

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH  
MEETING SUMMARY  
February 7-8, 2022**

The Open Session of the 95<sup>th</sup> meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened virtually by Zoom at 11:30 a.m. on Monday, February 7, 2022. 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 11:30 a.m. – 5:00 p.m. on February 7, 2022. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 10:00 a.m. – 11:00 a.m. on February 7, 2022 and from 11:00 a.m. until adjournment on September 8, 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)  
Wendy Chung (Columbia University)  
Mark Craven (University of Wisconsin, Madison)  
Patricia Deverka (University of California, San Francisco)  
Hal Dietz (Johns Hopkins University)  
Stephen Fodor (13.8 Inc.)  
Rafael Irizarry (Dana-Farber Cancer Institute)  
Gail Jarvik (University of Washington)  
Lynn Jorde (University of Utah)  
Iftikhar Kullo (Mayo Clinic)  
Lisa Parker (University of Pittsburgh)  
Len Pennacchio (University of California, Berkeley)  
Stephen Rich (University of Virginia)  
Peter Robinson (The Jackson Laboratory for Genomic Medicine)  
Olga Troyanskaya (Princeton University)

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

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

**APPROVAL OF MEETING SUMMARY**

The Council approved the September 2021 Meeting Summary by a unanimous vote.

**FUTURE NACHGR MEETING DATES**

- May 16-17, 2022
- September 19-20, 2022
- February 13-14, 2023
- May 15-16, 2023
- September 18-19, 2023

## **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** – *Expanding the Capacity to Conduct Gene x Environment Experiments* – Dr. Rick Woychik

Dr. Richard (Rick) Woychik, Director of the National Institute of Environmental Health Sciences (NIEHS) and the National Toxicology Program (NTP) gave a presentation on NIEHS's activities and potential collaborations between NIEHS and NHGRI. NIEHS is studying the impact of the environment on human health and is interested in a broad assortment of environmental exposures. Dr. Woychik provided information on successful collaborations between NIEHS and other institutes, including NHGRI, such as the Roadmap Epigenomics Program, the International Common Disease Alliance (ICDA), and the Genes, Environment, and Health Initiative. Dr. Woychik discussed the importance of genetic, epigenetic, and environmental influences on common diseases, and gave an overview of NIEHS-funded Personalized Environment and Genes Study (PEGS). PEGS aims to develop a resource for a variety of studies dedicated to clarifying the interaction of the genetic and environmental determinants of human health. Nearly 20,000 participants have enrolled and contributed biological samples to the PEGS resource. Dr. Woychik also gave an update on the proposed NIH-wide Initiative on Climate Change and Health (CCH). The President's Budget for Fiscal Year 2022 included \$100M for NIEHS to fund CCH research. NIEHS is encouraging all NIH institutes and centers to participate in the NIH-wide CCH Working Group that will help develop the strategic plan and to contribute to the CCH initiative.

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

## **PRESENTATION** – *Workshop Report: Future Directions of the NHGRI Analysis, Visualization, and Informatics Lab-space (AnVIL)* – Dr. Marylyn Ritchie

Dr. Marylyn Ritchie provided a summary of the Future Directions of the NHGRI AnVIL Workshop, which was sponsored by NHGRI and held in October 2021. The goal of the workshop was to identify gaps, challenges, and future opportunities related to NHGRI's investments in the AnVIL. Details of this workshop are available [online](#), including links to a workshop report and recordings of all presentations. The workshop was attended by 105 individuals and focused on four main topics: data submission and consortia engagement, analysis tools, infrastructure, and outreach and training. Several cross-cutting themes were discussed, including: (1) how cloud-based platforms can better serve the needs of genomics researchers; (2) what tools and services would better support clinical genomics researchers; and (3) how to improve interoperability with other cloud-based resources. Dr. Ritchie provided an overview of the strengths, weaknesses, opportunities, and threats (SWOT) analysis that was conducted by workshop participants.

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

## **CONCEPT CLEARANCE** – *Multi-Omics for Health and Disease (RFA)* – Dr. Joannella Morales

Dr. Joannella Morales presented the Multi-Omics for Health and Disease concept, which was developed following the [Multi-Omics of Disease Workshop](#) held in June 2021. Recent studies have shown that integrative ("multi-omics") analysis approaches can address biological

questions at a deeper resolution, improve the classification of disease into clinically relevant subgroups, and potentially identify biomarkers of health and disease. The goal of the proposed concept is to validate and enhance multi-omics approaches to identify biological changes related to disease, detect and assess molecular profiles, develop best practices and standards for data use and analysis, and create a multidimensional dataset and portal. Three funding opportunity announcements were proposed that would support the following: (1) Data Analysis and Coordination Center; (2) Disease Study Sites; and (3) Data Production Centers. Each Disease Study Site would focus on a disease area with evidence that integrative multi-omics would be impactful; recruit or re-consent 200-300 participants, 75% of which must be from underrepresented ancestral backgrounds; and collect specimens at a minimum of three timepoints. Council expressed enthusiasm for the concept overall, but also noted the importance and necessity of clarity and linkage between sample collection and data production due to the heterogenous disease types, samples, and techniques.

Council members approved the concept unanimously. A video of Dr. Morales's presentation and the related discussion can be found here: [\[link\]](#)

**PRESENTATION** – *Triennial Inclusion Report on Human Subjects Participation in NHGRI-Sponsored Research* – Christine Chang

Christine Chang gave the NHGRI Triennial Inclusion Report. All Institutes and Centers at NIH are mandated to ensure the inclusion of women and minority groups as participants in research projects as appropriate to the scientific question(s) under study. Data from NHGRI-sponsored projects in 2019, 2020, and 2021 were presented, along with comparisons to data from earlier reporting periods. While there was a substantial decrease in enrollment compared to the previous reporting period due to the ongoing COVID-19 pandemic, there were successful strategies employed to increase overall minority participant enrollment, but improvements are still needed. Council voted unanimously to accept the report.

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

**COUNCIL-INITIATED DISCUSSION**

Council members mentioned the National Academies of Sciences, Engineering, and Medicine (NAEM) work on the use of race, ethnicity, and ancestry as population descriptors in genomics research, and requested information about whether there is ongoing work to update the racial and ethnic categories used for grant applications to better reflect our contemporary understanding of race and ethnicity.

Council members noted the increase in the number of large complex datasets being generated brings a need for new methods for integrating and analyzing such data. Much of the discussion focused on how best to create and disseminate tools and information for data analysis, and Council suggested several future activities for NHGRI to consider such as workshops, travelling roadshows, presentations at national meetings, and sponsoring curriculum development work. NHGRI is currently developing two funding opportunity announcements related to cloud-based genomic data science (CGDS) education, which propose to use NIH-funded cloud platforms as a tool to enhance CGDS education at institutions serving groups underrepresented in the biomedical and data science fields. Additionally, NHGRI supports outreach activities through the AnVIL and other large programs. Council also acknowledged that future resources developed will have to be updated and there is a cost associated with this. The NHGRI Genomic Data

Science Working Group was suggested as a possible group to conduct further discussion on this topic.

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

## **CONFLICT OF INTEREST STATEMENT <sup>1</sup>**

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 248 applications, requesting \$180,266,787 (direct costs). The applications included: 162 research project applications (R01, R03, R21, or RM1); 14 Cooperative Agreement Research Center applications (U54); 2 Conference Applications (R13); 12 Education applications (R25); 16 Community Resource applications (U24); 6 Career Development applications (F99/K00, K99/R00, K01, or K08); 7 Institutional Training Grant applications (T32); 16 SBIR Phase I applications (R43); 10 SBIR Phase II applications (R44); 1 STTR Phase 1 application (R41); and 2 STTR Phase 2 applications.

### **NHGRI STAFF PRESENT**

Katie Bardsley, ERP  
Sarah Bates, CPLB  
Vence Bonham, IOD  
Joy Boyer, ERP  
Lawrence Brody, ERP  
Comfort Browne, ERP  
Dee Burkardt, ERP  
Alicia Caffi, DM  
Stephanie Calluori, ERP  
Lisa Chadwick, ERP  
Christine Chang, ERP  
Monika Christman, ERP  
Heather Colley, ERP  
Amanda Conti, PPAB  
Jyoti Dayal, ERP  
Valentina Di Francesco, ERP  
Argenia Doss, ERP  
Alvaro Encinas, CPLB  
Elise Feingold, ERP  
Adam Felsenfeld, ERP  
Ann Fitzpatrick, DM  
Colin Fletcher, ERP  
Anneliese Galczynski, ERP  
Prabarna Ganguly, CPLB  
Sean Garin, ERP  
Tina Gatlin, ERP  
Elena Ghanaim, PPAB  
Daniel Gilchrist, ERP  
Jenell Glover, ERP

Madison Goldrich, ERP  
Bettie Graham, ERP  
Eric Green, IOD  
Chris Gunter, IOD  
Linda Hall, ERP  
Zephaun Harvey, ERP  
Lucia Hindorff, ERP  
Carolyn Hutter, ERP  
Deanna Ingersoll, ERP  
Amber Jackson, ERP  
Cristina Kapustij, PPAB  
Dave Kaufman, ERP  
Jim Kees, ERP  
Se Rin (Julie) Kim, ERP  
Michael Lacy, DM  
Grace Lamoure, ERP  
Rongling Li, ERP  
Asiyah Lin, ERP  
Natalie Linear, ERP  
Nicole Lockhart, ERP  
Ebony Madden, ERP  
Teri Manolio, ERP  
Radu Marin, ITB  
William Maye, ITB  
Allison McCague, PPAB  
Keith McKenney, ERP  
Donna Messersmith, ECIB  
Jenn Montooth, CPLB  
Joannella Morales, ERP

Donna Morris, ERP  
Stephanie Morris, ERP  
Mukul Nerurkar, CPLB  
Allie Osgood, ERP  
Nguyen Park, ERP  
Mike Pazin, ERP  
Haleena Phillips, ERP  
Ajay Pillai, ERP  
Mindy Perilla, IRP  
Rudy Pozzatti, ERP  
Erin Ramos, ERP  
Kathleen Renna, ERP  
Renee Rider, ERP  
Enitza Rodriguez, ERP  
Ellen Rolfes, DM  
Kristen Ross, ERP  
Robb Rowley, ERP  
Ismail Safi, ERP  
Ella Samer, ERP  
Lorjetta Schools, ERP  
Baergen Schultz, ERP  
Jamil Scott, IOD

Shurjo Sen, ERP  
Michael Smith, ERP  
Grace Snyder, ERP  
Heidi Sofia, ERP  
Jerryl Somani, ITB  
Rene Sterling, ERP  
Ana Stevens, ERP  
Jennifer Strasburger, ERP  
Michelle Tallman, ERP  
Jill Thomas, CPLB  
Barbara Thomas, ERP  
Helen Thompson, ERP  
Jennifer Troyer, ERP  
Susan Vasquez, IOD  
Simona Volpi, ERP  
Harry Wedel, CPLB  
Chris Wellington, ERP  
Kris Wetterstrand, IOD  
Ken Wiley, ERP  
Sarah Wheelan, ERP  
Valerie Willis, ERP

ERP = Extramural Research Program  
CPLB = Communications and Public Liaison Branch  
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  
IRP = Intramural Research Program

*This NACHGR Meeting Summary was prepared by Baergen Schultz, NHGRI Scientific Program Analyst.*

05/16/2022

Date

*Rudy Pozzatti*

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

05/18/2022

Date

*Eric D. Green*

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

This report was approved by the NACHGR on May 16, 2022.