

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
May 20-21, 2024**

The Open Session of the 102nd meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:30 a.m. on May 20, 2024, with 12 Council members participating in person or 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 May 20, 2024. 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 May 20, 2024, and from 10:00 a.m. until adjournment on May 21, 2024 for the review, discussion, and evaluation of grant applications. This portion of the meeting on May 21st was closed to the public in accordance with the determination that it concerned matters exempt from mandatory disclosure under sections 552b(c)(4) and 552b(c)(6), Title 5 U.S.C., and section 1009(d) of the Federal Advisory Committee Act, as amended (5 U.S.C. §§ 1001-1014). Members absented themselves from the meeting during discussion of and voting on applications from their own institutions or other applications in which there was a potential conflict of interest (real or apparent).

COUNCIL MEMBERS PRESENT

Joe Beery (LunaDNA)
Kyle Brothers (University of Louisville)
Judy Cho (The Icahn School of Medicine at Mount Sinai Hospital)
Nancy Cox (Vanderbilt University)
Lynn Jorde (University of Utah)
Iftikhar Kullo (Mayo Clinic)
Timothy Reddy (Duke University)
Peter Robinson (The Jackson Laboratory)

AD HOC MEMBERS

Katrina Armstrong (Vagelos College of Physicians and Surgeons at Columbia University)
Kelly Frazer (University of California, San Diego)
Bruce Korf (University of Alabama, Birmingham)
Casey Overby Taylor (Johns Hopkins University)

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 NHGRI staff members, liaisons, and guests, which can be viewed here: [\[link\]](#)

APPROVAL OF MEETING SUMMARY

The Council approved the February 12-13, 2024 Meeting Summary by a unanimous vote.

FUTURE NACHGR MEETING DATES

- September 9-10, 2024
- February 10-11, 2025
- May 19-20, 2025
- September 15-16, 2025
- February 9-10, 2026
- May 18-19, 2026

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, [slides](#), and related documents are available here: [\[link\]](#)

PRESENTATION – *The NIAID Mission: Infectious Diseases and Beyond* – Dr. Jeanne Marrazzo

Dr. Marrazzo is the Director of the National Institute of Allergy and Infectious Diseases (NIAID). She presented an overview of the mission of NIAID, the institute's recent research that has led to public health interventions, and NIAID's budgetary changes over time. Dr. Marrazzo introduced the 2025-2029 NIAID Strategic Plan, which has a Request for Information (RFI) that is open at this time. The NIAID Centralized Sequencing Program for the Infectious Diseases Clinic facilitates intramural research with 10 collaborating institutes and centers (ICs). Dr. Marrazzo described the NIAID Clinical Genomics Program (NCGP) and a case study of CHAPLE disease, which showcased the power of merging genomic medicine with discovery and development of therapy. NIAID's opportunities for engagement include the Inborn Errors of Immunity/Primary Immunodeficiencies Notice of Special Interest (NOSI) and the NIAID-NHGRI Workshop: *Genomic Medicine XVI*. Currently, NIAID is monitoring a highly pathogenic avian influenza strain (H5 HPAI). Council discussion concerned the lack of funds for pandemic preparedness and the NIAID Strategic Plan. Council members were interested in learning about NIAID's view on climate change and the increased presence of vector-borne diseases in the US. There may be opportunities for ClinGen to collaborate with NIAID.

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

CONCEPT CLEARANCE – *RFA: Supporting Talented Early-Career Researchers in Genomics* – Dr. Lisa Chadwick

Dr. Chadwick sought concept approval for renewal of the Supporting Talented Early-Career Researchers in Genomics Request for Information (RFA). She expressed the increasing difficulty of early-stage investigators to receive an NIH grant. This RFA targets funding Early-Stage Investigators (ESI) and the Next Generation Researchers Initiative (NGRI). The Supporting Talented Early-Career Researchers in Genomics RFA is proposing another three-year RFA with one application due date each year, funding five-year awards from a \$2M/year set-aside for competing awards. There are no significant changes proposed from the original RFA. Council members were supportive of the proposed renewal to continue providing opportunities to ESIs. Council members noted that it may be better to have multiple receipt dates per year, so those applicants that are not funded can reapply sooner. NHGRI has an ESI webpage that provides several resources for applicants.

There were 12 votes to approve the concept, no objections, and no abstentions.

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

CONCEPT CLEARANCE – *RFA: Genome Research Experiences to Attract Talented Undergraduates into Genomic Fields to Enhance Diversity (GREAT)* – Dr. Ebony Madden

Dr. Madden sought concept approval for the renewal of the Genome Research Experiences to Attract Talented Undergraduates into Genomic Fields to Enhance Diversity (GREAT) RFA. The

GREAT program provides exposure to genomics research for undergraduate students enrolled at minority-serving institutions (MSIs) or Institutional Development Award (IDeA)-eligible states. Four applicants have been funded since the program began in Fiscal Year (FY) 2022, and one to two more applicants will be funded in FY2024. This RFA has allowed students to change their career paths and broaden their horizons. The renewal will continue to enhance diversity in undergraduate research. There are no significant changes proposed from the original RFA. Council members were supportive of the proposed renewal to continue providing undergraduate research opportunities to diverse populations. There was a discussion regarding the success of the program thus far and the application process. Council members noted NIH has many resources for new applicants, but they could be better publicized.

There were 12 votes to approve the concept, no objections, and no abstentions.

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

CONCEPT CLEARANCE – *PAR: NHGRI Predoctoral to Postdoctoral Transition Award for a Diverse Genomics Workforce – Dr. Lucia Hindorff*

Dr. Hindorff presented a concept for reissuing a Program Announcement (PAR) that will support investigators transitioning from predoctoral to postdoctoral positions in the form of up to two years of the predoctoral F99 phase and up to three years of the postdoctoral K00 phase. To increase accessibility, applicants from foreign institutions can now apply to the PAR. Five applications have been funded since the PAR began in FY2022. There are no significant changes proposed from the original PAR. Council members noted an increasing trend of doctoral graduates taking industry positions and forgoing a postdoctoral research experience. They asked if there is any evidence that the F99/K00 award might encourage more people to accept a postdoctoral research position. All of NHGRI's F99/K00 grantees have transitioned to the K00 phase, but this represents a very small sample size. Moving expenses represent a barrier for doctoral graduates transitioning to a postdoctoral position. NIH awards cannot support these costs, but institutions can.

There were 12 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: [\[link\]](#)

PRESENTATION – *Bettie J. Graham Leadership Awards for Enhancing Diversity, Equity, Inclusion, and Accessibility in the Genomics Workforce – Dr. Lucia Hindorff, Dr. Eric Green, Dr. Shilpa Garg, Dr. Shurjo Sen, and Dr. Nicola Mulder*

Dr. Hindorff introduced the *Bettie J. Graham Leadership Awards for Enhancing Diversity, Equity, Inclusion, and Accessibility in the Genomics Workforce*. Award categories include an Extramural Early-Stage Investigator, an Extramural Established Investigator, and an NHGRI Staff Member. Dr. Green announced the awardees.

The Extramural Early-Stage Investigator awardee is Dr. Shilpa Garg. She presented on her research activities, which included the hierarchical approach that her group has created for constructing a pangenome with class, genus, and species variation. Engaging diverse communities and increasing diversity, equity, inclusion, and accessibility (DEIA) in genomics education and outreach has been a cornerstone throughout her career.

The NHGRI Staff Member awardee is Dr. Shurjo Sen. He presented programs in which he has played a major role, including using NIH cloud computing platforms as a classroom for

broadening the genomics and data science workforce. While there have been some improvements in DEIA in the STEM workforce, it has been marginal. Cloud computing will help democratize access to computing resources, provide access to representative datasets, and enable virtual collaboration.

The Extramural Established Investigator awardee is Dr. Nicola Mulder. Dr. Mulder was very successful as an H3Africa grantee for the H3Africa Bioinformatics Network (H3ABioNet) and continues to perform research as the lead PI for the African Genomics Data Hub (AGDH). She presented on her progress and plans to establish and increase the genomics workforce in Africa. Her work has been, and will continue to be, accomplished through different methods of training and increasing accessibility to African data from biomedical research projects.

The three panelists responded to questions from Dr. Green during the discussion, such as when DEIA became a priority in their careers, how to improve diversity of the genomics workforce, what advice they give trainees and mentors, and how to better push forward an agenda at NIH to improve DEIA. Council Members were curious regarding the concept of “training the trainers,” advice to be a good mentor to trainees from underrepresented backgrounds, the ability to connect the trainees with research, and implications for how Artificial Intelligence (AI) will modulate the workforce and training.

A video of the awardee presentations and the panel discussion can be found here: [\[link\]](#)

PRESENTATION – *Impact of Genomic Variation on Function (IGVF) Consortium Update – Dr. Karen Mohlke and Dr. Jesse Engreitz*

Dr. Mohlke and Dr. Engreitz presented an overview of the NHGRI-funded IGVF consortium. The main goal of IGVF is to produce a genome-wide map of genomic variant effects. IGVF consists of Functional Characterization Centers, Predictive Modeling Projects, Mapping Centers, Regulatory Network Projects, and a Data and Administrative Coordinating Center. IGVF’s key outcomes are to create a resource to accelerate research throughout the community and discover biological insights about genomic variants, genome function, and disease. Dr. Engreitz explained some examples of how IGVF has addressed research challenges thus far, such as linking enhancers to target genes, interpreting genomic variants of uncertain significance, linking variants to functions for lipid traits and coronary artery disease, applying the Map-Perturb-Predict framework, and uniform data processing pipelines and standards, as well as the creation of the IGVF Data Resource. The Council discussion noted the diversity of the IGVF cohort, the collaboration within the consortium, and how IGVF addresses the difficulties of studying coronary artery disease. Council members commended the IGVF data portal and questioned what, if any, feedback IGVF has received from the community. Council members were curious as to how Drs. Mohlke and Engreitz see the relationship between IGVF and other current large-scale data-generation projects.

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

PRESENTATION – *Genomics Research to Elucidate the Genetics of Rare Disease (GREGoR) Consortium Update – Dr. Jennifer Posey*

Dr. Posey presented an overview of the NHGRI-funded GREGoR consortium. The mission of GREGoR is to significantly increase the proportion of Mendelian conditions with an identified genetic cause. This mission will be accomplished through three goals: creating a dataset with broad utility, increasing diagnostic yield, and developing genomics recommendations.

Addressing disparities in the field has been an ongoing consideration in pursuit of GREGoR's goals. Council members acknowledged how far GREGoR has come and commended GREGoR on their community outreach and providing data resources to the community. Council members were curious to know the upper boundary of GREGoR's success rate and what percentage of traits and disorders do not have a genetic cause identified. Dr. Green asked Dr. Posey to explain the difference between GREGoR's and the Undiagnosed Diseases Network's (UDN) goals. The GREGoR Consortium is attempting to advance the field of rare disease research in everything they do. They see a greater volume of patients than the UDN, and the UDN commits much more time and effort to in-person phenotyping than GREGoR.

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

REPORT – *Workshop Report: Advances in the Genetic Architecture of Complex Human Traits – Dr. Alexander Arguello and Dr. Nancy Cox*

Dr. Arguello was the lead Program Director responsible for the “Advances in the Genetic Architecture of Complex Human Traits” workshop, while Dr. Cox was part of the Scientific Organizing Committee. A summary of the workshop and other information can be found [here](#). Dr. Cox presented some major themes that emerged from the meeting, including different parts of genetic architecture (i.e., population and statistical vs. biological and mechanistic), the importance of context, and differences in complexity of traits. Open questions for future research include whether current quantitative genetic theories and models are sufficient to tackle the full variety of complex traits and how to precisely quantify and measure non-genetic factors. Council members noted two concerns relevant to this research field: (1) the sparsity of single-cell multi-omics data, and (2) the challenges of scaling protein analyses.

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

PRESENTATION – *Overview of the NHGRI Small Business Program – Dr. Renee Rider and Dr. Ian Nova*

Dr. Rider and Dr. Nova gave a presentation on the NHGRI Small Business Program, including what defines a small business, the application and funding process, an analysis of NHGRI's current Small Business portfolio, and future directions. The goals of the Small Business Program are to stimulate technological innovation, meet federal R&D needs, increase private sector commercialization of innovations, and foster and encourage participation by socially and economically disadvantaged persons, and women-owned small businesses. Small Business grants differ from research grants because they focus on product or service development. The two categories of Small Business Grants are Small Business Technology Transfer (STTR) and Small Business Innovation Research (SBIR). Council members were interested in the reasons why applications do not get funded (e.g., the technology is not ready or applicants are lacking skills in grantsmanship). Some outcomes of these small businesses include patent applications and commercialization. A project will begin soon to catalog the success stories of small businesses funded by NHGRI.

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

PRESENTATION – *Revisions for Fellowship Applications and Simplified Review Framework for Research Applications – Dr. Sarah Wheelan*

Dr. Wheelan presented information about the revisions to NIH applications and peer review that will go into effect in 2025, including a simplified review framework for most research project grants, improvements to the NRSA fellowship application and review process, and more. The simplified review framework is intended to reduce reviewer burden and will not affect applicants, but critiques and summary statements will be affected; Notice of Funding Opportunities (NOFOs) will be reissued to reflect the changes. The revisions to fellowship applications and review will focus on the candidate's scientific potential and research training plan to ensure that a broad range of candidates and research training contexts can be recognized as meritorious; NOFOs will be reissued to reflect the changes. There will also be changes to application instructions and training grant applications. Council members asked for clarification on some changes and commended the revisions. The triage process will likely not change.

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

COUNCIL-INITIATED DISCUSSION

Council members asked how more early-career investigators could serve as reviewers on NIH study sections. NHGRI staff commented that the Center for Scientific Review (CSR) has a program for early-career investigators. They get a reduced load of applications to review, and the Scientific Review Officer is careful to pair them with more experienced reviewers. The minimum criteria to be a reviewer is not defined by a specific policy, but most of the time reviewers have at least one R01.

The cost of educating the next generation of biomedical researchers is expensive. Housing costs are extremely expensive for fellows. There is no discussion at the NIH-level to address this problem.

Artificial Intelligence (AI) will displace individuals and change the workforce. In industry, AI is focusing on allowing a company to accelerate the time it takes to do research and to lower the cost. AI allows for information resources and datasets to be made available so that a more junior-level employee could direct and manage a research project, thereby reducing the overall cost of the process.

With H3Africa in close-out, NHGRI's global outreach should be reviewed at a future meeting.

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

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 284 applications, requesting a total of \$107,510,616 (direct costs). The applications included: 164 research project applications (R01, R03, R15 or R21); 57 research project cooperative agreement applications (U01); 9 education projects, cooperative agreement applications (UE5); 2 conference applications (R13); 5 education applications (R25); 3 community resource applications (U24); 16 career development applications (F99/K00, K99/R00, K01, or K08); 13 SBIR phase I applications (R43); 9 SBIR phase 2 applications (R44); 5 STTR phase one applications (R41); and 1 STTR phase II application (R42).

NHGRI STAFF PRESENT

Adam Felsenfeld, ERP

Afia Asare, ERP

Ajay Pillai, ERP
Alanna Kulchak Rahm, ERP
Alessandra Serrano Marroquin, ERP
Alexander Arguello, ERP
Alicia Caffi, ERP
Allison McCague, PPAB
Alvaro Encinas, OC
Amber Jackson, TiDHE
Anneliese Galczynski, ERP
Barbara Thomas, ERP
Ben Cubert, ERP
Carolyn Hutter, ERP
Chris Gunter, IOD
Chris Wellington, OGDS
Christine Chang, ERP
Colin Fletcher, ERP
Comfort Browne, ERP
Cristina Kapustij, PPAB
Daniel Gilchrist, ERP
Dave Kaufman, ERP
Deanna Ingersoll, ERP
Ebony Madden, TiDHE
Elena Ghanaim, OGDS
Elise Feingold, ERP
Enitza Rodriguez, ERP
Erin Ramos, ERP
Esperes Mfwilwakanda, ERP
Faye Brown, TiDHE
Heather Colley, ERP
Helen Thompson, OGDS
Ian Nova, ERP
Imani Boykin, ERP
Ismail Safi, ERP
Issel Lim, ERP
Jacob Jaffa, ITB
Jahnvi Narula, ERP
Jake Baroch, ERP
Jamil Scott, TiDHE
Jean Gao, OGDS
Jennifer Troyer, ERP
Jerryl Somani, ITB
Joannella Morales, ERP
Jyoti Dayal, ERP
Kayla Titilii-Torres, PPAB
Keith McKenney, ERP
Kimberly Ferguson, ERP
Kris Wetterstrand, ERP
Lawrence Brody, ERP
Lisa Chadwick, ERP
Lori Bonnycastle, IRP
Lucia Hindorff, TiDHE

Madji Lodoumgoto, OGDS
Marcia Morris, ERP
Maricela Trujillo, ERP
Maya VanZanten, ERP
Melinda Rose, ERP
Michelle Tallman, ERP
Mike Pazin, ERP
Molly Bird, PPAB
Monika Christman, ERP
Mukul Nerurkar, OC
Natalie Linear, ERP
Nephi Walton, ERP
Nicholas Nguyen, ERP
Nicolas Keller, OGDS
Nicole Lockhart, ERP
Rene Sterling, ERP
Renee Rider, ERP
Riley Wilson, ERP
Robb Rowley, ERP
Rongling Li, ERP
Sandhya Xirasagar, OGDS
Sara Currin, ERP
Sarah Anstice, ERP
Sarah Hutchinson, ERP
Sarah Wheelan, ERP
Sheethal Jose, ERP
Shurjo Sen, OGDS
Simona Volpi, ERP
Stephanie Morris, ERP
Susan Vasquez, IOD
Temesgen Fufa, ERP
Teri Manolio, ERP
Valentina Di Francesco, OGDS
Vanessa Campos, ERP
Vence Bonham, IOD
Weini Ogbagioris, TiDHE
Zephaun Harvey, ERP
Zo Bly, ERP

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 Jacob Baroch, NHGRI Scientific Program Analyst.

9/20/2024
Date

Rudy O. Pozzatti, Ph.D.

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

9/24/2024
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 September 9, 2024