

**NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH
MEETING SUMMARY
February 22-23, 2021**

The Open Session of the 92nd meeting of the National Advisory Council for Human Genome Research (NACHGR) was convened virtually at 11:30 AM Eastern Time (ET) on Monday, February 22, 2021. 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 AM – 6:00 PM on February 22, 2021. In accordance with the provisions of Public Law 92-463, the meeting was closed to the public from 11:00 AM until adjournment on February 23 for the review, discussion, and evaluation of grant applications.

COUNCIL MEMBERS PRESENT:

Jeffrey Botkin (University of Utah)
Howard Chang (Stanford University)
Wendy Chung (Columbia University)
Mark Craven (University of Wisconsin)
Patricia Deverka (Deverka Consulting, LLC)
Harry Dietz (Johns Hopkins University)
Stephen Fodor (13.8 Inc.)
Jonathan Haines (Case Western Reserve University)
Trey Ideker (University of California, San Diego)
Rafael Irizarry (Dana-Farber Cancer Institute)
Lisa Parker (University of Pittsburgh)
Len Pennacchio (University of California, Berkeley)
Sharon Plon (Baylor College of Medicine)
Stephen Rich (University of Virginia)

AD HOC ADVISORS PRESENT:

Olga Troyanskaya (Princeton University)

AD HOC DISCUSSANTS PRESENT:

Michael Boehnke (University of Michigan)
John Carpten (University of Southern California)
Howard Jacob (AbbieVie, Inc.)
Owen White (University of Maryland School of Medicine)

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 at: [\[link\]](#)

APPROVAL OF MINUTES

The Council approved the September 2020 Meeting minutes by a unanimous vote.

FUTURE NACHGR MEETING DATES:

- May 17-18, 2021
- September 13-14, 2021

- February 7-8, 2022
- May 16-17, 2022
- September 19-20, 2022

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 at: [\[link\]](#)

PRESENTATION – Final NIH Policy on Data Management and Sharing – Dr. Carrie Wolinetz

Dr. Carrie Wolinetz, Acting Chief of Staff and Associate Director of the NIH Office of Science Policy presented on the final NIH Policy for Data Management and Sharing. The purpose of this policy is to advance rigorous and reproducible research and to promote public trust in research. The policy applies to all NIH-supported research that generates scientific data. Applications will require a data management and sharing plan as part of the Budget Justification section. The Office of Science Policy is in the process of doing outreach to ensure the community is aware of the new policy. Tools for good data-sharing practices, guidance for extramural program staff, and other requested resources are being developed. NIH welcomes feedback from the research community about other materials that would be useful as the policy is implemented. The policy will be in effect beginning with applications submitted in January 2023.

A video of Dr. Wolinetz's presentation and the Council discussion can be found at: [\[link\]](#)

PRESENTATION – Building a Diverse Genomics Workforce: An NHGRI Action Agenda – Vence Bonham, JD

Vence Bonham, Senior Advisor to the NHGRI Director on Genomics and Health Disparities, gave a presentation on the newly released NHGRI Action Agenda for Building a Diverse Genomics Workforce. One of the major themes highlighted in the 2020 NHGRI Strategic Vision is the need for enhancing diversity of the genomics workforce. Major goals in the new Action Agenda strive to support exposure to genomics and the creation of networks for trainees from K-12 and undergraduates through independent genomic and clinical research careers. An additional goal is an ongoing evaluation of the Action Agenda to assess its effectiveness and determine if specified targets are being reached. Mr. Bonham also highlighted new partnerships with the American Society of Human Genetics, the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors including use of a survey to better understand the current diversity of the genomics workforce. NHGRI plans to work with other societies, industry, and academic institutions as well.

A video of Mr. Bonham's presentation and the related discussion can be found at: [\[link\]](#)

CONCEPT CLEARANCE – Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Promote Diversity (GREAT Program), RFA – Dr. Tina Gatlin

Dr. Tina Gatlin presented the Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Promote Diversity (or GREAT Program) RFA concept. The concept is part of a larger group of concepts being developed to enhance the diversity of the genomic workforce. The goal of the GREAT Program is to encourage undergraduates enrolled at diversity-serving institutions to train and pursue future careers in

genomics by providing research opportunities and educational experiences. The program will be a partnership between a diversity serving institution and a research-intensive institution with graduate-level genomics training. The institutions will be expected to create collaborative approaches for genomics education. Students will be part of a two-year program with opportunities to participate in research as well as other educational and professional development activities at both partnering institutions.

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

CONCEPT CLEARANCE – Grants for New Investigators to Promote Diversity in Genomics Research, RFA – Ms. Jyoti Dayal

Ms. Jyoti Dayal presented the Grants for New Investigators to Promote Diversity in Genomics Research RFA concept. The concept is part of a larger group of concepts being developed to enhance the diversity of the genomics workforce. The goal of this concept is to increase investigators in genomics from diverse backgrounds including underrepresented groups as defined by the Notice of NIH's Interest in Diversity Statement (NOT-OD-20-031). It is modeled on a National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) diversity R21 Funding Opportunity Announcement, which NHGRI currently co-sponsors. The new initiative aligns with the 2020 NHGRI Strategic Vision and the NHGRI Action Agenda for Building a Diverse Genomics Workforce. New and early stage investigators with interest in any research topic within NHGRI's mission would be eligible to apply.

NACHGR members approved the concept unanimously. A video of Ms. Dayal's presentation and the related discussion can be found at: [\[link\]](#)

CONCEPT CLEARANCE – Non-Human Primate dGTEX, RFA – Dr. Jennifer Troyer

Dr. Jennifer Troyer presented the Non-Human Primate dGTEX RFA concept. The goal of this concept is to examine gene-expression patterns in tissues across the developmental timeline in non-human primates. The project will establish data sets and samples for comparative developmental genomics and identify patterns that are human- or primate-specific. The project parallels other genotype-tissue expression projects such as the completed GTEX Common Fund project and the new developmental GTEX (dGTEX) project. The Non-Human Primate dGTEX project will generate comparable data sets to the previous projects in both prenatal and postnatal stages of development. Applicants are encouraged to use banked samples as much as possible and should leverage existing non-human primate resources in the National Primate Resource Center Consortium.

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

CONCEPT CLEARANCE – Investigator-initiated Research in Computational Genomics and Data Science, PAR – Dr. Daniel Gilchrist

Dr. Daniel Gilchrist presented plans to renew the Investigator-initiated Research in Computational Genomics and Data Science PAR concept. The purpose of the PAR renewal is to support research in computational genomics, data science, statistics for genomics, and bioinformatics. Supported work must be research-oriented, computational work that is relevant to basic or clinical genomics and broadly applicable to human health and disease. The renewal

will be updated to reflect the latest science and themes in the 2020 NHGRI Strategic Vision. Special review criteria supporting a broad definition of innovation and utility for the research community will also be added. Applicants from diverse backgrounds and communities underrepresented in biomedical sciences will be encouraged to apply.

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

CONCEPT CLEARANCE – Molecular Phenotypes of Null Alleles in Cells Pilot Project, RFA – Dr. Adam Felsenfeld

Dr. Adam Felsenfeld presented the Molecular Phenotypes of Null Alleles in Cells (MorPhic) Pilot Project RFA concept. This concept reflects one of the Compelling Genomics Research Projects in Biomedicine described in the 2020 NHGRI Strategic Vision. This pilot project includes a long-term goal of creating a catalog of molecular and cellular phenotypes of null alleles for all human genes. Such a catalog would provide basic information for all genes and fill in gaps between molecular and anatomical/physiological phenotypes. Phase one will be a four-year project to develop a pipeline, address barriers to scaling up, and troubleshoot challenges, all of which will serve to inform a potential second phase of the project. Phase one will focus on 1,000 protein-coding genes and test multiple approaches for mutagenesis and assays. There are three proposed components: (1) six data production centers, (2) 2-3 data analysis and validation centers and (3), a data resource center.

NACHGR members approved the concept with 10 votes for approval and 4 votes to oppose the concept. A video of Dr. Felsenfeld's presentation and the related discussion can be found at: [\[link\]](#)

PRESENTATION – Phenotypes and Exposure (PhenX) Toolkit – Dr. Erin Ramos

Dr. Erin Ramos presented an update on the Phenotypes and Exposure (PhenX) Toolkit. PhenX is an online catalog of recommended protocols for the research community to use with the goal of facilitating cross-study analysis and increasing the impact of research studies. Originally funded only by NHGRI, seven other NIH institutes now contribute funds to PhenX. The toolkit houses 863 data-collection protocols from 28 research domains such as demographics, various life stages, and specific disease areas. The most recent addition is a COVID-19 Research Collection. This two-part project captured information from the research community and then leveraged crowdsources to evaluate which COVID-19 protocols were of highest relevance. PhenX is also linked to a variety of other resources including, dbGaP and REDcap. To date, almost 400 NIH Funding Opportunity Announcements reference the PhenX Toolkit.

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

COUNCIL-INITIATED DISCUSSION

Council members suggested a number of potential presentation topics for future Council meetings: (1) information about returning results from polygenetic risk scores; (2) final reports from NHGRI programs that are sunsetting; (3) a discussion about data visualization research activities. Council also suggested hearing from Dr. Eric Lander or Dr. Alondra Nelson about the White House Office of Science and Technology Policy structure and priorities. Dr. Green noted that new NIH Institute Directors could also be invited to give presentations to the Council and discuss opportunities for collaborations with NHGRI.

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

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. Council members will absent themselves from the meeting when the Council discusses applications from their own institutions or when a conflict of interest may occur for other reasons.

REVIEW OF APPLICATIONS

In the Closed Session, the NACHGR reviewed 247 applications, requesting \$145,558,488 (direct costs). The applications included: 152 research project applications (R01, R03, R21, or RM1); 46 cooperative agreement applications (U01 or U24); 7 career development or career transition applications (K99/R00 or K01); 2 conference applications (R13); 3 education applications (R25); 6 institutional training applications (T32); 22 SBIR Phase I applications (R43); 7 SBIR Phase II applications (R44); 1 STTR Phase 1 application (R41), and 1 STTR Phase 2 application.

NHGRI STAFF PRESENT:

Sarah Bates, CPLB
Yasmeen Becket, IOD
Vence Bonham, IOD
Joy Boyer, ERP
Marie Brennan, ERP
Lawrence Brody, ERP
Lisa Brooks, ERP
Comfort Browne, ERP
Devon Bumbray-Quarles, ERP
Alicia Caffi, DM
Eileen Cahill, ERP
Lisa Chadwick, ERP
Christine Chang, ERP
Monika Christman, ERP
Heather Colley, ERP
Amanda Conti, PPAB
Luis Cubano, ERP
Christina Daulton, ECIB
Jyoti Dayal, ERP
Edith DeHaut, ERP
Valentina Di Francesco, ERP
Argenia Doss, ERP
Carla Easter, ECIB
Elise Feingold, ERP
Julia Fekecs, CPLB
Adam Felsenfeld, ERP
Kimberly Ferguson, ERP
Laurie Findley, ERP
Ann Fitzpatrick, DM
Colin Fletcher, ERP

Prabarna Ganguly, CPLB
Sean Garin, ERP
Sylvia Garvey, ERP
Tina Gatlin, ERP
Elena Ghanaim, PPAB
Daniel Gilchrist, ERP
Madison Goldrich, ERP
Bettie Graham, ERP
Chris Gunter, IOD
Linda Hall, ERP
Peggy Hall, ERP
Hwaida Hannoush, ERP
Lucia Hindorff, ERP
Carolyn Hutter, ERP
Deanna Ingersoll, ERP
Kimberly Jacoby Morris, ECIB
Alyssa Jones, CPLB
Cristina Kapustij, PPAB
Dave Kaufman, ERP
Jim Kees, ERP
Julie Kim, ERP
Natalie Kucher, ERP
Grace Lamoure, ERP
Rongling Li, ERP
Asiyah Lin, ERP
Natalie Linear, ERP
Nicole Lockhart, ERP
Ebony Madden, ERP
Teri Manolio, ERP
Allison McCague, PPAB

Donna Messersmith, ECIB
Joannella Morales, ERP
Donna Morris, ERP
Marcia Morris, ERP
Stephanie Morris, ERP
Ken Nakamura, ERP
Emily Neveux, DM
Briana Nuñez, ERP
Lisa Oken, ERP
Allie Osgood, ERP
Mike Pazin, ERP
Devona Perrineau, CPLB
Ajay Pillai, ERP
Natalie Pino, ERP
Erin Ramos, ERP
Alex Raphael, ERP
Kathleen Renna, ERP
Renee Rider, ERP
Enitza Rodriguez, ERP
Ellen Rolfes, DM
Robb Rowley, ERP
Lorjetta Schools, ERP

Baergen Schultz, ERP
Jamil Scott, OD
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
Barbara Thomas, ERP
Jill Thomas, CPLB
Jennifer Troyer, ERP
Susan Vasquez, IOD
Simona Volpi, ERP
Harry Wedel, CPLB
Chris Wellington, ERP
Kris Wetterstrand, IOD
Ken Wiley, ERP
Valerie Willis, ERP
Rosann Wise, ECIB

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
OD = Office of the Director
ITB = Information Technology Branch
PPAB = Policy and Program Analysis Branch

This NACHGR Meeting Summary document was prepared by Alex Raphael, NHGRI Scientific Program Analyst.

05/17/2021

Date

Rudy Pozzatti

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

05/20/2021

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 17, 2021

