

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
February 10-11, 2020

The Open Session of the 88th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 AM on Monday, February 10, 2020 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 10, 2020. 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 February 10, 2020; and from 8:30 AM until adjournment on February 11, 2020 for the review, discussion, and evaluation of grant applications.

NACHGR MEMBERS PRESENT

Harry Dietz (Johns Hopkins University School of Medicine)
Jonathan Haines (Case Western Reserve University)
Jonathan Pritchard (Stanford University)*
Sharon Plon (Baylor College of Medicine)
Trey Ideker (University of California, San Diego)
Gail Henderson (University of North Carolina)*
Brent Graveley (University of Connecticut)*
Jeffrey Botkin (University of Utah)
Wendy Chung (Columbia University)
Patricia Deverka
Stephen Fodor (13.8, Inc.)
Rafael Irizarry (Dana-Farber Cancer Institute)

AD HOC ADVISORS PRESENT

Mark Craven (University of Wisconsin)
Stephen Rich (University of Virginia)

**indicates participation by phone*

STAFF PRESENT FROM THE NATIONAL INSTITUTES OF HEALTH

Matthew Arnegard, ORWH	Argenia Doss, ERP
Sarah Bates, DPCE	Jyoti Dayal, ERP
Vence Bonham, OD and IRP	Carla Easter, DPCE
Joy Boyer, ERP	Elise Feingold, ERP
Comfort Browne, ERP	Adam Felsenfeld, ERP
Larry Brody, ERP	Laurie Findley, ERP
Eileen Cahill, ERP	Ann Fitzpatrick, DM
Lisa Chadwick, ERP	Prabarna Ganguly, DPCE
Alicia Caffi, DM	Tina Gatlin, ERP
Christine Chang, ERP	Dan Gilchrist, ERP
Joanna Chau, ERP	Bettie Graham, ERP
Monika Christman, ERP	Eric Green, OD
Heather Colley, ERP	Chris Gunter, OD
Luis Cubano, ERP	Linda Hall, ERP
Christine Daulton, DPCE	Hwaida Hannoush, ERP
Valentina di Francesco, ERP	Lucia Hindorff, ERP

Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Britny Kish, DPCE
Danuta Krotoski, NICHD
Darryl Leja, DPCE
Rongling Li, ERP
Nicole Lockhart, ERP
Marie-Lusie Brennan, ERP
Ebony Madden, ERP
Teri Manolio, ERP
Donna Messersmith, DPCE
Kim Jacoby Morris, DPCE
Briana Nuñez, ERP
Melissa Parisi, NICHD
Mike Pazin, ERP
Natalie Pino, ERP
Robb Rowley, ERP
Lorjetta Schools, ERP
Baergen Schultz, ERP

Shurjo Sen, ERP
Jason Shockey, OD
Catherine Sillari, ERP
Mike Smith, ERP
Heidi Sofia, ERP
Taylorlyn Stephan, ERP
Tasha Stewart, ERP
Jennifer Strasburger, ERP
Tiancheng Sun, ERP
Michelle Tallman, ERP
Cecelia Tamburro, ERP
Jennifer Troyer, ERP
Simona Volpi, ERP
Harry Wedel, ERP
Chris Wellington, ERP
Kris Wetterstrand, OD
Ken Wiley, ERP
Guopin Yu, CSR

OTHERS PRESENT FOR ALL OR A PORTION OF THE MEETING

Austin Booth, Research Triangle Institute
Rich Haspel, Beth Israel Deaconess Medical Center
Samata Katta, American Society of Human Genetics
Rachel Levinson, Arizona State University
Mona Miller, American Society of Human Genetics
Rhonda Schonberg, National Society of Genetic Counselors

INTRODUCTION OF NEW NACHGR 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=HZQBw9G2NaM>.

APPROVAL OF MINUTES

NACHGR members approved the September 2019 minutes by a unanimous vote with no edits, opposition or abstentions.

FUTURE NACHGR MEETING DATES

- May 18-19, 2020
- Sept. 14-15, 2020
- Feb. 22-23, 2021
- May 22-23, 2021
- Sept. 13-14, 2021

DIRECTOR'S REPORT

Dr. Eric Green gave his Director's Report, which included a series of updates on recent activities and awards 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/88th-Meeting-of-National-Advisory-Council-for-Human-Genome-Research>.

PRESENTATION – NHGRI Training Task Force Report – Wendy Chung

NACHGR Member Dr. Wendy Chung presented the findings and recommendations on behalf of the NHGRI Training Task Force. The group considered five central principles for evaluating ERP's efforts in training in genomics, data analysis and interpretation, and ELSI issues. These included: diversity/inclusion, multi-disciplinary training, modular and just-in-time curriculum development, data and knowledge democratization, and foundational knowledge. The Task Force gathered input for their recommendations via conversations with representatives of the training community, an analysis of the existing NHGRI training portfolio, and collated feedback from NHGRI Strategic Planning meetings, among other sources. The Task Force recommended that NHGRI increase funding for existing training mechanisms while also developing and expanding training efforts in clinical genomics, data science, and bioinformatics, with particular focus on the inclusion and representation of women and minority groups. The Task Force also advised NHGRI to develop and disseminate modular training programs for various career levels to increase the flexibility of training programs as new methods and careers in genomics emerge.

A video of Dr. Chung's presentation and the related discussion can be found at: <https://youtu.be/lx-7PWXQZE>.

CONCEPT CLEARANCE – Consortium for Understanding the Impact of Genomic Variation on Genome Function (RFA) – Mike Pazin and Dan Gilchrist

Drs. Mike Pazin and Dan Gilchrist presented the concept for the Consortium for Understanding the Impact of Genomic Variation on Genome Function. The concept was developed in large part from discussions and feedback from the [From Genome to Phenotype Workshop](#) and addresses a key component of the anticipated 2020 NHGRI Strategic Plan: how genome variation affects genome function. The program would have five essential components: Functional Characterization Centers to test variant impact, Mapping Centers to map functional activity, Regulatory Network Projects to map genome regulatory networks, Predictive Modeling Projects to enhance prediction of variant impact, and a Data Coordination Center to connect and serve the consortium components and build a community resource. The consortium would build upon the successes of past research projects, like [ENCODE](#) and [LINCS](#), while also interfacing with a number of existing international initiatives, such as [IHEC](#), [HuBMAP](#), and the [Human Cell Atlas](#). NHGRI will carefully consider how to structure the RFAs to optimize the organization of the consortium, as each of these components is highly complex and the success of the proposed consortium relies on the efficient coordination among all members.

Council members approved the concept, with 10 votes to approve, 2 votes to oppose, and one council member abstaining. A video of Dr. Pazin's and Dr. Gilchrist's presentations can be found at: <https://youtu.be/dUqHvO1-X3A>.

CONCEPT CLEARANCE – *Developmental Genotype-Tissue Expression (RFA) – Jyoti Dayal*

Jyoti Dayal presented the Developmental Genotype-Tissue Expression (dGTEx) concept, which will be an addition to the very successful [GTEx Common Fund Program](#) which was concluded in 2016. Developed in collaboration with NICHD, dGETx aims to study gene expression patterns across early post-natal, prepubertal, and post-pubertal developmental states. By doing so, the program would establish an atlas of tissue-specific gene expression data, enhance our understanding of regulatory variation across developmental states, and create a biobank of tissue samples and data to enable future research studies. Samples will be collected in the form of blood, multiple bulk tissues, and single cells, and whole-genome sequence and RNA-Seq data will be generated. NHGRI will provide funding and program management for the Laboratory, Data Analysis, and Coordinating Centers (the subjects of this concept), while NICHD will manage and fund the tissue procurement centers. NICHD's council has approved the tissue procurement concept.

NACHGR Members approved the concept unanimously. A video of Jyoti Dayal's presentation and the related discussion can be found at: <https://youtu.be/3di0FDBLXlc>.

CONCEPT CLEARANCE – *Advancing Genomic Medicine Research (RFA) – Christine Chang*

Christine Chang presented the Advancing Genomic Medicine Research concept, which was developed based on previous NACHGR feedback and the [Strategic Planning Workshop on Genomics in Medicine & Health](#). This concept would build upon the existing Investigator-Initiated Genomic Medicine Research PARs and aims to grow the support for research projects in genomic medicine outside of large consortia. The proposed concept would stimulate innovation and advance understanding of the implementation of genomic information in clinical settings. NHGRI selected the RFA mechanism for this program to enhance grantee communication and accelerate genomic medicine research progress through annual meetings and other methods of cross-project interactions. NHGRI intends to encourage applications that encompass racial and ethnic diversity in their study design, as well as those from investigators who are new to the field of genomic medicine for the purpose of broadening the field.

NACHGRI Members approved the concept unanimously. A video of Christine Chang's presentation and the related discussion can be found at: <https://youtu.be/B7IAxWP6IMk>.

REPORT – *Genomic Medicine Working Group Update – Teri Manolio*

Dr. Teri Manolio gave an update on behalf of the [Genomic Medicine Working Group of NACHGR](#). This working group is charged with advising NHGRI on research needed to evaluate and implement genomic medicine. The group organizes regular [meetings](#) that convene clinicians, researchers, and experts from many institutions to discuss specific issues surrounding the adoption and implementation of genomic medicine. These meetings have led to a number of critical key initiatives that have benefitted the global genomic medicine research community, including NHGRI's [ClinGen](#) and [eMERGE](#) programs. The upcoming 2020 Genomic Medicine XIII meeting will focus on topics and issues concerning genomics and public health. The group also convenes monthly via teleconference to review and promote [key findings and innovations](#) in a number of genomic medicine focus areas, including clinical implementation and pharmacogenomics.

A video of Dr. Manolio's presentation and related discussion can be found at: <https://youtu.be/nr-wnzt4wwc>.

REPORT – Genomics and Society Working Group Update – Jeffery Botkin

NACHGR Member Dr. Jeffery Botkin provided an update about the [Genomics and Society Working Group of NACHGR](#), which provides NHGRI with feedback and recommendations on ELSI-related activities. In the past year, the group convened via teleconference in May 2019 and January 2020. Dr. Botkin stated that major discussion topics at the May meeting were lessons learned from the Canadian Genomics in Society / [GE3LS](#) research programs. These included how to consider the impact of ELSI research (especially in diverse populations) and improving bioethics research across the NIH. In January, the group discussed a preliminary analysis of ELSI trainee career paths, the use of genetic genealogy in forensic investigations, and genome editing and somatic tissue interventions, among other topics. The working group also devoted significant time in the past year to discussing education, training, and research areas likely to align with the 2020 Strategic Plan.

A video of Dr. Botkin's presentation and the related discussion can be found at: <https://youtu.be/D-ypqJYL5HE>.

PUBLIC HEARING – Proposed NHGRI Organizational Changes – Ellen Rolfes and Eric Green

Ellen Rolfes and Dr. Eric Green provided an overview about proposed changes to [NHGRI's organizational structure](#). These changes would specifically affect the three branches in the Division of Policy, Communication, and Education (DPCE): the Communications and Public Liaison Branch (CPLB), the Policy and Program Analysis Branch (PPAB), and the Education and Community Involvement Branch (ECIB). The proposal calls for moving into the Office of the Director, and both PPAB and ECIB into the Division of Genomics and Society (DG&S). DG&S would then be comprised of three components: the ELSI Research Program, PPAB and ECIB. The reorganization would not have any effect on budget or staffing. Following the required approval from NIH and notification of Congress, a final public hearing is planned for February 19, 2020.

A video of Ellen Rolfes' and Dr. Green's presentations and the related discussion can be found at: <https://youtu.be/v1GtnPaLhPs>.

COUNCIL-INITIATED DISCUSSION

NACHGR Members requested a future presentation that would provide information about ELSI research activities supported by other NIH Institutes. NACHGR also requested a future presentation that would provide guidance and guidelines for how investigators should report time and effort commitments on grant applications, as well as guidance and best practices to manage scientific collaborations with investigators who work at foreign institutions. Dr. Green noted there are individuals in senior leadership positions at NIH with expertise to speak to these matters and they will be invited to a future NACHGR meeting.

A video of the Council-initiated discussion can be found at: <https://youtu.be/SW7cBRNvkM>.

CONFIDENTIALITY AND CONFLICT OF INTEREST¹

Dr. Rudy Pozzatti, NACHGR Executive Secretary read the Confidentiality and Conflict of Interest Policy to NACHGR Members and asked the members to sign the forms provided to them.

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 235 applications, requesting \$98,115,209 (direct costs). The applications included: 163 research project applications (R01, R03, R15, R21 and RM1); 26 cooperative agreement applications (U01 or U24); 7 institutional training applications: 5 career development or career transition applications (K99/R00 or K01); 5 conference applications (R13); 5 education applications (R25); 14 SBIR Phase I applications (R43); 5 SBIR Phase II applications (R44), and 4 STTR Phase 1 applications (R41) and 1 STTR Phase 2 application. A total of 177 applications totaling \$72,081,459 were recommended for funding by the NACHGR.

This NACHGR Meeting Summary document was prepared by Harry Wedel, NHGRI Scientific Program Analyst.

5/20/2020

Date

Rudy O. Pozzatti

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

5/20/2020

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 May 18, 2020.

¹ NACHGR Members absented themselves from the meeting when the NACHGR discussed applications from their institutions or in which a conflict of interest may occur. Members signed a statement to this effect. This did not apply to "en block" votes.