

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
September 19-20, 2022

The Open Session of the 97th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:00 a.m. on Monday, September 19, 2022. Nine Council members attended the meeting in person; seven members participated 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 10:00 a.m. – 5:30 p.m. on September 19, 2022. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 9:00 a.m. – 9:30 a.m. on September 19, 2022, and from 11:00 a.m. until adjournment on September 20, 2022, 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 School of Medicine)
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)
Stephen Rich (University of Virginia)
Peter Robinson (The Jackson Laboratory for Genomic Medicine)
Olga Troyanskaya (Princeton University)

AD HOC MEMBERS

Judy Cho (The Icahn School of Medicine at Mount Sinai Hospital)
Nancy Cox (Vanderbilt University)
Timothy Reddy (Duke 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 16, 2022, Meeting Summary by a unanimous vote.

FUTURE NACHGR MEETING DATES

- February 13-14, 2023
- 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 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 – NIAMS Research: Focus on Genomics and Genetics – Dr. Lindsey Criswell

Dr. Lindsey Criswell, Director of the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), presented an overview of ongoing research activities supported by NIAMS, highlighting advances in genetics and genomics and opportunities for collaboration. After a brief overview of the NIAMS mission, budget, and history, Dr. Criswell described the NIAMS current Strategic Plan for Fiscal Years 2020 to 2024 which promotes development of patient-centered, personalized ways to improve health outcomes in the core areas of the NIAMS mission. NIAMS and NHGRI are collaborating on several ongoing projects including the Genome Research in African American Scleroderma Patients (GRASP) Consortium and Accelerating Medicines Partnership (AMP) Bespoke Gene Therapy Consortium. Data science is another ideal area for collaboration between the two institutes, and Dr. Eric Green highlighted the opportunity to connect data science working groups that are operational at both NHGRI and NIAMS.

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

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

Dr. Malia Fullerton, Chair of the NACHGR Genomics and Society Working Group, presented an annual report of the working group's activities. The Genomics and Society Working Group provides advice on short- and long-term planning and priority setting for genomics and society research activities supported by NHGRI, with particular focus on the Ethical, Legal, and Social Implications (ELSI) Research Program. Dr. Fullerton highlighted emerging ELSI research priorities in the areas of social determinants of health, health equity, structural factors and social justice, gene-environment interaction research, and genomics beyond healthcare. ELSI research is needed to guide efforts to address structural inequalities in genomics research. Council members highlighted the importance of community involvement and looking to disadvantaged communities as a source of inspiration and solutions rather than a locus of study because of unfortunate circumstances.

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

CONCEPT CLEARANCE – Genomic Community Resources PAR Renewal – Mr. Chris Wellington

Mr. Chris Wellington presented renewal plans for the Genomic Community Resources Program Announcement with Special Receipt, Referral or Review considerations (PAR), a longstanding grant program at NHGRI. Projects that could be supported by this PAR include genomic informatics resources, comprehensive identification and collections of genomic features, resources to support genomics community-building efforts, and sample repositories for genomic studies. The renewal plan proposes the U24 PAR be reissued for three years with three receipt dates per year. Council members emphasized the need for the program; however, they also raised questions about challenges with review of resource grants as well as maintenance costs.

There were 16 votes to approve the concept, no objections, and no abstentions. A video of Mr. Wellington's presentation and the related discussion can be found here: [\[link\]](#)

CONCEPT CLEARANCE – Genomics and Health Equity Initiatives RFAs – Dr. Lucia Hindorff

Dr. Lucia Hindorff presented the Investigator-Initiated Research in Genomics and Health Equity concept which includes four Request for Applications (RFAs) for R01 and R21 projects. The concept is guided by the need to maximize the usability of genomics for all members of the public, including the ability to access genomics in healthcare, as described in NHGRI's 2020 Strategic Vision. A Future Directions in Genomics and Health Equity Virtual Workshop held in April 2022 helped inform this concept. The concept aims to support investigator-initiated research in genomics and health equity across NHGRI's scientific areas through the development of approaches, dissemination of data, and implementation of metrics or interventions, with the goal of advancing the equitable use of genomics to improve health in all U.S. populations. Council members were enthusiastic about this concept and mentioned the importance of tracking outcomes to measure impact.

There were 16 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 – Entry Level Training Modules (ELM) PAR – Ms. Renee Rider

Ms. Renee Rider presented the Entry Level Training Modules (ELM) concept, which aims to increase genomics knowledge in the entry-level genomics research workforce and enhance diversity within the field. The concept employs modules, stand-alone units of curricula that supplement existing training, developed by lead sites and used for genomics education at partner institutions. A total cost of \$1.8M is proposed to support three R25 awards for three years. Council members were supportive of this concept and suggested tracking outcomes and using the NHGRI website to share the educational content developed by these grants. Questions were raised about the lifespan of the modules, but the staff believes the grantees will be self-motivated to update the content.

There were 16 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 – Center for Inherited Disease Research; Contract Renewal – Dr. Lawrence Brody

Dr. Larry Brody presented a proposed renewal of the Center for Inherited Disease Research (CIDR), which was founded in 1996. CIDR is an integrated program that reviews and carries out large-scale genomic research projects for investigators. The facility has flexibility as to what genomic services are offered and is structured so that it may be closed if its services are no longer required. CIDR is currently supported by 10 NIH institutes and provides shared expertise, governance, resources, and projects. NHGRI's support is in-kind through personnel. Council members highlighted the strength of the wrap-around services that make CIDR unique and productive. They also suggested that additional efforts to disseminate information about the program could be useful. Questions were raised about potential CLIA certification issues and tracking the prevalence of samples from underrepresented populations.

There were 16 votes to approve the concept, no objections, and no abstentions. A video of Dr. Brody's presentation and the related discussion can be found here: [\[link\]](#)

REPORT – Human Genome Reference Program Update – Dr. Deanna Church and Dr. Martin Hirst

Drs. Deanna Church and Martin Hirst, members of the Human Pangenome Reference Consortium's (HPRC) Scientific Advisory Board, presented an update on the activities and progress of the Human Genome Reference Program. The consortium aims to generate a

reference human genome sequence that is more inclusive and representative of the diversity of the world's population, called a pangenome, while prioritizing quality and developing a new, non-linear genome reference structure. The team has developed a multi-center genome sequencing effort optimized for efficiency and quality. They have harnessed the potential of ultra-long reads with innovative long-read assembly methods and automated assembly standards. The HPRC is establishing a framework to define diversity and prioritize the study of cell lines from globally diverse participants to enable the generation of a truly representative pangenome. Implementation and adoption of the Pangenome model will be additional challenges.

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

WORKSHOP REPORT – *Capturing RNA Sequences & Transcript Diversity Workshop – Dr. Jennifer Strasburger and Dr. Brenton Graveley*

Dr. Jennifer Strasburger, a Program Director in the Division of Genome Sciences, and Dr. Brenton Graveley, the Chair of the Department of Genetics and Genome Sciences and Associate Director of the Institute for Systems Genomics at UConn Health, provided an overview of a May 2022 workshop titled, "Capturing RNA Sequence and Transcript Diversity, from Technology Innovation to Clinical Application." The goal of the workshop was to determine current capabilities, needs, and prospects for comprehensive characterization and understanding of the true diversity of all RNAs and their modifications at a chemical and structural level in relation to normal and disease states. Opportunities were identified in RNA sequencing, the study of RNA structure, RNA-binding proteins, RNA biology, and the use of RNA as a therapeutic and drug target. Materials from the workshop can be found on the meeting website [here](#).

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

COUNCIL-INITIATED DISCUSSION

Council members expressed the view that the NIH needs to take on a bigger role in educating the community about the risks of sharing data, as well as how progress in biomedical research is hindered when data are not shared. A balanced approach is essential for ethical and legal data sharing that builds trust. Additional efforts are needed to gather specific information from the community about the challenges of data sharing.

It was suggested that NHGRI consider establishing a single resource for reprocessing of genomic variants. Currently there is a huge and repetitive burden on investigators, and it is worth considering how data are stored, shared, and provided.

Council members raised the issue of the current shortage of applicants to fill the available positions for postdoctoral fellows.

Dr. Stephen Sherry, the new Director of the National Center for Biotechnology Information was suggested as a speaker for a future Council meeting. A presentation on where NHGRI may be able to lead in the field of gene therapy was also suggested.

Council members voiced the value of consistent publication of public access policies across NIH and federal funding agencies. The effects of open access also need to be seriously evaluated.

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 207 applications, requesting \$75,891,708 (direct costs). The applications included: 158 research project applications (R01, R03, or R21); 1 R15 application; 2 Conference applications (R13); 10 Education applications (R25); 6 Community Resource applications (U24); 13 Career Development applications (F99/K00, K99/R00, K01, K08, or K18); 10 SBIR Phase I application (R43); 6 SBIR Phase II applications (R44); and 1 STTR Phase 1 application.

NHGRI STAFF PRESENT

Alexander Arguello, ERP
Katie Bardsley, ERP
Jake Baroch, ERP
Sarah Bates, OC
Zo Bly, ERP
Vence Bonham, IOD
Joy Boyer, ERP
Lawrence Brody, ERP
Comfort Browne, ERP
Devon Bumbray-Quarles, ERP
Alicia Caffi, DM
Stephanie Calluori, ERP
Lisa Chadwick, ERP
Christine Chang, ERP
Monika Christman, ERP
Heather Colley, ERP
Amanda Conti, PPAB
Sara Currin, ERP
Jyoti Dayal, ERP
Edith DeHaut, ERP
Valentina Di Francesco, OGDS
Argenia Doss, ERP
Alvaro Encinas, OC
Elise Feingold, ERP
Kimberly Ferguson, ERP
Adam Felsenfeld, ERP
Ann Fitzpatrick, DM
Colin Fletcher, ERP
Brianna Foster, ERP
Anneliese Galczynski, ERP
Elena Ghanaim, OGDS
Daniel Gilchrist, ERP
Jenell Glover, ERP
Bettie Graham, ERP
Chris Gunter, IOD

Peggy Hall, ERP
Zephaun Harvey, ERP
Joe Henke, ITB
Lucia Hindorff, TIDHE
Albert Hinman, ERP
Sarah Hutchinson, ERP
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Amber Jackson, TIDHE
Cristina Kapustij, PPAB
Dave Kaufman, ERP
Jim Kees, ERP
Se Rin (Julie) Kim, OGDS
Michael Lacy, DM
Rongling Li, ERP
Asiyah Lin, OGDS
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
Keith McKenney, ERP
Donna Messersmith, ECIB
Jenn Montooth, OC
Joannella Morales, ERP
Donna Morris, ERP
Marcia Morris, ERP
Stephanie Morris, ERP
Jahnavi Narula, ERP
Mukul Nerurkar, OC
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
Melinda Rose, ERP
Kristen Ross, ERP
Robb Rowley, ERP
Ismail Safi, ERP
Ella Samer, ERP
Lorjetta Schools, TiDHE
Jamil Scott, TiDHE
Shurjo Sen, OGDS
Michael Smith, ERP

Heidi Sofia, ERP
Jerryl Somani, ITB
Sonja Soo, OC
Rene Sterling, ERP
Jennifer Strasburger, ERP
Michelle Tallman, ERP
Jill Thomas, OC
Barbara Thomas, ERP
Helen Thompson, OGDS
Jennifer Troyer, ERP
Maya VanZanten, ERP
Susan Vasquez, IOD
Simona Volpi, ERP
Chris Wellington, OGDS
Kris Wetterstrand, IOD
Ken Wiley, ERP
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 Katie Bardsley, NHGRI Scientific Program Analyst.

02/14/2023

Date

Rudy Pozzatti, Ph.D.

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

02/17/2023

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 February 14, 2023