

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
May 18-19, 2020**

The Open Session of the 89th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened virtually at 12:30 PM on Monday, May 18, 2020 on Zoom. 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 12:30 PM – 4:30 PM on May 18, 2020. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 11:00 AM – 12:00 PM on May 18, 2020 and from 11:00 AM until adjournment on May 19, 2020 for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT:

Jeffrey Botkin (University of Utah)
Wendy Chung (Columbia University)
Mark Craven (University of Wisconsin)
Patricia Deverka (Deverka Consulting, LLC)
Harry Dietz (Johns Hopkins University)
Steve Fodor (13.8, Inc.)
Jonathan Haines (Case Western Reserve University)
Gail Henderson (University of North Carolina)
Trey Ideker (University of California, San Diego)
Rafael Irizarry (Dana-Farber Cancer Institute)
Sharon Plon (Baylor College of Medicine)
Stephen Rich (University of Virginia)

AD HOC ADVISORS PRESENT:

Howard Chang (Stanford University)
Lisa Parker (University of Pittsburgh)
Len Pennacchio (Department of Energy, Joint Genome Institute)
Olga Troyanskaya (Princeton University)

STAFF PRESENT FROM NHGRI

Sarah Bates, CPLB
Vence Bonham, IOD
Joy Boyer, ERP
Lawrence Brody, ERP
Lisa Brooks, ERP
Comfort Browne, ERP
Gloria Butler, DM
Alicia Caffi, DM
Eileen Cahill, ERP
Ron Carter, DM
Lisa Chadwick, ERP
Christine Chang, ERP
Joanna Chau, ERP
Monika Christman, ERP
Luis Cubano, ERP
Jennifer Czajkowski, DM

Jyoti Dayal, ERP
Edith DeHaut, ERP
Ernesto Del Aguila, CPLB
Valentina Di Francesco, ERP
Chris Donohue, CPLB
Argenia Doss, ERP
Carla Easter, ECIB
Jack Eidsness, DM
Laura Eisenman, ERP
Elise Feingold, ERP
Adam Felsenfeld, ERP
Kimberly Ferguson, ERP
Laurie Findley, ERP
Ann Fitzpatrick, DM
Colin Fletcher, ERP
Prabarna Ganguly, CPLB

Sylvia Garvey, ERP
Tina Gatlin, ERP
Elena Ghanaim, PPAB
Daniel Gilchrist, ERP
Bettie Graham, ERP
Chris Gunter, IOD
Linda Hall, ERP
Peggy Hall, ERP
Rebecca Hong, PPAB
Joe Henke, DM
Lucia Hindorff, ERP
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Kimberly Jacoby Morris, ECIB
Arnold Jang, DM
Cristina Kapustij, PPAB
Dave Kaufman, ERP
Jimmie Kees, ERP
Julie Kim, ERP
Natalie Kucher, ERP
Rongling Li, ERP
Darryl Leja, CPLB
Nicole Lockhart, ERP
Ebony Madden, ERP
Kerri Magerowski, ERP
Teri Manolio, ERP
Marcia Morris, ERP
Stephanie Morris, ERP
Ken Nakamura, ERP

Mukul Nerurkar, CPLB
Emily Neveux, DM
Briana Nuñez, ERP
Mike Pazin, ERP
Ajay Pillai, ERP
Natalie Pino, ERP
Erin Ramos, ERP
Alex Raphael, ERP
Ellen Rolfes, DM
Robb Rowley, ERP
Lorjetta Schools, ERP
Baergen Schultz, ERP
Shurjo Sen, ERP
Catherine Sillari, ERP
Michael Smith, ERP
Heidi Sofia, ERP
Jerryl Somani, DM
Taylorlyn Stephan, ERP
Jennifer Strasburger, ERP
Michelle Tallman, ERP
Cecelia Tamburro, ERP
Barbara Thomas, ERP
Jennifer Troyer, ERP
Susan Vasquez, IOD
Simona Volpi, ERP
Harry Wedel, 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
Katherine Lambertson, Genetic Alliance
Mona Miller, American Society of Human Genetics
Rhonda Schonberg, National Society of Genetic Counselors
Sharon Terry, Genetic Alliance

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 2020 Minutes by a unanimous vote.

FUTURE NACHGR MEETING DATES

- Sep. 14-15, 2020
- Feb. 22-23, 2021
- May 17-18, 2021
- Sep. 13-14, 2021

- Feb. 7-8, 2022
- May 16-17, 2022

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: [\[link\]](#)

CONCEPT CLEARANCE – *Research Experiences in Genomic Research for Genetic Counselors (PAR) – Dr. Luis Cubano*

Dr. Luis Cubano presented the concept for Research Experiences in Genomic Research for Genetic Counselors (PAR). This concept is intended to increase the number of genetic counselors in the research workforce, in response to the [NHGRI Training and Education Task Force Report](#) presented by NACHGR Member Dr. Wendy Chung at the February 2020 NACHGR Meeting. The proposed initiative will provide funding for R25 research education programs to train graduates of genetic counseling master's programs in genomics research and integrate them as core members of research groups. This proposal will fill a necessary niche in providing rigorous research opportunities and professional development, especially as genetic counselors become increasingly important in genomic medicine research.

NACHGR Members approved the concept unanimously. A video of Dr. Cubano's presentation and the related discussion can be found at: [\[link\]](#)

CONCEPT CLEARANCE – *Research Experiences in Genomic Research for Data Scientists (PAR) – Dr. Luis Cubano*

Dr. Luis Cubano presented the concept for Research Experiences in Genomic Research for Data Scientists (PAR), which is also a product of the recommendations from the [NHGRI Training and Education Task Force Report](#) to train graduates of data science master's programs in clinical and basic genomics research and increase the genomic data science research workforce. Similar to the counterpart concept for genetic counselors, grantee institutions will create genomics research training programs for master's-level data scientists. Examples of research projects include using genomic data analysis methods to find phenotypically relevant genomic variants and building researcher and clinician-friendly predictive models and data-visualization tools.

NACHGR Members approved the concept unanimously. A video of Dr. Cubano's presentation and the related discussion can be found at: [\[link\]](#)

CONCEPT CLEARANCE – *Technology Development for Single-Molecule Protein Sequencing (RFAs) – Dr. Tina Gatlin*

Dr. Tina Gatlin presented the Technology Development for Single-Molecule Protein Sequencing (SMPS) concept, designed to fund novel and high-risk investigator-initiated projects that will significantly accelerate innovation and technology development in the sequencing of single protein molecules. Recent advances in nanopore techniques and Edman chemistry have created a significant opportunity to develop new technologies that can facilitate low-abundance protein detection and single-cell analysis at high throughput. Such technologies would further genomics research, particularly in genotype-to-phenotype mapping, functional genomics, and multi-omic molecular diagnostics. Despite the significant risk associated with this concept,

advancement of single-molecule protein sequencing technology could potentially lower the cost and increase accessibility of proteomic data, thereby transforming proteomics in the same way that next-generation DNA sequencing revolutionized genomics.

NACHGR Members approved the concept unanimously. A video of Dr. Gatlin's presentation and the related discussion can be found at: [\[link\]](#)

CONCEPT CLEARANCE – Investigator-initiated Research on Genetic Counseling Processes and Practice (RFA) – Dr. Ebony Madden

Dr. Ebony Madden presented the Investigator-initiated Research on Genetic Counseling Processes and Practice concept. As genetic counselors become increasingly important in the era of genomic medicine, the shortage of genetic counselors and clinical geneticists will require more efficient strategies for delivering genetic testing results, particularly with the rise of telehealth in the post-COVID-19 era. NHGRI seeks to fund research projects to study the implementation and optimization of novel genetic counseling practices for genomic medicine. Research topics can vary in range – from implementation of technical solutions to increase genetic counseling bandwidth to developing methods for genetic counseling in medically underserved areas. Responsive applications would include research personnel experienced with challenges in genetic counseling and projects with generalizable findings.

NACHGR Members approved the concept unanimously. A video of Dr. Madden's presentation and the related discussion can be found at: [\[link\]](#)

COUNCIL-INITIATED DISCUSSION

Presentations requested at the February 2020 NACHGR Meeting will be postponed to the next in-person NACHGR Meeting. In addition to these pending presentations, NACHGR requested a program presentation from Genome Sequencing Program (GSP) summarizing the findings and outcomes from the last 5 years.

NACHGR inquired about NHGRI's administrative and scientific response to the COVID-19 pandemic. NHGRI is committed to working with grantees to manage the drastic impact of the pandemic on the research community. NIH-wide announcements and notices have been released to provide guidance on grants and applications (e.g., Dr. Mike Lauer's [blog](#)), and grantees are advised to refer to their program officers, local institution, and IRB for guidance on research activities (e.g., participant recruitment and engagement). NACHGR emphasized the need for NIH-wide policy on managing no-cost extensions, especially when research progress may be stalled for months. Additionally, grantees and program officers should be cognizant of making budgetary changes to their grants due to travel restrictions during the pandemic. On the research side, NHGRI has been involved in key discussions and coordination for COVID-19 data and research, including international collaborations.

NACHGR Members expressed their full support for the new initiatives to fund recent graduates of data science and genetic counseling master's programs, but they also noted that such funding opportunities should also be made available earlier in the educational pipeline to master's, undergraduate, and high school students. NACHGR requested a presentation on the various NIH educational and training programs and the types of trainees they support (e.g., T32 grants are limited to PhDs and MD/PhDs), so that NACHGR Members can better assess NIH-wide funding opportunities for trainees at all levels. NACHGR also recommended that NHGRI develop strategies to provide more

training opportunities with competitive funding for undergraduate and high school students, so that they may consider genomics research as a viable career option.

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

CONFIDENTIALITY AND CONFLICT OF INTEREST¹

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 247 applications, requesting \$84,984,683 (direct costs). The applications included: 203 research project applications (R01, R03, R15, R21, R35, or RM1); 6 cooperative agreement applications (U24); 7 career development or career transition applications (K99/R00 or K01); 3 conference applications (R13); 3 education applications (R25); 1 institutional training application (T32); 14 SBIR Phase I applications (R43); 9 SBIR Phase II applications (R44); and 1 STTR Phase 1 application (R41).

This Council Meeting Summary document was prepared by Joanna Chau, NHGRI Scientific Program Analyst.

9/28/2020

Date

Rudy Pozzatti

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

9/28/2020

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 14, 2020

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.