

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
February 12-13, 2024

The Open Session of the 101st meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened at 10:30 a.m. on February 12, 2024, with 13 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 February 12, 2024. 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 February 12, 2024, and from 11:00 a.m. until adjournment on February 13, 2024, 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)
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 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 September 18-19, 2023, Meeting Summary by a unanimous vote.

FUTURE NACHGR MEETING DATES

- May 20-21, 2024
- September 9-10, 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, [slides](#), and related documents are available here: [\[link\]](#)

PRESENTATION – *Research at the Intersection of Translational Science and Genomics* – Dr. Joni Rutter, Director National Center for Advancing Translational Science (NCATS).

Dr. Rutter noted that the NCATS research mission is focused on the many thousands of diseases that don't have effective therapeutic interventions. The NCATS mission statement is, "Turn Research Observations into Health Solutions through Translational Science." NCATS has three long-term goals. (1) Achieve a five-fold increase in the number of disease treatments; (2) Increase inclusivity across all groups of people in the areas that NCATS supports; and (3) Reduce by 50% the length of time needed to develop new diagnostics and therapeutics. Dr. Rutter described some of NCATS' collaborative work with NASA scientists and the Rare Disease Clinical Network (which involves 20 extramural groups working together to develop diagnoses faster in order to shorten the diagnostic odyssey experienced by patients with a rare disease). Another research priority for NCATS is their support of gene therapy technology. Dr. Rutter described research activity for somatic cell gene editing work, research efforts being conducted by the Bespoke Gene Therapy Consortium, and the Platform Vector Gene Therapy project focused on gene delivery mechanisms using AAV vectors. They are currently planning to conduct clinical trials on three ocular diseases, three neurology diseases and two systemic diseases, all using AAV vectors for gene delivery. Dr. Rutter concluded her presentation describing the work of the Clinical Translational Science Awards Program that involves more than 60 academic medical institutions throughout the United States.

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

CONCEPT CLEARANCE – *Building Partnerships and Broadening Perspectives to Advance ELSI Research* – Dr. Rene Sterling

Dr. Sterling presented the concept for *Building Partnerships and Broadening Perspectives to Advance ELSI Research*. This Request for Applications (RFA) seeks to advance Ethical, Legal, and Social Implications (ELSI) research by broadening the types of knowledge, skills, experience, expertise, and perspectives brought to bear in the field through transdisciplinary ELSI research led by research teams with community representatives. As Dr. Sterling emphasized, expanding involvement in ELSI research can lead to greater innovation and creativity and help illuminate a path towards greater equity in genomics. Eligible applicants would include domestic organizations receiving less than \$30M in total funding from NIH for the past three fiscal years. Council members were supportive of the proposed program as it would offer valuable ways of strengthening and broadening the ELSI workforce. There was discussion surrounding how collaborators would be chosen and whether awards would be staggered by cycle.

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

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

CONCEPT CLEARANCE – *NHGRI Predoctoral to Postdoctoral Transition Award for Ethical, Legal, and Social Implications (ELSI) Research (F99/K00)* – Dr. Rene Sterling

Dr. Sterling presented the concept for *NHGRI Predoctoral to Postdoctoral Transition Award for Ethical, Legal, and Social Implications (ELSI) Research (F99/K00)*. Based on a review of the ELSI research program training portfolio, fellowship opportunities for graduate fellows have been limited in the ELSI research program. This notice of funding opportunity (NOFO) would seek to address this gap by providing funding for predoctoral to postdoctoral transition for ELSI research scholars. Council members were supportive and enthusiastic. There was discussion

around how student loans would be affected by the grants and whether the program would support someone moving from one discipline to another.

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

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

CONCEPT CLEARANCE – *Population Screening in Primary Care* – Dr. Simona Volpi

Dr. Volpi presented the concept for *Population Screening in Primary Care*. The concept proposes three Requests for Applications (RFAs) for pilot programs for population screening for common, actionable genomic conditions predominantly in the primary care setting. The three RFAs would focus on population screening for three genetic conditions classed as Tier 1 by the CDC – Hereditary Breast and Ovarian Cancer syndrome (HBOC), Lynch Syndrome, and Familial Hypercholesterolemia (FH). As Dr. Volpi emphasized, there would be a focus on community engagement and implementation science. The program would consist of a coordinating center, a sequencing center, and 4-5 clinical groups teamed with primary care physicians working in diverse settings and populations. The program would operate for five years. Council members were supportive as it sounds like a natural follow-up to the Genomic Medicine XV workshop while also incorporating innovative topics. There was discussion about the size of the budget, how the budget would be allocated, and program sustainability. There was a recommendation to expand to support implementation of population screening for additional conditions.

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

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

CONCEPT CLEARANCE – *RFA Renewal: ELSI Congress Research Conference* – Dr. Nicole Lockhart

Dr. Lockhart presented the concept for *ELSI Congress Research Conference*. This concept seeks to renew a Request for Applications (RFA) which funds a biannual meeting for the ELSI research community known as the ELSI Congress. The congress is a hybrid, multi-day meeting held every two years that routinely attracts around 300-400 attendees. The sessions include presentations of invited and peer-reviewed work. There are also dedicated sessions for trainees and early career scholars. Dr. Lockhart pointed out that updates to the concept in this renewal emphasize maximum accessibility for people with disabilities. This RFA would support three meetings beginning in Fiscal Year 2025. Council members were supportive overall given the meeting's success so far. There was discussion around expanding the meeting to include people who are typically overlooked as attendees, to provide sponsorships, and to give travel funds for attendees.

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

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

CONCEPT CLEARANCE – *Enhancing Reuse of NHGRI Data Assets (ERND)* – Dr. Shurjo Sen

Dr. Sen presented the concept for *Enhancing Reuse of NHGRI Data Assets (ERND)*. This request for applications (RFA) seeks R03 (Small Grant Program) applications for research

performing secondary analysis on existing datasets on NHGRI's secure cloud platform for genomics – the Analysis, Visualization, and Informatics Lab-Space (AnVIL). There are now over 600,000 genome sequences stored in AnVIL. At an AnVIL community workshop there was a unanimous recommendation that NHGRI should create ways to engage the broader genomics community and provide support and training for them to start using AnVIL. This award would fund 6-8 Small Grant Program applications per cycle for two years each. Council members were overall supportive as it seems critical to the field to begin this process. There was discussion around providing resources for applicants to browse existing data ahead of time, the waiting process to gain access to AnVIL, and creating large scale summary statistic results of the AnVIL data.

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

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

CONCEPT CLEARANCE – *PAR Renewal: Investigator-Initiated Research in Computational Genomics and Data Science* – Dr. Dan Gilchrist

Dr. Gilchrist presented the concept for *Investigator-Initiated Research in Computational Genomics and Data Science*. This concept seeks to renew a Program Announcement with special review criteria or receipt dates (PAR) for notices of funding opportunities (NOFOs) in investigator-initiated research in computational genomics and data science. The award was first published in 2018, renewed in 2021, and is now set to expire in 2024. Dr. Gilchrist emphasized that funding opportunities received a strong response from investigators and continue to do so and provide a significant source of funding and investment in innovative research in computational genomics and data science. The existing funding opportunities support R01 and R21 applications and this renewal proposes to add the R15 activity code. Council members were very supportive of the proposal and consider it a critical program and funding source. There was discussion around exploring issuing small parallel small business opportunities.

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

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

PRESENTATION – *Evaluation of the Centers of Excellence in Genomic Science (CEGS) Program* – Dr. Chris Gunter

Dr. Chris Gunter, Senior Advisor to the NHGRI Director on Genomics Engagement, provided an evaluation of the Centers of Excellence in Genomic Science (CEGS) Program. Dr. Gunter explained that NHGRI is committed to transparency and program assessment and intends to conduct mixed methods program evaluations and make the results available through publication. CEGS is acting as the flagship grant mechanism for this evaluation process. CEGS is unique in that it is a “high-risk, high-reward” program that focuses on transformative approaches and outputs. In order to carry out the program evaluation, surveys were constructed, and semi-structured interviews were held with CEGS Principal Investigators (PIs). Dr. Gunter noted some recency bias in the results of the survey. The results of the survey found that overall, CEGS programs produced more publications and more highly cited papers than comparison R01s but produced fewer papers per \$1M of investment. It was suggested this is likely due to infrastructure requirements. The survey also yielded a list of breakthrough techniques, resources, and approaches that the CEGS research projects contributed to, including RNA-seq and single cell ATAC-seq. Participants also reported that participation in

CEGS allowed them to engage in education and outreach activities that would not have existed otherwise, and it was suggested that CEGS work to provide even more training opportunities. Another suggestion for improvement of the CEGS program was further outreach to increase diversity of participants. Council members acknowledged the impact of the CEGS program and were enthusiastic about the data in the report, but suggested caution in bias from the low response rate as well as citation bias.

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

PRESENTATION – *Advancing Genomic Medicine through Genome Informed Risk Assessment – eMERGE Network Update* – Dr. Eimear Kenny

Dr. Kenny provided an update about the Electronic Medical Records and Genomics (eMERGE) Network. The presentation focused on the Genome Informed Risk Assessment (GIRA), a method to bring Polygenic Risk Scores (PRS) into clinical care. GIRA combines monogenic risk, clinical risk, polygenic risk, and familial risk to develop a report for a patient on 11 specified conditions. Dr. Kenny noted that one of these conditions is not polygenic and that, at the moment, pediatric patients only receive a report on four specified conditions. The risk report is integrated into a prospective study, in which a study staff returns the high-risk report and the report is integrated into the Electronic Health Record (EHR). Nearly one third of the GIRA that have been returned so far have been high-risk results. In the future, the eMERGE network is primed to tackle more sophisticated risk stratification schema (such as using AI or ML), incorporate more datatypes, and utilize more sophisticated outcome assessment tools. Council members were interested in the results coming from the GIRA studies, asked about lifestyle interventions for participants, and recommended thinking about scalability of the program.

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

REPORT – *Genomic Medicine Working Group of Council Annual Report* – Dr. Teri Manolio

Dr. Manolio, the Director of the Division of Genomic Medicine, presented the annual report on the Genomic Medicine Working Group (GMWG) of Council. Dr. Manolio provided a year in review of the papers highlighted by the working group as "Accomplishments in Genomic Medicine," and invited nominations for future accomplishments from the group. The Year in Review was published in the *American Journal of Human Genetics*, as it has been since 2019. Dr. Manolio provided a report on the most recent Genomic Medicine Meeting (GM XV) and presented on the group's budget. Council members noted their support for the program.

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

REPORT- – *Genomic Data Science Working Group of Council Annual Report* – Dr. Shannon McWeeney and Dr. Anshul Kundaje

Working Group Chairs Dr. McWeeney and Dr. Kundaje presented the annual report on the Genomic Data Science Working Group (GDSWG). The group currently has 12 members as of February 2024. Its main roles are to advise NHGRI on plans related to genomic data science and provide input to the Director and other institute leaders about trans-NIH issues related to data science. In 2023, the group discussed topics such as new initiatives about ML/AI tools, NHGRI training in genomics and data science, and funding early-stage investigators. Dr. McWeeney provided a Portfolio Review of the Genomic Community Resource PARs. Upcoming Working Group discussion topics will include phenotypic data collection and sharing across

NHGRI programs, genomics research transition to the cloud, unification of knowledgebases, and others. Council members asked questions about the Global Core Biodata Resource.

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

COUNCIL-INITIATED DISCUSSION

There was discussion about a potential year-long Continuing Resolution (CR). NHGRI has modeled 5%, 10%, and 15% cuts to the institute's budget. The group discussed how budget cuts would affect pay lines, and Dr. Green agreed to follow up with a full projection at the May Council meeting. There was additional discussion about the unionization of NIH intramural postdoctoral fellows as well as other possible presentations for future Council meetings, including a presentation on NHGRI keeping pace with NIH for early-stage investigators. Dr. Green informed the group that Dr. Bertagnolli will speak at the September Council meeting.

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

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 202 applications, requesting a total of \$89,279,541 (direct costs). The applications included: 124 research project applications (R01, R03, R15, and R21); 3 phase 1 exploratory development applications (UG3); 9 research project with complex structure (UM1 and RM1) applications; 2 specialized center applications (U54); 2 conference applications (R13); 21 education applications (R25); 2 community resource applications (U24 and U24); 10 Career Development applications (F99/K00, K99/R00, K01, and K08); 3 institutional training grant applications (T32); 9 SBIR Phase I applications (R43); 11 SBIR Phase 2 applications (R44); 4 STTR phase one applications (R41); 1 STTR phase II application (R42) and 1 commercialization readiness (SB1) application.

NHGRI STAFF PRESENT

Adam Felsenfeld, ERP
Afia Asare, ERP
Ajay Pillai, ERP
Alanna Kulchak Rahm, ERP
Alessandra Serrano Marroquin, ERP
Alexander Arguello, ERP
Alicia Caffi, DM
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

Brandon Meiklejohn, ITB
Britny Kish, OC
Candace Holt, ERP
Caprina Pipion, ERP
Carolyn Hutter, ERP
Chris Gunter, IOD
Chris Wellington, OGDS
Christina Daulton, TIDHE
Christine Chang, ERP
Christopher Donohue, OC
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
Imani Boykin, ERP
Ismail Safi, ERP
Jacob Jaffa, 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
Kayla Titalli-Torres, PPAB
Keith McKenney, ERP
Kenya Smith, OC
Kimberly Ferguson, ERP
Kris Wetterstrand, ERP
Lawrence Brody, ERP
Lisa Chadwick, ERP
Liz Dietz, OC
Lori Bonnycastle, ERP
Lucia Hindorff, TiDHE
Madji Lodoungoto, OGDS
Marcia Morris, ERP
Maricela Trujillo, ERP

Mauresa Pittman, OC
Maya VanZanten, ERP
Melinda Rose, ERP
Michelle Tallman, ERP
Mike Pazin, ERP
Molly Bird, PPAB
Monika Christman, ERP
Mukul Nerurkar, OC
Natalie Linear, ERP
Nephi Walton, 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 Zo Bly, NHGRI Scientific Program Analyst.

05/21/2024

Date

Rudy Pozzatti, Ph.D.

Rudy Pozzatti, Ph.D.

Executive Secretary

National Advisory Council for Human Genome Research

05/23/2024

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 May 20, 2024