

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
September 13-14, 2021**

The Open Session of the 94th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened virtually by Zoom at 11:30 a.m. on Monday, September 13, 2021. 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 11:30 a.m. – 5:00 p.m. on September 13, 2021. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 5:30 p.m. until adjournment on September 13, 2021 and from 11:00 a.m. until adjournment on September 14, 2021 for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT:

Joe Beery (LunaDNA)
Laura Bierut (Washington University School of Medicine)
Kyle Brothers (University of Louisville)
Howard Chang (Stanford University)
Wendy Chung (Columbia University)
Mark Craven (University of Wisconsin, Madison)
Hal Dietz (Johns Hopkins University)
Stephen Fodor (13.8 Inc.)
Rafael Irizarry (Dana-Farber Cancer Institute)
Gail Jarvik (University of Washington)
Lynn Jorde (University of Utah)
Iftikhar Kullo (Mayo Clinic)
Lisa Parker (University of Pittsburgh)
Len Pennacchio (University of California, Berkeley)
Stephen Rich (University of Virginia)
Peter Robinson (The Jackson Laboratory for Genomics Medicine)
Olga Troyanskaya (Princeton University)

**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 here: [\[link\]](#)

APPROVAL OF MINUTES

The Council approved the May 2021 Meeting Minutes by a unanimous vote.

FUTURE NACHGR MEETING DATES

- February 7-8, 2022
- May 16-17, 2022
- September 19-20, 2022
- February 13-14, 2023
- May 15-16, 2023
- September 18-19, 2023

ANNOUNCEMENT OF NEW NHGRI SCIENTIFIC DIRECTOR

Dr. Charles Rotimi will lead NHGRI's intramural research program as the new NHGRI Scientific Director. Dr. Rotimi is a leading genetic epidemiologist and genomics researcher with expertise in statistical genetics, and health disparities. He is currently an NIH Distinguished Investigator, Director of the trans-NIH Center for Research on Genomics and Global Health, and Chief of the NHGRI Metabolic, Cardiovascular, and Inflammatory Disease Genomics Branch. Dr. Rotimi is also a leader in the development of global genomics expertise and resources. He has been a vocal advocate for increased ancestral diversity in research participants and in the international scientific workforce.

DIRECTOR'S REPORT

Dr. Eric Green gave his Director's Report, which included a series of updates about NHGRI, NIH, and the broader genomics research community. A [video](#) of his presentation and the [slides](#) are available here: [\[link\]](#)

PRESENTATION – *The National Cancer Institute: Leading Cancer Research in 2021 and Beyond – Dr. Ned Sharpless*

Dr. Norman (Ned) Sharpless, Director of the National Cancer Institute (NCI), gave a presentation on the NCI's activities and goals. In the annual plan and budget proposal, NCI will request more funds for clinical trials, computer-based drug design, precision prevention, tumor dynamics, and more. Through the Cancer Genome Atlas (TCGA), NCI and NHGRI investigators have published the Pan-Cancer Atlas. This work has led to the discovery of multiple new drug targets, and modernized cancer research. TCGA has generated 2.5 petabytes of genomic, epigenomic, transcriptomic, and proteomic data from over 11,000 patients, on 33 different tumor types and, 10 rare cancers. Dr. Sharpless also provided information and previews of NCI initiatives such as the NCI Cancer Research Data Commons, Childhood Cancer Data Initiative, the Sherlock Lung Project, and the Connect for Cancer Prevention Study. Novel uses of sequencing data include investigating mutational signatures to determine tumor etiology and blood-based multi-cancer early detection tests.

A video of the presentation and the related discussion can be found here: [\[link\]](#)

PRESENTATION – *Genomics, Genetics, and the Eye – Dr. Michael Chiang*

Dr. Michael Chiang, Director of the National Eye Institute (NEI), gave an overview of the NEI's strategic plan and potential collaborations between the NEI and NHGRI. The mission of the NEI is to eliminate vision loss and improve quality of life through vision research. The institute will publish its strategic plan this coming November and will focus on seven cross-cutting areas of emphasis: from genes to disease mechanisms, biological and neuroscience, immune system and eye health, regenerative medicine, data science, individual quality of life, and public health and disparities research. NEI is also funding The National Ophthalmic Disease Genotyping and Phenotyping Network (eyeGENE), which aims to facilitate research into the causes and mechanisms of rare inherited eye diseases and accelerate pathways to treatments. eyeGENE consists of a national network of ophthalmic CLIA-certified labs, private and academic clinical organizations and their patients, and the greater vision research community.

Dr. Chiang discussed the need to create large-scale curated datasets that can be housed in databases and shared with the research community, particularly with networks of investigators who will study complex interactions among genes on the path to understanding disease mechanisms. Challenges such as privacy and de-identification of data will also need to be

addressed. One indication of collaboration between NHGRI and NEI is NEI's decision to sign on to a PAR related to the Clinical Genome Resource (ClinGen), an NHGRI research resource. ClinGen has a clinical ocular domain working group with expert panels for glaucoma, retinal gene panels and X-linked inherited diseases.

A video of Dr. Chiang presentation and the related discussion can be found here: [\[link\]](#)

CONCEPT CLEARANCE – *Initiative to Maximize Research Education in Genomics: Courses (PAR) – Dr. Tina Gatlin*

Dr. Tina Gatlin presented the Initiative to Maximize Research Education in Genomics: Courses concept. This concept is a renewal of an R25 program to support the offering of advance level courses that are intended to disseminate new techniques, methods, analyses, and knowledge related to all research areas of NHGRI. This program will be open to participants nationwide who are at a master's level or beyond, and course materials must be made available to the public. Courses should also align with the NHGRI Strategic Vision. Council suggested creating a clearing house for course materials and establishing metrics for long term impact.

Council members approved the concept unanimously. A video of Dr. Gatlin's presentation can be found here: [\[link\]](#)

CONCEPT CLEARANCE – *Enhancing Diversity in Cloud-based Genomic Data Science Education (two RFAs) -- Dr. Shurjo Sen*

Dr. Shurjo Sen presented the Enhancing Diversity in Cloud-based Genomic Data Science Education concept. The goal of this concept is to facilitate the development of a diverse genomics workforce with training in computational genomics and data science (CGDS). The program will use NIH-funded cloud platforms as a tool to enhance CGDS education at institutions serving groups underrepresented in the biomedical and data science fields. Eligible institutions include historically black colleges and universities (HBCUs), Hispanic-serving institutions, and colleges and universities with predominantly female enrollment. The two RFAs will support individual sites for curriculum development through R25 grants and a single CGDS Hub (U24 grant) that will act as the facilitator/coordinator for cloud-based CGDS education, and partner with the sites to support faculty at eligible institutions to create and adopt CGDS education content.

Council members approved the concept unanimously. A video of Dr. Sen's presentation can be found here: [\[link\]](#)

ANNUAL REPORT — *Data Science Working Group of NACHGR – Dr. Mark Craven*

Council member Dr. Mark Craven provided an annual report on the NHGRI Genomic Data Science Working Group of NACHGR. The working group advises NHGRI on plans related to genomic data science outside of council meetings and provides input to the NHGRI Director and other Institute leaders about trans-NIH issues related to data science. Members address broad challenges, including data management, analysis, computing and data science policy. Some topics in 2020-2021 included renewal of two NHGRI Computational Genomics and Data Science FOAs, NHGRI AnVIL project updates, overview of training and outreach activities for broadening and bridging genomics/data science communities, and the use and development of artificial intelligence and machine learning approaches in biomedical research.

A video of Dr. Craven's presentation can be found here: [\[link\]](#)

ANNUAL REPORT — *Community Engagement in Genomics Working Group of NACHGR – Dr. Greta Goto and Dr. Maya Sabatello*

Dr. Greta Goto and Dr. Maya Sabatello provided an overview of the NHGRI Community Engagement in Genomics Working Group of NACHGR. The working group aims to assess the gaps and needs of diverse communities related to genomics and genomic medicine, provide input about NHGRI activities designed to increase genomic literacy, work in partnership with NHGRI to build relationships and trust with communities, and increase understanding of the views and priorities of diverse communities. This past year, the working group provided feedback to the Morehouse School of Medicine on their engagement in the genomics toolkit project and co-authored a publication on structural racism in the era of the COVID-19 pandemic. In the coming year, the working group will publish a journal article detailing the complexities and intersectionality of trust and mistrust in precision medicine research. Working group members are also collaborating with NHGRI's Inter-Society Coordinating Committee for Practitioner Education in Genomics.

A video of Dr. Goto's and Dr. Sabatello's presentation can be found here: [\[link\]](#)

PRESENTATION – *Report on Multi-Omics in Health and Disease Workshop –Dr. Joannella Morales and Dr. Howard Chang*

Dr. Joannella Morales and Dr. Howard Chang gave a presentation on the importance of multi-omics research and the June 2021 Multi-Omics in Health and Disease Workshop sponsored by NHGRI. The integration of multiple omics studies such as genomics, transcriptomics, proteomics, epigenomics, and metabolomics has led to increased insights and comprehensive assessments using high throughput technologies. Moreover, NHGRI aims to extend multi-omics studies of human disease and health into clinical settings. Thus, NHGRI hosted a workshop to brainstorm focus areas in the multi-omics space. Workshop participants provided recommendations on broad categories such as ensuring diversity and inclusion, developing high throughput technologies, methods, and analytical tools, enhancing cohort studies, and establishing consensus and best practices.

A video of Dr. Morales' and Dr. Chang's presentation can be found here: [\[link\]](#)

COUNCIL-INITIATED DISCUSSION

Council members asked for an update on the impact of the pandemic on NHGRI early career stage investigators and what NHGRI can do for these investigators.

For future Council meetings, Council members recommended presentations from other NIH Institute Directors. Council also requested information about how two new research programs, KOMP2 and MorPhiC will interact and/or integrate. NHGRI will need to wait until applications for these projects are received to know how the research community has responded to the FOAs to determine what the relationship will be between KOMP2 and MorPhiC. Lastly, Council members requested an update on how AnVIL is being used and what plans have been made for its sustainability.

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

CONFLICT OF INTEREST STATEMENT

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 155 applications, requesting \$56,813,966 (direct costs). The applications included: 110 research project applications (R01, R15, or R21); 2 education applications (R25); 6 cooperative agreement applications (U24); 8 career development applications (K99/R00, K01 or K08); 1 conference applications (R13); 17 SBIR Phase I applications (R43); 8 SBIR Phase II applications (R44); 2 STTR Phase 1 applications (R41), and 1 STTR Phase 2 application.

NHGRI STAFF PRESENT:

Katie Bardsley, ERP
Sarah Bates, CPLB
Yasmeen Beckett, IOD
Vence Bonham, IOD
Joy Boyer, ERP
Lawrence Brody, ERP
Comfort Browne, ERP
Alicia Caffi, DM
Stephanie Calluori, ERP
Lisa Chadwick, ERP
Christine Chang, ERP
Monika Christman, ERP
Heather Colley, ERP
Amanda Conti, PPAB
Priscilla Crockett, ERP
Luis Cubano, ERP
Jyoti Dayal, ERP
Edith DeHaut, SRB
Christopher Donohue, IOD
Argenia Doss, ERP
Laura Eisenman, ERP
Alvaro Encinas, CPLB
Elise Feingold, ERP
Adam Felsenfeld, ERP
Kimberly Ferguson, ERP
Ann Fitzpatrick, DM
Colin Fletcher, ERP
Valentina Di Francesco, ERP
Anneliese Galczynski, ERP
Prabarna Ganguly, CPLB
Sean Garin, ERP
Katy Garvey, DM
Tina Gatlin, ERP
Elena Ghanaim, PPAB
Daniel Gilchrist, ERP

Jenell Glover, ERP
Madison Goldrich, ERP
Bettie Graham, ERP
Chris Gunter, IOD
Linda Hall, ERP
Peggy Hall, ERP
Zephaun Harvey, ERP
Edith DeHaut, ERP
Joe Henke, ITB
Lucia Hindorff, ERP
Nichole Holm, ERP
Belén Hurle, ECIB
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Amber Jackson, ERP
Cristina Kapustij, PPAB
Dan Kastner, IRP
Dave Kaufman, ERP
Denise Kennedy, DM
Se Rin (Julie) Kim, ERP
Grace Lamoure, ERP
Darryl Leja, CPLB
Rongling Li, ERP
Asiyah Lin, ERP
Natalie Linear, ERP
Nicole Lockhart, ERP
Ebony Madden, ERP
Teri Manolio, ERP
Radu Marin, ITB
Joannella Morales, ERP
Donna Morris, ERP
Marcia Morris, ERP
Stephanie Morris, ERP
Ken Nakamura, ERP
Mukul Nerurkar, CPLB

Emily Neveux, DM
Allie Osgood, ERP
Mike Pazin, ERP
Devona Perrineau, CPLB
Haleena Phillips, ERP
Ajay Pillai, ERP
Rudy Pozzatti, ERP
Erin Ramos, ERP
Kathleen Renna, ERP
Enitza Rodriguez, ERP
Ellen Rolfes, DM
Kristen Ross, ERP
Charles Rotimi, IRP
Robb Rowley, ERP
Ismail Safi, ERP
Ella Samer, ERP
Lorjetta Schools, ERP
Baergen Schultz, ERP
Jamil Scott, IOD
Shurjo Sen, ERP

Michael Smith, ERP
Grace Snyder, ERP
Heidi Sofia, ERP
Jerryl Somani, ITB
Rene Sterling, ERP
Ana Stevens, ERP
Jennifer Strasburger, ERP
Michelle Tallman, ERP
Jill Thomas, CPLB
Barbara Thomas, ERP
Helen Thompson, ERP
Jennifer Troyer, ERP
Susan Vasquez, IOD
Simona Volpi, ERP
Harry Wedel, CPLB
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP
Valerie Willis, ERP
Rachael Zisk, CPLB

ERP = Extramural Research Program
CPLB = Communications and Public Liaison Branch
IOD = Immediate Office of the Director
DM = Division of Management
ECIB = Education and Community Involvement Branch
ITB = Information Technology Branch
PPAB = Policy and Program Analysis Branch
IRP = Intramural Research Program

This NACHGR Meeting Summary was prepared by Se Rin (Julie) Kim, NHGRI Scientific Program Analyst.

02/09/2022

Date

Rudy Pozzatti

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

02/09/2022

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 February 7, 2022