

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
February 13-14, 2023**

The Open Session of the 98th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:30 a.m. on Monday, February 13, 2023, with 14 Council members participating virtually by 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 10:30 a.m. – 6:00 p.m. on February 13, 2023. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 9:00 a.m. – 10:00 a.m. on February 13, 2023, and from 11:00 a.m. until adjournment on February 14, 2023, for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT

Laura Bierut (Washington University)
Kyle Brothers (University of Louisville)
Mark Craven (University of Wisconsin, Madison)
Hal Dietz (Johns Hopkins University)
Gail Jarvik (University of Washington)
Lynn Jorde (University of Utah)
Iftikhar Kullo (Mayo Clinic)
Lisa Parker (University of Pittsburgh)
Len Pennacchio (Lawrence Berkeley National Laboratory)
Timothy Reddy (Duke University)
Stephen Rich (University of Virginia)
Olga Troyanskaya (Princeton University)

AD HOC MEMBER

Judy Cho (The Icahn School of Medicine at Mount Sinai Hospital)

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

Dr. Rudy Pozzatti began the Open Session with the introduction of new Council members, new members of the NHGRI staff, liaisons, and guests, which can be viewed here: [\[link\]](#)

APPROVAL OF MEETING SUMMARY

The Council approved the September 19-20, 2022 Meeting Summary by a unanimous vote.

FUTURE NACHGR MEETING DATES

- May 15-16, 2023
- September 18-19, 2023
- February 12-13, 2024
- May 20-21, 2024
- September 16-17, 2024

DIRECTOR'S REPORT

Dr. Eric Green gave the 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 – *Building a Diverse Genomics Workforce Action Agenda: Implementation Plan and Progress Report – Mr. Vence Bonham*

Mr. Vence Bonham, Acting Deputy Director, NHGRI, updated Council on the implementation plan and a progress report for the last two years of the "Building a Diverse Genomics Workforce Action Agenda." By working with institutes and centers across NIH, NHGRI's Training, Diversity, and Health Equity (TiDHE) Office staff is working to enhance diversity of the genomics workforce by supporting new initiatives, advancing genomic health equity, and conducting health disparities research. Council members responded positively about these programs and are looking forward to hearing about metrics that will be used to evaluate progress of these programs at future Council meetings. There was discussion of how to expand the implementation plan to the wider community, particularly through virtual seminars or in-person conferences.

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

PRESENTATION – *Outstanding Award for Diversity, Equity, Inclusion, and Accessibility (DEIA) in the Genomics Workforce – Ms. Lorjetta Schools*

Ms. Schools presented the NHGRI's Award for Enhancing Diversity, Equity, Inclusion, and Accessibility (DEIA) in the Genomics Workforce. This annual award was established to highlight the accomplishments of extramural researchers and NHGRI staff's efforts towards DEIA in the human genetics and genomics workforce.

The 2022 Outstanding Award for DEIA in the Genomics Workforce/Established Investigator was awarded to the Broad Institute's Pardis Sabeti, MD, PhD, for elevating DEIA in the study of infectious diseases. Her accomplishments include the co-founding of the *African Centre of Excellence for Genomics of Infectious Disease* (ACEGID), support for scientists in Africa to enable the first diagnosis of Ebola in Sierra Leone and Nigeria, co-founding Operation Outbreak, and co-creating an outbreak science textbook for high school students.

The 2022 Outstanding NHGRI Staff Award for DEIA in the Genomics Workforce was awarded to Bettie Graham, PhD, for her career-long efforts to advance diversity in NHGRI extramural training. Her accomplishments include development of the First Minority Action Plan for NHGRI, involvement in the UNITE Initiative at NIH, expansion of the Eligibility of the Loan Repayment Program, and involvement in the review of pay increases for NHGRI's and NCAT's Title 42 recipients.

The 2022 NHGRI Staff Award for DEIA Innovation in the Genomics Workforce was awarded to Ann Mc Cartney, PhD, for centering Indigenous perspectives in genomic data science. Dr. Mc Cartney is currently a Visiting Fellow in the Genome Informatics Section, NHGRI. Her accomplishments within this realm include teaching "Intro to Genomics" session for the summer internship for Indigenous People in Genomics USA/Canada. She is one of the faculty organizers of the first IndigiData workshop, and she chaired the working group for indigenous peoples and local communities for data sharing and management. Dr. McCartney is a co-investigator on a

U24 award for creating a COVID-19 tribal data repository, and is the chair of the European Reference Genome Atlas pilot project.

After the awards were presented, Dr. Eric Green moderated a fireside chat with the awardees. They discussed topics such as Dr. Sabeti's experience working with African investigators, Dr. Graham's experience with diversity programs throughout her 50 years of federal service, and Dr. Mc Cartney's views on diversity on the global scale as she traveled to New Zealand and Africa.

A video of Ms. Schools' presentation and Dr. Green's fireside chat with awardees can be found here: [\[link\]](#)

CONCEPT CLEARANCE – PAR Renewal: ELSI Program Announcements for R01, R03, and R21 Applications – Dr. Dave Kaufman

Dr. Kaufman, a Program Director in NHGRI's Division of Genomics and Society, sought approval to renew the current Ethical, Legal, and Social Implications (ELSI) program announcements that will expire in July 2023. The Council members focused on the issue that R21 awards are difficult to complete in two years due to the complexity of work often performed in ELSI research. Council members expressed support for the proposal to offer R21 awards with three years of funding.

The concept was approved by Council with 12 votes to approve, no objections, and 1 abstention.

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

CONCEPT CLEARANCE – RFA Renewal: Center for ELSI Resources and Analysis – Dr. Nicole Lockhart

Dr. Lockhart, a Program Director in NHGRI's Division of Genomics and Society, sought approval for the *Center for ELSI Resources and Analysis* concept. She noted that the renewal would provide a platform to share ELSI research products, enable synthesis and curation of ELSI research findings, and facilitate collaborations and uptake of ELSI research. ELSI research products are often used outside of NHGRI-funded research projects, in part because this field represents a great diversity of disciplines. The renewal will be an open-competition RFA beginning in Fiscal Year 2024 with direct costs up to \$6 million over five years. This concept renewal received significant support among Council members as it allows for creativity and has a wide range of applicability. There was discussion of how this program will be evaluated externally, how the user interface will be set up, and how standards and best practices will be implemented in a way that will ensure continuity from the work done in the initial funding phase and into the future.

There were 13 votes to approve the concept, no objections, and no abstentions.

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

CONCEPT CLEARANCE – PAR Renewal: Diversity Action Plan – Dr. Lucia Hindorff

On behalf of the NHGRI Extramural Training Team, Dr. Hindorff presented the Diversity Action Plan (DAP) Renewal. The DAP is being reissued to highlight the NIH Notice of Interest in

Diversity, the NHGRI Diversity Action Plan Agenda, and the new NHGRI- and NIH-wide FOAs for trainees from diverse backgrounds. Proposed changes include focusing on students in undergraduate and post-baccalaureate programs, summer- and semester-long training experiences, increasing diversity among mentors, expanding approaches for recruitment in minority-serving institutions and community colleges, and adding language to the PAR for evidence-based evaluation approaches. Council members were very supportive of the proposed amendments to diversify the students and mentors in the undergraduate/post-baccalaureate programs. Emphasis was placed on the importance of having diverse mentors in these programs as they bring unique perspectives and could help mitigate barriers faced in academics.

There were 13 votes to approve the concept, no objections, and no abstentions.

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

CONCEPT CLEARANCE – *RFA Renewal: Advancing Genomic Medicine Research* – Ms. Christine Chang

Ms. Chang presented the renewal for the *Advancing Genomic Medicine Research* (AGMR) RFA. This RFA aims to address research gaps related to the use of genomic information and to advance the application of genomics in clinical care. Proposed changes include a Notice of Special Interest (NOSI) for Small Business applications (R43/R44); having one receipt date per year in July; and increasing the budget and length of support from two to three years for R21 applications. The goals of these proposed changes are to encourage applications from new investigators and to continue advancing diversity. Council members noted that increasing the project period across "R-series" awards could decrease the number of awards made over time. Ms. Chang addressed these concerns, and Council members agreed that increasing the project period is necessary for these high-impact projects.

There were 13 votes to approve the concept, no objections, and no abstentions.

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

CONCEPT CLEARANCE – *RFA Renewal: Human Genome Reference Program* – Dr. Adam Felsenfeld

Dr. Felsenfeld presented three Funding Opportunity Announcements (FOAs) for the *Human Genome Reference Program* RFA renewal. These RFAs included a new emphasis on community adoption for the Human Pangenome Reference Center; development of criteria for prioritizing utility factors for basic and clinical studies for the Center for High-Quality Reference Genomes; and a new FOA for Informatics Tools for the Pangenome Reference that will facilitate the uptake and use of the Pangenome Reference. The overall goal for these FOAs is to improve the human genome reference sequence and reduce gaps in reference genomes. Council members were supportive of the renewal, particularly noting the pangenome resource as an example of research at the forefront of genomics. Council members also pointed out the need to support the development of tools and methods for investigators who want to use the pangenome. Accessibility for engaging the community is crucial as they bring new, diverse, and creative ideas that can broaden pangenome use.

There were 13 votes to approve the concept, no objections, and no abstentions.

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

REPORT – Genomic Medicine Working Group of Council: Update of Meetings, Activities, and Accomplishments – Dr. Teri Manolio

Dr. Manolio presented updates from the Genomic Medicine Working Group of Council. She reviewed the charge and responsibilities of the Working Group (WG) and noted the key advances made in genomic medicine over the past year. The WG also produces an annual manuscript entitled, *Genomic Medicine Year in Review*, which is published in the *American Journal of Human Genetics*. The WG also helps to plan an annual Genomic Medicine Workshop that discusses the research agenda for Genomic Medicine. The most recent such workshop was held in September 2022 and focused on the topic of "Genomic Learning Healthcare Systems." Dr. Manolio reviewed the key recommendations from this meeting. She also presented a high-level review of the current portfolio of Genomic Medicine research supported by NHGRI. Council members responded positively to the report and showed curiosity about efforts to address gaps in current research, next steps, and the timeframe of these efforts.

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

REPORT – Genomic Data Science Working Group of Council Update – Dr. Mark Craven

Dr. Craven is a Council member and serves on the Genomic Data Science Working Group. He presented an overview of the Working Group functions, membership, 2022 NHGRI-focused topics in data science, trans-NIH efforts and initiatives, and upcoming Working Group discussion topics. In 2022, the Working Group focused on future directions and added components to support clinical research of the Genomic Data Science Analysis, Visualization, and Informatics Lab-Space (AnVIL), the newly implemented NIH Data Management and Sharing Policy, and the new NHGRI Office of Genomic Data Science (established in early 2022). Trans-NIH topics covered by the Working Group include collaborations between the NIH Office of Data Science Strategy and NHGRI, the NIH transition to the cloud, and NIH STRIDES initiative.

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

REPORT – Community Engagement in Genomics Working Group of Council Update – Dr. Maya Sabatello and Ms. Greta Goto

Dr. Sabatello of Columbia University and Ms. Goto of the Prader-Willi Syndrome Alaska Parent Group, serve as co-chairs of the Community Engagement in Genomics Working Group. They provided an update on the activities of this Working Group of Council, focusing on two projects: (1) the intersectionality and trust in genomic medicine research; and (2) community outreach mapping. Community outreach efforts seek to approach members of medically underserved communities such as people with disabilities, the LGBTQI+ community, and Indigenous communities. Council expressed gratitude for this Working Group's efforts in engaging underserved communities and providing resources that are helpful with the challenge of engaging minority populations. Council members also took a forward-thinking perspective, encouraging collaboration with the National Center for Advancing Translational Sciences (NCATS), which also sponsors a lot of community engagement research.

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

Review of the Statement of Understanding between NACHGR and NHGRI– Dr. Rudy Pozzatti

Dr. Pozzatti reviewed the Statement of Understanding (SOU) between NACHGR and NHGRI. This document is reviewed annually at the February Council meeting. It details the responsibilities and authorities of the Council and the NHGRI staff, including the types of applications and actions that must be brought to the Council for presentation and review. Dr. Pozzatti noted two changes that had been made to the SOU since the review in February 2022. These include one change made to the process of conducting Special Council Review when a principal investigator has \$2 million in total funding. The second change will allow NHGRI to convene a council meeting via electronic format, absent any national disaster.

There were 12 votes to approve the Statement of Understanding, no objections, and no abstentions.

A video of this discussion can be found at: [\[link\]](#)

COUNCIL-INITIATED DISCUSSION

Council members expressed an interest in inviting new directors from other NIH institutes to future Council meetings to discuss their research priorities. There have been leadership changes at several institutes in the past few years; thus, there are several candidates to consider.

Concerns over the issue of graduate student pay disparities was raised as a major issue affecting all of biomedical research. The gap between NIH graduate student stipends and what is actually paid to the students is growing. Council members expressed how this is adding budgetary pressure to investigators and their institutions. While the importance of this issue was acknowledged, it was noted that NIH is not able to make a direct change as Congress decides the stipend levels.

There were discussions regarding the inclusion of journal fees in grant budgets due to the disparities in costs across many journals. Based on current practices, these fees need to be budgeted for in the grant.

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 184 applications, requesting a total of \$81,742,282 (direct costs). The applications included: 115 research project applications (R01, R03, or R21); 2 R15 application; 2 conference applications (R13); 6 RM1 applications: 16 education applications (R25); 13 Community Resource applications (U24); 5 Institutional

Training applications (T32); 5 Career Development applications (F99/K00, K99/R00); 15 SBIR Phase I application (R43); 3 SBIR Phase II applications (R44); and 2 STTR Phase 1 application.

NHGRI STAFF PRESENT

Ernesto Del Aguila, OC
Alexander Arguello, ERP
Katie Bardsley, ERP
Jake Baroch, ERP
Sarah Bates, OC
Zo Bly, ERP
Danielle Buice, IRP
Vence Bonham, IOD
Lawrence Brody, ERP
Comfort Browne, ERP
Danielle Buice, IRB
Devon Bumbray-Quarles, ERP
Gloria Butler, DM
Lisa Chadwick, ERP
Eggerton Campbell ERP
Christine Chang, ERP
Paul Cheung, DM
Monika Christman, ERP
Heather Colley, ERP
Sara Currin, ERP
Jyoti Dayal, ERP
Christina Daulton, ECIB
Edith DeHaut, ERP
Alvaro Encinas, OC
Valentina Di Francesco, OGDS
Elise Feingold, ERP
Juila Fekecs, OC
Kimberly Ferguson, ERP
Adam Felsenfeld, ERP
Ann Fitzpatrick, DM
Colin Fletcher, ERP
Brianna Foster, ERP
Temesgen Fufa, ERP
Anneliese Galczynski, ERP
Elena Ghanaim, OGDS
Daniel Gilchrist, ERP
Jenell Glover, OGDS
Bettie Graham, ERP
Chris Gunter, IOD
Peggy Hall, ERP
Zephaun Harvey, ERP
Lucia Hindorff, TiDHE
Sarah Hutchinson, ERP
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Amber Jackson, TiDHE

Cristina Kapustij, PPAB
Dave Kaufman, ERP
Jim Kees, ERP
William Kibby, ITB
Britny Kish, OC
Se Rin (Julie) Kim, OGDS
Darryl Leja, OC
Rongling Li, ERP
Natalie Linear, ERP
Nicole Lockhart, ERP
Ebony Madden, TiDHE
Teri Manolio, ERP
Radu Marin, ITB
Iman Martin, ERP
William Maye, ITB
Allison McCague, PPAB
Imani McGregor, ERP
Julius Militante, ERP
Ava Miller, IRP
Jenn Montooth, OC
Joannella Morales, ERP
Donna Morris, ERP
Marcia Morris, ERP
Stephanie Morris, ERP
Jahnvi Narula, ERP
Emily Neveux, DM
Nguyen Park, ERP
Mike Pazin, ERP
Ajay Pillai, ERP
Caprina Pipion, ERP
Mauresa Pittman, OC
Colette Pollard, OGDS
Erin Ramos, ERP
Renee Rider, ERP
Enitza Rodriguez, ERP
Anna Rogers, OC
Ellen Rolfes, DM
Robb Rowley, ERP
Ellaha Sadat, OC
Ismail Safi, ERP
Lorjetta Schools, TiDHE
Jamil Scott, TiDHE
Shurjo Sen, OGDS
Heidi Sofia, ERP
Kenya Smith, OC
Jeryl Somani, ITB

Sonja Soo, OC
Rene Sterling, ERP
Jennifer Strasburger, ERP
Nicola Sugden, OC
Michelle Tallman, ERP
Barbara Thomas, ERP
Helen Thompson, OGDS
Jennifer Troyer, ERP
Maricela Trujillo, ERP

Maya VanZanten, ERP
Susan Vasquez, IOD
Simona Volpi, ERP
Chris Wellington, OGDS
Kris Wetterstrand, IOD
Riley Wilson, ERP
Sarah Wheelan, ERP
Sandhya Xirasagar, OGDS

ERP = Extramural Research Program
OC = Office of Communications
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
TiDHE = Training, Diversity, and Health Equity Office
OGDS = Office of Genomic Data Science
IRP = Intramural Research Program

This NACHGR Meeting Summary was prepared by Jenell Glover, NHGRI Scientific Program Analyst.

6/22/2023

Date

Rudy Pozzatti, Ph.D.

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

6/23/2023

Date

Eric Green, M.D, Ph.D.

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

This report was approved by the NACHGR on May 15, 2023