=zOy4FfP8gA&index=1&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ>

**APPROVAL OF MINUTES FOR THE MAY 2017 NACHGR MEETING**

The Council approved the Minutes by unanimous vote without any edits or changes.

**FUTURE MEETING DATES**

- Feb. 12-13, 2018
- 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. A video of his presentation can be found at: <https://www.youtube.com/watch?v=2EIR3l6B9ns&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=2>

**SPECIAL PRESENTATION** – Dr. Eric Green presented a U.S. flag to NACHGR member Dr. Aviv Regev, to commemorate her becoming a citizen of the United States of America. A video of this presentation can be found at: <https://www.youtube.com/watch?v=9cTuDemANUM&index=3&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ>

### **PRESENTATION** – *Next Generation Researchers Initiative*

Dr. Lawrence Tabak presented on the Next Generation Researchers Initiative. A video of this presentation can be found at: [https://www.youtube.com/watch?v=O3jxAN3j\\_P4&index=4&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ](https://www.youtube.com/watch?v=O3jxAN3j_P4&index=4&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ). Dr. Tabak spoke about the challenges of maintaining a highly diversified workforce of biomedical investigators in an era of hyper-competition to obtain NIH grant support. His talk focused on NIH's current efforts to provide funding for both early-career and mid-career investigators.

### **REPORT** – *Update on the eMERGE Network*

Dr. Rex Chisholm made a presentation about the Electronic Medical Records and Genomics (eMERGE) Network. A video of this presentation can be found at: <https://www.youtube.com/watch?v=5MsEX5b3Tgo&index=5&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ>. Dr. Chisholm presented an overview of the research activities and accomplishments of the eMERGE Network over its three funding cycles that date back to 2007. In phase 1, eMERGE investigators explored the challenges of using electronic health records to conduct genomics research. In phase 2, investigators examined how genomic findings could be applied to clinical care. In phase 3, investigators are using genome sequencing to improve genomic discovery and advance genomic medicine implementation.

### **REPORT** – *The Cancer Genome Atlas (TCGA): A Decade of Discovery*

Dr. Carolyn Hutter reported on TCGA. A video of this presentation can be found at: [https://www.youtube.com/watch?v=zbd\\_p8MaQlc&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=6](https://www.youtube.com/watch?v=zbd_p8MaQlc&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=6). Dr. Hutter presented a summary of the achievements of The Cancer Genome Atlas program – a ten-year collaboration involving NHGRI and the National Cancer Institute.

## **REPORT – National Academy of Medicine Report on Evidence Framework for Genetic Testing**

Dr. Wendy Chung discussed the NAM Report on Evidence Framework for Genetic Testing. A video of this presentation can be found at:

<https://www.youtube.com/watch?v=fCqDX6hluR4&index=7&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ>. Dr. Chung summarized a report prepared by a working group of the National Academy of Medicine. The group was charged with reviewing the scientific literature on the evidence base for different types of genetic testing; to make recommendations to advance the evidence base to improve patient care; and to recommend a framework to be used for decision-making regarding the use of genetic tests.

## **REPORT – Genomic Medicine Working Group Activities in 2017**

Dr. Teri Manolio reported on the Genomic Medicine Working Group activities in 2017. A video of this presentation can be found at:

[https://www.youtube.com/watch?v=o\\_ZT776jqPs&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=8](https://www.youtube.com/watch?v=o_ZT776jqPs&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=8). Dr. Manolio gave an update on the activities of the Genomic Medicine Working Group over the past year.

## **REPORT – Update on the NSIGHT Program**

Dr. Anastasia Wise gave an update about the Newborn Sequencing in Genomic Medicine and Public Health (INSIGHT) program. A video of this presentation can be found at:

<https://www.youtube.com/watch?v=gZ8FrrHqTIU&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=9>. Dr. Wise summarized the progress that has been made by the investigators who are participating in the NSIGHT program.

## **COUNCIL-INITIATED DISCUSSION**

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

[https://www.youtube.com/watch?v=aBSmb\\_xHjqc&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=10](https://www.youtube.com/watch?v=aBSmb_xHjqc&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=10)

## **ANNOUNCEMENTS AND ITEMS OF INTEREST**

A report was sent to NHGRI from the National Society of Genetic Counselors describing their recent activities. This report and a full listing of the Open Session agenda can be found at:

<https://www.youtube.com/watch?v=VgPdgvzNBAI&list=PL1ay9ko4A8snp23vGcZ2knqilGG993vSQ&index=11>

## **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 NACHGR reviewed 131 applications, requesting \$62,334,463 (total cost). The applications included: 76 research project applications (R01, R03, R15 or R21); 2 cooperative agreement applications (U01); 11 ELSI applications (R01, R21, or K01); 3 research center applications (U41); 3 conference application (R13); 5 career transition award applications (K99/R00); 1 institution training application (T32); 10 SBIR Phase I applications (R43); 12 SBIR Phase II applications (R44); 4 STTR Phase 1 applications (R41); 1 STTR Phase II application (R42), 2 Research Education applications (R25), and 1 clinical investigator application (K08). A total of 77 applications totaling \$29,576,811 were recommended by the NACHGR.

This Council Meeting Summary document was prepared by Laura Skow, Program Analyst at NHGRI.

2/23/2018

Date

*Rudy Pozzatti*

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

2/21/2018

Date

*Eric 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.