

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH  
MEETING SUMMARY  
September 18-19, 2023**

The Open Session of the 100<sup>th</sup> meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:30 a.m. on September 18, 2023, with 13 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 September 18, 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 September 18, 2023, and from 11:00 a.m. until adjournment on September 19, 2023, for the review, discussion, and evaluation of grant applications.

**COUNCIL MEMBERS PRESENT**

Laura Bierut (Washington University)  
Iftikhar Kullo (Mayo Clinic)  
Nancy Cox (Vanderbilt University)  
Lynn Jorde (University of Utah)  
Gail Jarvik (University of Washington)  
Judy Cho (The Icahn School of Medicine at Mount Sinai Hospital)  
Timothy Reddy (Duke University)  
Kyle Brothers (University of Louisville)  
Lisa Parker (University of Pittsburgh)  
Peter Robinson (The Jackson Laboratory)  
Joe Beery (LunaDNA)  
Len Pennacchio (Lawrence Berkeley National Laboratory)  
Howard Chang (Stanford 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 May 15-16, 2023, Meeting Summary by a unanimous vote.

**FUTURE NACHGR MEETING DATES**

- February 12-13, 2024
- May 20-21, 2024
- September 9-10, 2024
- February 24-25, 2025
- May 19-20, 2025
- September 8-9, 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, [slides](#) and related documents are available here: [\[link\]](#)

## **Commemorating the 100<sup>th</sup> Meeting of NACHGR**

The September 2023 Council Meeting marked the 100<sup>th</sup> meeting of NACHGR. Dr. Eric Green was joined by Dr. Francis Collins, former Director of NHGRI and NIH to commemorate this milestone.

A video of the commemoration can be found here: [\[link\]](#)

## **Honoring Bettie Graham**

After 52 years of federal service, Dr. Bettie J. Graham is retiring from her role as Director of the Division of Extramural Operations (DEO) at NHGRI. To honor her unparalleled commitment to the genomics research community, the Outstanding NHGRI Staff Award for DEIA in the Genomics Workforce will be renamed to the Bettie J. Graham Leadership Award.

Mr. Vence Bonham delivered comments from the research community praising Dr. Graham for the impact she has made during her time at the institute. Dr. Francis Collins continued the celebration with a song dedicated to Dr. Graham.

A video of the commemoration can be found here: [\[link\]](#)

## **PRESENTATION – *Catalyzing Data Science in Research Through Collaborations – Dr. Susan Gregurick***

Dr. Gregurick is the Associate Director for Data Science and the Director of the Office of Data Science Strategy (ODSS) at NIH. She presented an overview of the goals of ODSS, followed by NIH-wide efforts in: 1) creating Findable, Accessible, Interoperable, and Reusable (FAIR) Data; 2) developing and sustaining software; 3) cloud utilization; 4) federating NIH data platforms, and 5) artificial intelligence. Dr. Gregurick highlighted the many funding opportunities that have and will continue to support data science initiatives in the intramural and extramural research communities. She also pointed out collaborations across the data ecosystem that NHGRI is involved in, such as the NIH Cloud Platform Interoperability (NCPI) effort that integrates with the NHGRI Analysis, Visualization, and Informatics Lab-Space (AnVIL). Council members were interested in learning about the accessibility of results in addition to data, the integration of ethical, legal, and social implications (ELSI) research, the use of Large Language Models (LLMs), and resources needed to support data infrastructure.

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

## **CONCEPT CLEARANCE – *Machine Learning and Artificial Intelligence Tools to Advance Genomic Translational Research – Dr. Sandhya Xirasagar***

Dr. Xirasagar presented the concept for *Machine Learning and Artificial Intelligence Tools to Advance Genomic Translation Research*. The research from this notice of funding opportunity (NOFO) will carry out the objective of spurring development of novel ML/AI tools to explore their potential in advancing genomic translational research. As Dr. Xirasagar emphasized, the increasing implementation of ML/AI in biomedical research indicates a need for a dedicated effort to understand applicability and translate findings into medicine with ELSI considerations. The program timeline will include a design phase and a validation phase; the goal is to fund 3-4 sites and a coordinating center to design and develop tools according to a standardized ELSI and FAIR data framework. Council members were supportive of the proposed program as it is

forward-thinking, incorporates ELSI research, and can leverage existing datasets across NHGRI. There was discussion surrounding effective ways to embed ELSI research, integrating the work of groups focused on different diseases as a first target, and the potential of receiving applications with narrow phenotypic or multi-omic data.

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

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

**CONCEPT CLEARANCE – RFA Renewal: NHGRI Technology Development Coordinating Center – Dr. Stephanie Morris**

Dr. Morris sought concept approval of an RFA for the renewal of the NHGRI Technology Development Coordinating Center (TDCC). The TDCC is responsible for coordinating technology development initiatives across NHGRI, including the Genome Technology Program (GTP) and Centers of Excellence in Genomic Science (CEGS) program. The four-year award was first launched in 2021 with the goal of the center being to enhance integration across the multi-component GTP. Since then, the TDCC has: 1) facilitated opportunities for collaborations, 2) developed outreach strategies and resources, and 3) supported innovative small-scale work. The renewal will continue to build on these existing activities. One five-year U24 award will be made in Fiscal Year 2025; the \$1.5 million total cost per year budget will include \$750k per year for the Opportunity Funds Program to support pilot projects in priority areas. Council members showed support for the renewal, given its success over the past two years. Discussion revolved around communication between GTP and CEGS, as well as the use of the opportunity funds funding mechanism.

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

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

**CONCEPT CLEARANCE – Genomic Medicine eConsult Service – Ms. Renee Rider**

Ms. Rider presented the concept for an RFA that will support implementation and evaluation of clinician-to-clinician genomic medicine eConsult services. The eConsult service is intended to promote delivery of actionable recommendations, improved care and health equity while decreasing patient burden. In the current landscape, many services are confined to single institutions, and few exist with a genomic medicine specialization. Ms. Rider explained that the proposal is to fund two sites that will test diverse service models, identify local barriers, and collaborate to discuss metrics and solutions to obstacles. NHGRI will fund the sites for five years and will seek co-funding from other NIH institutes and centers and federal agencies to expand the number of sites, project scope, and evaluation of the service. Council members emphasized that this proposal addresses an important gap in the U.S. healthcare system and will improve health access and equity. Questions arose about licensing implications, sustainability of the model, and the logistics of achieving 1000 consults a year.

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

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

**CONCEPT CLEARANCE** – *PAR Renewal: Genomic Research Experiences for Data Scientists*  
– Dr. Sandhya Xirasagar

Dr. Xirasagar introduced the *Genomics Research Experiences for Data Scientists* Program Announcement with special receipt, referral or review considerations (PAR) renewal. The goal of the PAR is to complement educational activities offered by master's programs in data science fields and bring early-stage data scientists into genomics research through experimental and computational research activities. It will be reissued as an R25 activity code for three years with one receipt date per year. The application budget limit is \$250,000 direct cost per year, not exceeding five years. Council members were enthusiastic and expressed support for the renewal, noting they look forward to the outcomes.

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

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

**CONCEPT CLEARANCE** – *PAR Renewal: Research Experiences in Genomic Research for Genetic Counselors* – Ms. Heather Colley

Ms. Colley sought approval for the renewal of the *Research Experiences in Genomic Research for Genetic Counselors* PAR. The purpose of the initiative is to foster the involvement of graduates of genetic counseling master's programs in genomics research and support their development as members of a research group. Through the R25 mechanism, awardees will receive up to \$250,000 total cost per year to support a minimum of two trainees with a 50% commitment to research for 6-24 months. Discussion about the renewal highlighted excitement for the training opportunity. Council members suggested flexibility about the time commitment, particularly in cases where genetic counselors may want to commit more time to research. They also raised the idea of awarding certificates to trainees who complete the program, though it was noted that certification criteria may vary by institution.

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

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

**REPORT** – *Genomics & Society Working Group of Council Annual Report* – Dr. Malia Fullerton

Dr. Fullerton is a co-chair of the Genomics & Society Working Group (GSWG), which provides advice on priorities for genomics and society initiatives at NHGRI, particularly within the ELSI Research Program. In the 2023 annual report, the GSWG addressed insights about ELSI research and its role in addressing structural inequities, as well as the need to broaden ELSI expertise to include new perspectives. Continuing priorities for the group include: 1) health equity; 2) structural factors and social justice; 3) gene and environment interaction research, and 4) social determinants of health. New priorities focus on diversity in the ELSI research portfolio, as well as next steps following the National Academies of Sciences, Engineering and Medicine (NASEM) Population Descriptors Report. Regarding the NASEM report, the GSWG advised that Council consider requiring applicants to explain their use of population descriptors, how to incentivize the adoption of NASEM recommendations, and support novel research on new frameworks and methods related to genetic similarity.

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

**PRESENTATION** – *Office of Communications and History of Genomics Program Update* – Ms. Sarah Bates and Dr. Christopher Donohue

Ms. Bates, NHGRI Communications Director, and Dr. Donohue, NHGRI Historian, provided overviews of the Office of Communications (OC) and the History of Genomics Program. As Ms. Bates highlighted, science communication is integral to NHGRI’s strategy and must be considered within social, historical, and ethical contexts. The OC’s goal is to utilize a variety of platforms to inform audiences about the importance of genomics and research; such work has earned recognition within and outside NIH for creative communication efforts. Dr. Donohue followed with a description of the History of Genomics Program, which leverages the unique resources of NHGRI and the OC to develop oral histories, digital archives, and lectures that capture the history of NHGRI and the field of genomics. The millions of artifacts digitized has resulted in NHGRI having the largest digital archive in biology, which continues to grow as more documents and information are added. In building the archive, the program applies FAIR principles and seeks to make files and datasets accessible to the public through resources like ArchivesSpace and SharePoint. Council members were excited to hear about the initiatives of the OC and History of Genomics Program, and suggested opportunities for engaging with archival efforts in other fields.

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

**PRESENTATION** – *Using Machine Learning and Artificial Intelligence Methods in History of Genomics Archival Research* – Spencer Hong and Thomas Stoeger

Mr. Hong and Dr. Stoeger from the laboratory of Dr. Luis Amaral at Northwestern University delivered a presentation on ML/AI approaches to enrich archival research in the history of genomics. Their work engages domain expertise, computational methods, and privacy protection to generate knowledge and data products. Across a diverse set of documents, the computational approaches can identify the type, content, handwriting, and potentially personally identifiable information. The researchers have used the tools to develop communication networks that track the dynamic nature of NHGRI’s history and interactions across and outside the institute. Council members commended the researchers on the work they presented and encouraged them to explore connections with other institutes and fields to fully understand the reach genomics has had on biomedical research and society.

A video of Mr. Hong’s, and Dr. Stoeger’s presentation and the related discussion can be found here: [\[link\]](#)

**REPORT** – *Clinical Sequencing Evidence-Generating Research (CSER)* – Gail Jarvik and Kyle Brothers

Council members Dr. Jarvik and Dr. Brothers presented a report on the Clinical Sequencing Evidence Generating Research (CSER) program, which concluded in 2023. In Phase 1 (2011-2016), the goal of CSER was to explore the applications of genomic sequence data to the care of patients within active clinical settings and develop best practices and approaches. In the following phase (2017-2023), CSER moved from “exploratory” to “evidence-generating” research, to generate evidence to determine the clinical utility of genome sequencing. Tools developed by the consortium include a comprehensive guide to interpreting genomic reports, a pathogenicity calculator, and other research resources that have been migrated to the NHGRI

AnVIL platform. Dr. Jarvik detailed Phase 1 progress in the investigation of additional findings, testing the ACMG/AMP guidelines for variant pathogenicity classification, and developing guidance on informed consent documents and responsible return of results. Dr. Brothers highlighted Phase 2 progress in harmonized measures for clinical utility, participant diversity and inclusion, and perceived utility of patients and their families. In addition to the 327 site-specific papers, the legacy of CSER in improving the practice of genomic medicine is illustrated by key evidence generated across the phases in genomic variant interpretation, clinical management, and adaptability into diverse care settings. Council members were excited to hear about the consortium's progress and achievements, pointing out that CSER is a prime example of how genomic medicine can be successfully implemented to achieve NHGRI's strategic vision.

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

## **COUNCIL-INITIATED DISCUSSION**

There was discussion around the promotion and accessibility of the NIH *All of Us* Research Program resource, as receiving access to non-public data for educational purposes can be burdensome. NHGRI staff commented that a trans-NIH liaison committee has hosted discussion on lowering the barrier to entry for some data types; similarly, the *All of Us* Research Program has multiple funding opportunities available to encourage use of the extensive datasets.

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

## **REVIEW OF APPLICATIONS**

In the Closed Session, the NACHGR reviewed 180 applications, requesting a total of \$65,763,869 (direct costs). The applications included: 135 research project applications (R01, R03, R15 or R21); 1 conference application (R13); 5 education applications (R25); 6 Community Resource applications (U24); 14 Career Development applications (F99/K00, K99/R00, K01, or K18); 12 SBIR Phase I application (R43), and 7 SBIR Phase II applications (R44).

## **NHGRI STAFF PRESENT**

Adam Felsenfeld, ERP  
Afia Asare, ERP  
Ajay Pillai, ERP  
Alessandra Serrano Marroquin, ERP  
Alexander Arguello, ERP  
Allison McCague, PPAB  
Alvaro Encinas, OC  
Amber Jackson, TIDHE  
Ann Fitzpatrick, DM  
Anna Rogers, OC  
Anneliese Galczynski, ERP  
Ava Miller, IRP  
Barbara Thomas, ERP  
Ben Cubert, ERP  
Beth Tuck, ECIB  
Bettie Graham, ERP

Britny Kish, OC  
Caprina Pipion, ERP  
Carolyn Hutter, ERP  
Chris Gunter, IOD  
Chris Wellington, OGDS  
Christina Daulton, TIDHE  
Christine Chang, ERP  
Colette Pollard, OGDS  
Colin Fletcher, ERP  
Comfort Browne, ERP  
Cristina Kapustij, PPAB  
Daniel Gilchrist, ERP  
Danielle Buice, IRP  
Darryl Leja, OC  
Dave Kaufman, ERP  
Deanna Ingersoll, ERP

Devon Bumbray-Quarles, ERP  
Donna Morris, ERP  
Ebony Madden, TiDHE  
Edith DeHaut, ERP  
Eggerton Campbell ERP  
Elena Ghanaim, OGDS  
Elise Feingold, ERP  
Ellaha Sadat, OC  
Ellen Rolfes, DM  
Emily Neveux, DM  
Enitza Rodriguez, ERP  
Erin Ramos, ERP  
Ernesto Del Aguila, OC  
Esperes Mfwilwakanda, ERP  
Faye Brown, TiDHE  
Gloria Butler, DM  
Heather Colley, ERP  
Helen Thompson, OGDS  
Ian Nova, ERP  
Iman Martin, ERP  
Ismail Safi, ERP  
Jahnvi Narula, ERP  
Jake Baroch, ERP  
Jamil Scott, TiDHE  
Jenn Montooth, OC  
Jennifer Strasburger, ERP  
Jennifer Troyer, ERP  
Jerryl Somani, ITB  
Jessica Chong, ERP  
Jim Kees, ERP  
Joannella Morales, ERP  
Juila Fekecs, OC  
Jyoti Dayal, ERP  
Karyn Roberts, ERP  
Kenya Smith, OC  
Kimberly Ferguson, ERP  
Kris Wetterstrand, IOD  
Lawrence Brody, ERP  
Lisa Chadwick, ERP  
Liz Dietz, OC  
Lorjetta Schools, TiDHE  
Lucia Hindorff, TiDHE  
Madji Lodoumgoto, OGDS  
Marcia Morris, ERP  
Maricela Trujillo, ERP

Mauresa Pittman, OC  
Maya VanZanten, ERP  
Michelle Tallman, ERP  
Mike Pazin, ERP  
Molly Bird, PPAB  
Monika Christman, ERP  
Mukul Nerurkar, OC  
Natalie Linear, ERP  
Nephi Walton, ERP  
Nguyen Park, ERP  
Nicholas Keller, OGDS  
Nicholas Nguyen, ERP  
Nicola Sugden, OC  
Nicole Lockhart, ERP  
Paul Cheung, DM  
Peggy Hall, ERP  
Radu Marin, ITB  
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 Bates, OC  
Sarah Hutchinson, ERP  
Sarah Wheelan, ERP  
Shurjo Sen, OGDS  
Simona Volpi, ERP  
Sonja Soo, OC  
Stephanie Morris, ERP  
Susan Vasquez, IOD  
Temesgen Fufa, ERP  
Teri Manolio, ERP  
Theo Tiffney, OC  
Valentina Di Francesco, OGDS  
Vanessa Campos, ERP  
Vence Bonham, IOD  
Weini Ogbagioris, TiDHE  
William Kibby, ITB  
William Maye, ITB  
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 Maya VanZanten, NHGRI Scientific Program Analyst.*

02/12/2024

Date

*Rudy Pozzatti, Ph.D.*

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

02/12/2024

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 February 12, 2024**