

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH  
MEETING SUMMARY  
May 15-16, 2023**

The Open Session of the 99<sup>th</sup> meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:30 a.m. on Monday, May 15, 2023, with four 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 May 15, 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 May 15, 2023, and from 11:00 a.m. until adjournment on May 16, 2023, for the review, discussion, and evaluation of grant applications.

**COUNCIL MEMBERS PRESENT**

Joe Beery (LunaDNA)  
Laura Bierut (Washington University)  
Kyle Brothers (University of Louisville)  
Howard Chang (Stanford University)  
Nancy Cox (Vanderbilt 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)  
Peter Robinson (The Jackson Laboratory for Genomic Medicine)  
Olga Troyanskaya (Princeton University)

**AD HOC MEMBER**

Deepak Kumar (North Carolina Central 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 members of the NHGRI staff, liaisons, and guests, which can be viewed here: [\[link\]](#)

**APPROVAL OF MEETING SUMMARY**

The Council approved the February 13-14, 2023, Meeting Summary by a unanimous vote.

**FUTURE NACHGR MEETING DATES**

- September 18-19, 2023
- February 12-13, 2024
- May 20-21, 2024
- September 16-17, 2024
- February 24-25, 2025
- May 19-20, 2025

## **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 – NHGRI Intramural Research Program Report – Dr. Charles Rotimi**

Dr. Charles Rotimi, NHGRI Scientific Director, provided an overview of his personal and scientific background, then presented the goals, vision, and current activities of NHGRI's Intramural Research Program. In its 30<sup>th</sup> year since establishment, the NHGRI Intramural Research Program continues to advance the frontiers of genetics and genomics. In Dr. Rotimi's words, the NHGRI Intramural Research Program, "Aims to be world leaders in the translation of genomic knowledge into tools and approaches for improving the treatment, prognosis, and prevention of rare and common diseases." Dr. Rotimi explained what makes the NHGRI Intramural Research Program unique, highlighted a subset of its faculty members, indicated key accomplishments the program has made to date, and revealed areas where the program hopes to expand and improve.

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

## **PRESENTATION – NHGRI Intramural Research Program Blue Ribbon Panel Report – Council members Dr. Gail Jarvik and Dr. Lynn Jorde**

Drs. Jarvik and Jorde presented the third Blue Ribbon Panel (BRP) Report for the NHGRI Intramural Research Program since its establishment in 1993. This report is part of a trans-NIH requirement for each NIH Institute or Center's Intramural Research Program to undergo a review by a panel of outside experts roughly every ten years. This review panel consisted of eight experts with broad perspectives on genetics and genomics in addition to considerable knowledge of NHGRI.

Dr. Jorde briefed Council on the review process, which was comprised of five virtual BRP meetings, one in-person two-day meeting, numerous ad hoc meetings with various staff members, and close consideration of the five White Papers that highlighted "big questions" to be addressed by the Intramural Research Program. Dr. Jorde explained to Council that overall, the BRP had a highly favorable opinion of the NHGRI Intramural Research Program. Drs. Jorde and Jarvik presented the BRP report's findings in the areas of direction, faculty recruitment, collaboration, diversity, policies/procedures, and suggestions for new initiatives. Overall, the BRP found the NHGRI Intramural Research Program to be an impressive success. The faculty and trainees have been highly productive and the quality of the research has been outstanding. The dedication to diversity and inclusion was viewed as impressive, and the leadership has been excellent. The BRP looks forward to the next ten years of shared success.

Council members thanked the panel for their assessment and positive evaluation. The BRP Report is a public document available on the [NHGRI website](#).

A video of Drs. Jarvik and Jorde's presentation can be found here: [\[link\]](#)

## **PRESENTATION – NASEM Report on the Use of Race, Ethnicity, and Ancestry as Population Descriptors – Dr. Aravinda Chakravarti, NYU School of Medicine, and Dr. Charmaine Royal, Duke University**

In September 2021, NHGRI, along with 13 other NIH Institutes and Centers, asked the National Academies of Sciences, Engineering, and Medicine to conduct a study focused on the use of population descriptors in genetics and genomics research. In February 2022, a committee was convened by the National Academies and had its first meeting with NIH representatives. In March 2023, the committee released its report focused on the use of race, ethnicity, and other population descriptors in genetics and genomics research. The two co-chairs of the committee, Dr. Aravinda Chakravarti and Dr. Charmaine Royal, presented this report to Council.

Dr. Chakravarti explained that the committee's charge included assessing the use of race, ethnicity, and genetic ancestry in the basic science of genetics and genomics, developing approaches to advance the appropriate use of population descriptors, proposing best practices, discussing obstacles to implementation and adoption of best practices, and proposing potential implementation strategies. This report was necessary due to the urgent need to address issues surrounding race and racism in science and society, the increased accessibility of genomics data, the overarching aim for large-scale genomic studies to include more diverse groups of people, and the advanced methods of analyzing genomic variation. The report was divided into two sections: (1) Past and current use of population descriptors and (2) Recommendations.

Dr. Royal briefed Council on the recommendations developed by the committee. The committee generated 13 recommendations that fell into three categories: (1) Requisites, (2) Guidance for Researchers, and (3) Implementation and Accountability. Under the category of requisites, the committee found that NHGRI should avoid typological thinking, measure environmental factors, and engage communities and participants. Under the category of Guidance for Researchers, the committee suggested that NHGRI researchers should tailor their use of population descriptors to the type and purpose of their study. Under the category of Implementation and Accountability, the committee noted that the research ecosystem has many participants who individually and collectively share responsibility for making changes and helping researchers implement the report's recommendations.

Council received the report with enthusiasm and thanked the co-chairs for their work.

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

### **PRESENTATION – *Update on the H3Africa Program – Dr. Jennifer Troyer***

Dr. Troyer, a Program Director in NHGRI's Division of Genome Sciences, presented a summary of the Human Heredity and Health in Africa (H3Africa) program. H3Africa was an NIH Common Fund program that reached its tenth and final year of funding in 2022. NIH Common Fund programs are required to benefit all (or most) NIH institutes and centers. They are also catalytic and transformative in nature and are time-limited, with the expectation to be a high-risk, high-reward endeavors. NHGRI had a prominent role in the H3Africa program in terms of peer review, grants management, and program management.

The goal of H3Africa was to enable a genomics research revolution in Africa and allow African scientists to take ownership of African research. The program aimed to facilitate an African-based research approach to the study of genomic and environmental determinants of common diseases, with the goal of improving the health of African populations. To reach their goals, the program needed to improve capacity development and build collaborative networks. The H3Africa program was comprised of eight different project types: Biomedical Research Projects, Collaborative Research Centers, Biorepositories, Coordinating Center for ELSI Research

Projects, Collaborative ELSI Centers, Informatics Network (H3ABioNet), and Bioinformatics Training Program. Among the different projects, numerous working groups, and the steering committee, H3Africa created a diverse network of researchers focused in different areas. These groups conducted outreach to communities, governments, and local regions to promote the use of genomics in healthcare and in health research. The working groups produced public policies and guidelines used by some governments and regions as well as research tools and products that conducted phenotype harmonization, training programs, pharmacogenomics, and a genotyping array specific for African populations. In addition to the working groups of the consortia, there were numerous individual projects that produced a large amount of data.

A major outcome of the project was the set of over 700 H3Africa papers published to date. In addition to the publications, several resources were created from the project and are currently available. One resource is the bioinformatics network, H3ABionet. This resource connects 35 sites in 17 different countries with local expertise and jointly established competencies in genome data analysis, workflow development, and data management. In addition to H3ABionet, H3Africa created Biorepositories that are International Society for Biological and Environmental Repositories (ISBER) compliant and play a critical role in supporting the collection and storage of African biospecimens that are linked to diseases relevant to African populations. Finally, H3Africa established degree-granting programs across Africa with hundreds of successful graduates. There were also thousands of people trained through classes, courses, workshops, and webinars run by different groups within H3Africa.

H3Africa demonstrated that African genomics cannot be effectively conducted without African genomes and African scientists. The World Health Organization is now considering genomics for global health in low- and middle-income settings, and there are developing plans for networks of genomics centers of excellence across Africa.

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

### **CONCEPT CLEARANCE – RFA: Genomics-Enabled Learning Health Systems – Dr. Robb Rowley**

Dr. Rowley, a Program Director in NHGRI's Division of Genomic Medicine, presented the Network of Genomics-Enabled Health Systems (gLHS) RFA concept. This concept aims to establish a network of institutions with a track record of using gLHS approaches in their health system and developing these practices into implementation resources. This concept will identify two to four implementation projects and pursue those projects network-wide. The implementation projects will be used to increase system-wide and across systems interoperability and refine resources for broader sharing. Validated tools and resources will also be established for sites implementing gLHS. The program proposed providing \$5.3M in funds over five years and will issue two U01 RFAs, one for a single coordinating center and one to develop four to six clinical sites. Council members were supportive of the concept, especially given its alignment with the 2020 NHGRI Strategic Vision. Council members noted the opportunity to synergize other NHGRI-funded programs like the AnVIL platform. They also suggested partnering with other institutions to increase the number of clinical sites and the creation of an advisory panel with stakeholders in the program.

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

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

## **CONCEPT CLEARANCE – PAR: Developing Novel Theory and Methods for Understanding the Genetic Architecture of Complex Traits – Dr. Alexander Arguello**

Dr. Arguello, a Program Director in NHGRI's Division of Genome Sciences, presented the Developing Novel Theory and Methods for Understanding the Genetic Architecture of Complex Traits PAR. This concept aims to support novel theory and methods development for genetic and non-genetic sources of complex trait variation across individuals, families, and populations. Council members were enthusiastic about the concept and the opportunities it would present to use existing data. Council suggested pointing applicants to specific datasets in the PAR.

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

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

## **COUNCIL-INITIATED DISCUSSION**

Council members expressed interest in hearing about the professional development and general well-being of NHGRI-supported students and trainees impacted by the COVID-19 pandemic. Additionally, they noted the disproportional effects of the pandemic related to gender, race, and other categories on the students, trainees, and junior investigators who have been supported by NHGRI. Council members asked NHGRI to determine how these individuals have been affected and discussed ways in which NHGRI can best support them.

Council members suggested inviting the National Center for Advancing Translational Sciences (NCATS) Director, Dr. Joni Rutter, to present at a future Council meeting.

Given NHGRI's leadership role in data access, and the nature of the concepts presented during this Council meeting, Council members would like to hear from an expert in the field who could discuss genomic data access and NHGRI's evolving strategy for data privacy and sharing.

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 226 applications, requesting a total of \$150,163,814 (direct costs). The applications included: 109 research project applications (R01, R03, or R21); 3 conference applications (R13); 67 cooperative agreement (U01) applications; 3 multi-project center cooperative agreement applications (U54); one UG3 cooperative agreement application; 7 career development applications (K01 or K08); 10 community resource applications (U24); 7 career development applications (F99/K00 or K99/R00); 10 SBIR Phase I application (R43); 5 SBIR Phase II applications (R44); 2 STTR Phase 1 applications (R41); and 1 STTR Phase 2 applications (R42).

## **NHGRI STAFF PRESENT**

Alexander Arguello, ERP  
Katie Bardsley, ERP  
Jake Baroch, ERP  
Sarah Bates, OC  
Zo Bly, ERP  
Vence Bonham, IOD  
Comfort Browne, ERP  
Gloria Butler, DM  
Alicia Caffi, ERP  
Stephanie Calluori, ERP  
Lisa Chadwick, ERP  
Christine Chang, ERP  
Paul Cheung, DM  
Monika Christman, ERP  
Heather Colley, ERP  
Sara Currin, ERP  
Stephanie Davis (on detail from NHLBI)  
Jyoti Dayal, ERP  
Christina Daulton, ECIB  
Edith DeHaut, ERP  
Christopher Donohue, OC  
Alvaro Encinas, OC  
Valentina Di Francesco, OGDS  
Elise Feingold, ERP  
Kimberly Ferguson, ERP  
Adam Felsenfeld, ERP  
Colin Fletcher, ERP  
Temesgen Fufa, ERP  
Elena Ghanaim, OGDS  
Daniel Gilchrist, ERP  
Bettie Graham, ERP  
Chris Gunter, IOD  
Peggy Hall, ERP  
Zephaun Harvey, ERP  
Lucia Hindorff, TiDHE  
Sarah Hutchinson, ERP  
Deanna Ingersoll, ERP  
Amber Jackson, TiDHE  
Cristina Kapustij, PPAB  
Dave Kaufman, ERP  
Jim Kees, ERP  
Se Rin (Julie) Kim, OGDS  
Mike Lacy, ERP  
Rongling Li, ERP  
Natalie Linear, ERP

Nicole Lockhart, ERP  
Ebony Madden, TiDHE  
Iman Martin, ERP  
William Maye, ITB  
Allison McCague, PPAB  
Imani McGregor, ERP  
Keith McKenney, ERP  
Donna Messersmith, ECIB  
Patricia Messick, IRP  
Julius Militante, ERP  
Joannella Morales, ERP  
Stephanie Morris, ERP  
Jahnavi Narula, ERP  
Nguyen Park, ERP  
Mike Pazin, ERP  
Ajay Pillai, ERP  
Caprina Pipion, ERP  
Colette Pollard, OGDS  
Sangeetha Raghavan, ERP  
Renee Rider, ERP  
Enitza Rodriguez, ERP  
Ellen Rolfes, DM  
Robb Rowley, ERP  
Ismail Safi, ERP  
Jamil Scott, TiDHE  
Shurjo Sen, OGDS  
Heidi Sofia, ERP  
Jerryl Somani, ITB  
Sonja Soo, OC  
Rene Sterling, ERP  
Jennifer Strasburger, ERP  
Michelle Tallman, ERP  
Barbara Thomas, ERP  
Helen Thompson, OGDS  
Kayla Titalii-Torres, PPAB  
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 Helen Thompson, NHGRI Scientific Program Analyst.*

09/20/2023

Date

*Rudy Pozzatti, Ph.D.*

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

09/20/2023

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 September 18, 2023