NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH MEETING SUMMARY

May 16-17, 2022

The Open Session of the 96th meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened virtually by Zoom at 11:30 a.m. on Monday, May 16, 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 May 16, 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 May 16, 2022, and from 11:00 a.m. until adjournment on May 17, 2022, for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT

Laura Bierut (Washington University School of Medicine)
Kyle Brothers (University of Louisville)
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)
Len Pennacchio (University of California, Berkeley)
Stephen Rich (University of Virginia)
Olga Troyanskaya (Princeton University)

AD HOC MEMBERS

Hagan Bayley (University of Oxford)
Joe Beery (LunaDNA)
Peter Robinson (The Jackson Laboratory for Genomic Medicine)
Jay Shendure (University of Washington)

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 viewed here: [link]

APPROVAL OF MEETING SUMMARY

The Council approved the February 7, 2022 Meeting Summary by a unanimous vote.

FUTURE NACHGR MEETING DATES

- September 19-20, 2022
- February 13-14, 2023
- May 15-16, 2023
- September 18-19, 2023
- February 12-13, 2024
- May 20-21, 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 <u>video</u> of his presentation and the <u>slides</u> are available here: [link]

PRESENTATION – The Future of Nursing Research: Innovation, Collaboration, and Impact – Dr. Shannon Zenk

Dr. Shannon Zenk, Director of the National Institute of Nursing Research (NINR) presented a talk on the future of nursing research and the activities of NINR with the release of their new strategic plan. Dr. Zenk started with an overview of NINR's history and the importance of nursing research, with a special focus on health inequalities. NINR released its 2022-2026 strategic plan 10 days ago, addressing challenges and goals through the "five research lenses," giving different perspectives to examine health challenges in the field and allowing for a broad scope for NINR research. The full strategic plan can be found on the NINR webpage (ninr.nih.gov). NINR is collaborating with other NIH institutes/centers on projects such as the Climate Change and Health Initiative, the Social Determinants of Health Research Committee, and the Community Partnerships to Advance Science for Society (ComPASS) program. Dr. Zenk also touched on opportunities and best practices for collaboration between NINR and NHGRI.

A video of Dr. Zenk's presentation and the related discussion can be found here: [link]

PRESENTATION - Chief Officer for Scientific Workforce Diversity Update - Dr. Marie Bernard

Dr. Marie Bernard, the NIH Chief Officer for Scientific Workforce Diversity, presented an update on the NIH Diversity, Equity, Inclusion, and Accessibility (DEIA) strategic plan and related activities. The strategic plan is currently being drafted as a blueprint for short-term and long-term goals that address racial, ethnic, and gender disparities at the NIH. The plan will be finalized in late spring or early summer to convey priorities for the next five years. The strategic plan aligns well with the diversity action agenda put forth by NHGRI in 2021. Dr. Bernard also gave an update on the new NIH UNITE initiative and its activities, including two of the FOAs to address health disparities and the ComPASS program. Internal workforce initiatives include the Power of an Inclusive Workforce recognition project to represent the range of individuals working in the biomedical sciences. Externally, the Faculty Institutional Recruitment for Sustainable Transformation (FIRST) program was set up to encourage institutional cultural changes around the US.

A video of Dr. Bernard's presentation and the related discussion can be found here: [link]

CONCEPT CLEARANCE – Analysis, Visualization, and Informatics Lap-Space (AnVIL) [RFA Renewal] – Mr. Chris Wellington and Dr. Ken Wiley

Mr. Chris Wellington and Dr. Ken Wiley presented the renewal plans of the Analysis, Visualization, and Informatics Lap-Space (AnVIL) program. The presentation started with an overview and timeline of the AnVIL program and its current status, such as the 4 petabytes of data across 20 research networks that has been uploaded to date. There was an explanation of the AnVIL lab space structure with a focus on outreach and user engagement. The future goals will be met by the two arms of the program, with the first RFA increasing tool availability, improving interoperability, and addressing barriers to cloud computing through a limited competition funding opportunity announcement. The second will be an open competition RFA focused on clinical resources to serve the community by leveraging the existing AnVIL efforts, titled the AnVIL Clinical Resource (ACR). The budget is approximately \$8M total cost per year over the next 5 years.

Council members approved both concepts unanimously. A video of Mr. Wellington's and Dr. Wiley's presentations and the related discussion can be found here: [link]

CONCEPT CLEARANCE – Diversity Genome Research Centers [RFA] – Dr. Ebony Madden

Dr. Ebony Madden presented two Diversity Genomes Research Centers RFA concepts. The concepts grew from the diversity and inclusion research informing the 2020 NHGRI Strategic Vision and the subsequent Action Agenda. The diversity genome research centers would help increase diversity in the genomics research workforce by supporting research activities and infrastructure enhancement at minority serving institutions (MSI). Two Request for Applications (RFAs) are proposed. One for U54 full-scale genome research centers, and a second that features a two-stage project using the UG3/UH3 activity code to develop infrastructure and genomics research capacity on the path to a U54 center. There will be opportunities for collaborations with well-resourced institutions, but the majority of the funds must go to the MSIs. A total cost of \$42.4M is proposed for both RFAs that will support 4 full-scale centers for 5 years and UG3/UH3 projects for up to 7 years.

Council members approved both concepts unanimously. A video of Dr. Madden's presentation and the related discussion can be found here: [link]

WORKSHOP REPORT - Future Directions in Genomics and Health Equity - Dr. Judy Cho

Dr. Judy Cho is the Dean of Translational Genetics and Director of the Charles Bronfman Institute for Personalized Medicine at the Icahn School of Medicine at Mount Sinai. She presented a report on the Future Directions in Genomics and Health Equity Research workshop that took place on April 6-7, 2022. The workshop featured keynote presentations, panel discussions, and breakout groups over the two days. Selected discussion points were highlighted, focusing on health disparities in genomics, identifying research gaps and opportunities, and addressing structural factors to support health equity in genomics research. Opportunities that NHGRI could pursue were also presented, including diversifying the genomics workforce and addressing the challenge of collecting genomic data from diverse populations. Priorities were developed from the breakout room discussion and input from workshop participants. There is a manuscript in preparation for a peer-reviewed publication. Information and video recordings from the workshop can be found here.

A video of Dr. Cho's presentation and the related discussion can be found here: [link]

COUNCIL-INITIATED DISCUSSION

Council members requested a presentation on big-data efforts at the NIH that could be leveraged to support work at NHGRI. Dr. Susan Gregurick from ODSS was suggested.

In addition, Dr. Lindsay Criswell from NIAMS was suggested as a possible speaker since she is a relatively new institute director.

Council members also requested a future presentation on ARPA-H to hear about collaborative opportunities. Once an ARPA-H Director is announced, that individual should be invited to give a presentation to the Council.

Another suggestion was given for a presentation and discussion about the biomedical ethics issues that are specific to research projects involving big data and genomics.

A final suggestion was a request for a presentation on data privacy and regulations at the NIH in relation to the global landscape. Council members volunteered to provide names for potential speakers.

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 182 applications, requesting \$82,202,713 (direct costs). The applications included: 119 research project applications (R01, R03, or R21); 25 Cooperative Agreement applications (U01, UM1, or U54); 1 Conference Application (R13); 3 Education applications (R25); 10 Community Resource applications (U24); 13 Career Development applications (F99/K00 or K99/R00); 8 SBIR Phase I application (R43); 2 SBIR Phase II applications (R44); and 1 STTR Phase 1 application.

NHGRI STAFF PRESENT

Alexander Arguello, ERP Katie Bardsley, ERP Sarah Bates, OC 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, OGDS

Argenia Doss, ERP Alvaro Encinas, OC Elise Feingold, ERP Adam Felsenfeld, ERP Ann Fitzpatrick, DM Colin Fletcher, ERP

Anneliese Galczynski, ERP Prabarna Ganguly, OC

Sean Garin, ERP Elena Ghanaim, OGDS Daniel Gilchrist, ERP Jenell Glover, OGDS Madison Goldrich, ERP Bettie Graham, ERP Eric Green, IOD Chris Gunter, IOD Linda Hall, ERP

Zephaun Harvey, ERP Lucia Hindorff, TiDHE Carolyn Hutter, ERP Deanna Ingersoll, ERP Amber Jackson, TiDHE 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, OGDS
Natalie Linear, ERP
Nicole Lockhart, ERP
Ebony Madden, TiDHE
Teri Manolio, ERP
Radu Marin, ITB
William Maye, ITB
Allison McCague, PPAB

Allison McCague, PPAB Keith McKenney, ERP Donna Messersmith, ECIB

Jenn Montooth, OC Joannella Morales, ERP Donna Morris, ERP Stephanie Morris, ERP Mukul Nerurkar, OC Allie Osgood, ERP Nauven 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, TiDHE Baergen Schultz, ERP Jamil Scott, TiDHE Shurjo Sen, OGDS

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

ERP = Extramural Research Program

OC = Office of Communications

Michael Smith, ERP

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 Sean Garin, NHGRI Scientific Program Analyst.

Date Rudy Pozzatti, Ph.D. Executive Secretary

National Advisory Council for Human Genome Research

Date Eric Green, M.D, Ph.D.

Chairman

National Advisory Council for Human Genome Research

This report was approved by the NACHGR on September 19, 2022