

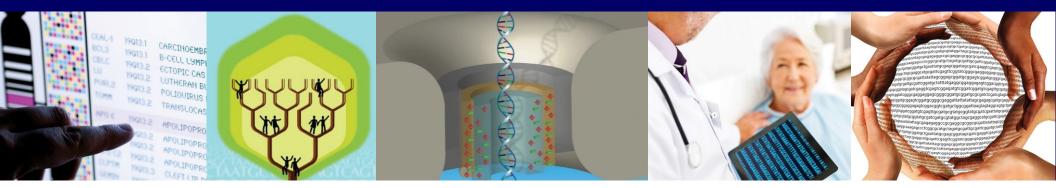
NIH National Human Genome Research Institute



DIRECTOR'S REPORT

Eric Green, M.D., Ph.D. Director, NHGRI

May 2022



NIH National Human Genome Research Institute	egin you	ur search here				Q		fy⊡
About Genomics Research Funding	Rese	arch at NHGRI	Health	Careers & Training	News & Events	About NH	GRI	
Director's Report-R	ela	ted D	ocur	nents				
May 2022		or's Report 🙏 or's Report 👔						
	No.	Relevant Doc	uments					
	1	NHGRI Celebr	ates Bettie	Graham's 50 Yea	rs of Federal Servic	9		
		NHGRI Office	of Genomi	c Data Science (O	GDS)			
		OGDS Website	e					
		OGDS News R	elease					
		PAR-20-100 - 0	Genomic Co	ommunity Resour	ces (U24)			
		Genomics Res Website	search to El	ucidate the Gene	tics of Rare Disease	s (GREGoR)	Progran	n
	2	NHGRI Genon Program Web		ence Analysis, Vi	sualization, and Info	ormatics La	ıb-space	(AnVIL)
		Bridge to Arti	ficial Intelli	igence (Bridge2AI) Program Website			

genome.gov/DirectorsReport

Document #

Open Session Presentations

Presentations:

The Future of Nursing Research: Innovation, Collaboration, and Impact Shannon Zenk

Chief Officer for Scientific Workforce Diversity Update Marie Bernard

Open Session Presentations

Concept Clearances:

Diversity Genome Research Centers Ebony Maddon

Analysis, Visualization, and Informatics Lab-Space (AnVIL) Chris Wellington and Ken Wiley

Workshop Report:

Future Directions in Genomics and Health Equity Judy Cho

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- **II. General NIH Updates**
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- V. NIH Common Fund/Trans-NIH
- VI. NHGRI Communications, Policy, and Education
- VII. NHGRI Intramural Research Program

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NHGRI Celebrates Bettie Graham's 50 Years of Federal Service



Departure of Extramural Program Director



Tina Gatlin, Ph.D.

New Extramural Program Director



Alexander Arguello, Ph.D.



Chris Wellington, B.S. Computational Genomics and Data Science Program Director



Shurjo Sen, Ph.D. Computational Genomics and Data Science Program Director

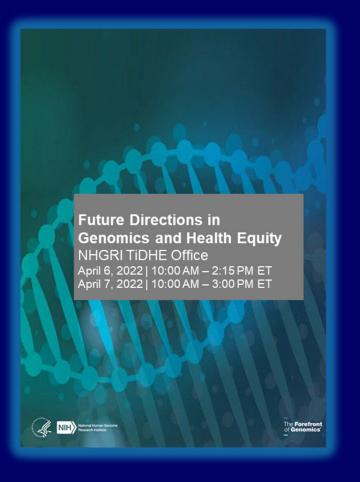


Asiyah Yu Lin, M.D., Ph.D. Data and Technology Advancement (DATA) Scholar



Elena Ghanaim, M.A. Policy Advisor for Data Science and Sharing

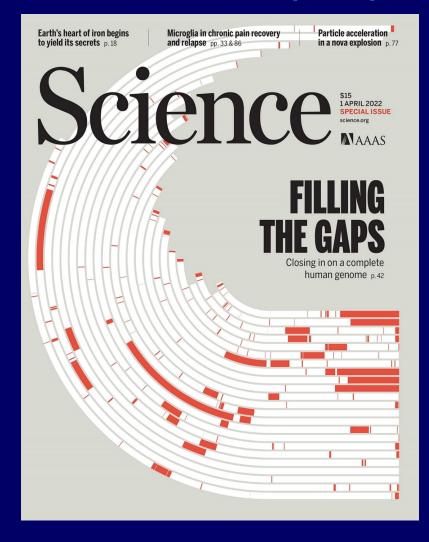
Future Directions in Genomics and Health Equity Research Workshop



 Identify research gaps and opportunities that help address health disparities and improve health equity in genomics

Over 300 participants

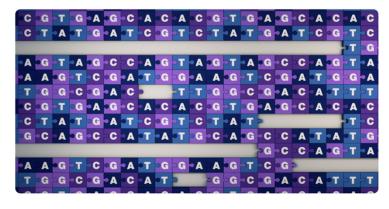
Telomere-to-Telomere (T2T) Consortium



Telomere-to-Telomere (T2T) Consortium

Top Tweet earned 200K impressions

Scientists in the Telomere-to-Telomere (**#T2T**) consortium have published the first complete, gapless human genome sequence. This can lead to a better understanding of our genomic variations and provide insight for missing heritability and human disease. genome.gov/T2T pic.twitter.com/tDpQ5cGnQo



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SCIENTIFIC Subscribe \equiv AMERICAN. SHARE GENETICS | OPINION **Completing the Human Genome Sequence (Again)** The Telomere-to-Telomere consortium just sequenced the tricky final 10 percent of the essentially complete human genome By Eric D. Green on March 31, 2022

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Acting Science Advisor to President Biden



Francis Collins, M.D., Ph.D.

Acting Director, Office of Science and Technology Policy



Alondra Nelson, Ph.D.

New Commissioner, Food and Drug Administration



Robert Califf, M.D.

Ned Sharpless Steps Down as Director, National Cancer Institute



Norman Sharpless, M.D.

Douglas Lowy, M.D.

New CEO, Foundation for the NIH





Julie Gerberding, M.D., M.P.H.

NIH Implements Harassment Reporting Requirements for Grantees

NIH National Institutes of Health

Tracker: 6

Became Law

THE NIH DIRECTOR

May 10, 2022

Congress strengthens NIH's ability to address harassment in NIH-funded activities

🗟 🖬 f 🔰 +

Ensuring a safe workplace where both people and science can thrive has long been the goal of NIH policies on addressing harassment. However, a 2018 National Academies report found "no evidence that current policies, procedures, and approaches have significantly reduced sexual harassment in academic sciences, engineering, and medicine." The report also found that a culture of harassment with persistent microaggressions is driving women away from science. It was crystal clear that NIH and other federal agencies needed to do more to end the culture of harassment and ensure women and other groups their rightful place in science.

F Citation 🖾 Subscribe 🖪 Share/Save 🗩 Site Feedback H.R.2471 - Consolidated Appropriations Act, 2022 117th Congress (2021-2022) | Get alerts LAW Hide Overview X Sponsor: Rep. Jeffries, Hakeem S. [D-NY-8] (Introduced 04/13/2021) Committees House - Foreign Affairs | Senate - Foreign Relations Committee Meetings: 04/21/21 2:00PM Latest Action: 03/15/2022 Became Public Law No: 117-103. (All Actions) There have been 8 roll call votes Roll Call Votes: The House Rules Committee Print, joint explanatory statements, and H.Rept. 117-269 from the Notes: House Rules Committee are available

Introduced > Passed House > Passed Senate > Resolving Differences >

CONGRESS.GOV

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To President

Sian In

Fiscal Year 2022 Appropriations

	FY2022 Enacted Labor-HHS Appropriation	Change from FY2021
NIH	\$45 B*	\$2.25 B (5.3%)
NHGRI	\$639.0 M	\$23.3 M (3.8%)

Fiscal Year 2023 Appropriations

	FY2023 President's Budget	Change from FY2022
NIH	\$49 B**	\$4 B (8.9%)

* Includes \$1 billion for ARPA-H** Includes \$5 billion for ARPA-H

Advanced Research Projects Agency for Health (ARPA-H)

 To benefit the health of all Americans by catalyzing health breakthroughs that cannot readily be accomplished through traditional research or commercial activity



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Mourning the Loss of Arthur Riggs





International Common Disease Alliance (ICDA)

2nd ICDA Virtual Scientific Plenary Meeting in March

New Executive Committee

 Virtual Community Town Halls in April



Global Biodata Coalition (GBC)







Guy Cochrane, D.Phil.

Elected to National Academy of Sciences

Howard Cedar Job Dekker Michael Elowitz Mark Johnston Erik Jorgensen Rasmus Nielsen Julie Segre Jay Shendure Len Zon



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Centers for Excellence in Genomic Science (CEGS)

New Awards:



David Haussler -University of California, Santa Cruz

> Center for Live Cell Genomics



Lucila Ohno Machado -University of California, San Diego

Center for Admixture Science and Technology



Samie Jaffrey - Weill Medical College of Cornell University

Center for Genomic Information Encoded by RNA Nucleotide Modifications

PAR-22-107 Next due date: June 22, 2022

Centers of Excellence in Genomic Science (RM1 Clinical Trial Optional)



Clinical Sequencing Evidence-Generating Research Program

Genetics in Medicine (2021) ==, 1–11	Genetics Medicine		
ELSEVIER	w.journals.elsevier.com/genetics-in-medicine		
ARTICLE			
Genome sequencing as a first-line diagnostic test for			
hospitalized infants			

- Utility of genome sequencing in infant cohort
- Of 367 infants that received genome sequencing, 30% received a genetic diagnosis
- 57% of detected genomic variants not detected by other genetic tests
- Participants from ethnically and racially diverse backgrounds as well as rural, medically underserved areas



Clinical Genome Resource (ClinGen) Justice, Equity, Diversity, and Inclusion Action Plan

- Contribute data, tools, and resources to the genomic knowledgebase to enhance equity in patient care
- Develop recommendations for the use of population descriptors in genetics and genomics
- Foster diversity, equity, and inclusion in the ClinGen workforce



Get Started - About Us - Curation Activities - Working Groups - Expert Panels - Documents - Tools Q

Justice, Equity, Diversity, and Inclusion (JEDI) Advisory Board

ClinGen is dedicated to building authoritative resources that define the clinical relevance of genes and variants for use in precision medicine and research. As part of our commitment to enhance diversity across clinical genomics, we have created a detailed action plan that organizes specific activities of the ClinGen grants in the format of Domains. An external Justice, Equity, Divserity, and Inclusion (JEDI) Advisory Board will periodically review progress on the JEDI Action Plan and provide feedback to the ClinGen Steering Committee.

Click <u>HERE</u> to access ClinGen's full JEDI plan. Please note the JEDI plan is still undergoing review and is not final/may change in the future.

Membership 🔮



Global Genomic Medicine Collaborative (GGMC)

Vision: Realizing the full potential of genomic medicine worldwide to improve individual and population health

Mission: To identify barriers and solutions, lead and develop programs, and global leverage expertise to bring genomic medicine to low-resource settings worldwide

International Hundred K+ Cohorts Consortium (IHCC) Global Genomic Medicine Consortium (G2MC) Potential additional programs

2021 Annual Report

Technology Development Program



Funding Opportunities:

 Technology Development for Single-Molecule Protein Sequencing

RFA-HG-21-001 (R01, also linked R21 RFA and R43/R44 PAR)

Next due date: June 15, 2022

 Advancing Genomic Technology Development for Research and Clinical Application
NOT-HG-21-018, Notice of Special Interest (NOSI)
R01, R21, R41/R42, & R43/R44
Standard receipt dates

Technology Development Program

Outreach events organized by the Technology Development Coordinating Center (TDCC) at the Jackson Laboratory



 Understanding Cells in Context: Spatial Transcriptomics

May 23, 2022

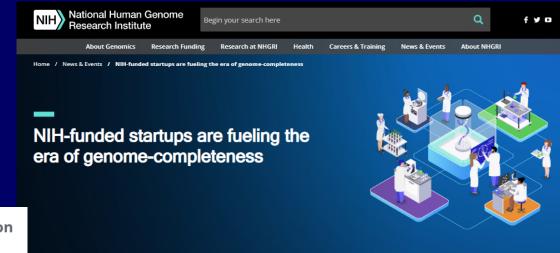
 Understanding Recent Advances in Genome Technology Development July 7, 2022

Both events are free, virtual, and open to the public

Small Business Program Grantee Success Stories

Features NHGRI-funded companies Circulomics, Arima Genomics, and Dovetail Genomics

"Small businesses are the heartbeat of innovation in the United States, but they face serious challenges when it comes to raising money to develop their ideas. Front-end money like that provided by NHGRI SBIR grants allows entrepreneurs to focus more on technology building and less on scrambling for early investments. "



PRABARNA GANGULY, PH.D. | FEBRUARY 03, 2022 | SPRESS CONTACT

Small businesses with NHGRI grants were instrumental in contributing to the completion of the human genome sequence, redefining the future of genomics.

Training and Career Development Program



- 7th Annual Training and Career Development Program Meeting
- Hosted by Duke University
- 227 in-person attendees and 157 virtual attendees

Diversity, Equity, Inclusion, and Accessibility (DEIA) in the Genomics Workforce Award

- Genomics Workforce DEIA Award
- Extramural investigators: Early-Stage Investigator (ESI) Established Investigator(s)
- Nominees must have NHGRI grant funding
- Due date: September 1, 2022





Genomics Workforce Diversity Funding Opportunities

 RFA-HG-22-004: Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Enhance Diversity (GREAT) Receipt date: July 1, 2022

 RFA-RM-22-008: NIH Institutional Recruitment for Sustainable Transformation (FIRST) Program: FIRST Cohort Receipt date: July 12, 2022

 PAR-20-153: NIH Science Education Partnership Award (SEPA) Receipt date: July 13, 2022

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Human Heredity & Health in Africa (H3Africa)

> Nat Commun. 2022 Feb 22;13(1):926. doi: 10.1038/s41467-021-27917-x.

Short- and long-read metagenomics of urban and rural South African gut microbiomes reveal a transitional composition and undescribed taxa

Fiona B Tamburini ¹, Dylan Maghini ¹, Ovokeraye H Oduaran ², Ryan Brewster ³, Michaella R Hulley ² ⁴, Venesa Sahibdeen ⁴, Shane A Norris ⁵ ⁶, Stephen Tollman ⁷ ⁸, Kathleen Kahn ⁷ ⁸, Ryan G Wagner ⁷ ⁸, Alisha N Wade ⁷, Floidy Wafawanaka ⁷, F Xavier Gómez-Olivé ⁷ ⁸, Rhian Twine ⁷, Zané Lombard ⁴, H3Africa AWI-Gen Collaborative Centre; Scott Hazelhurst [#] ⁹ ¹⁰, Ami S Bhatt [#] ¹¹ ¹² ¹³ > Epigenomics. 2022 Jan;14(1):11-25. doi: 10.2217/epi-2021-0310. Epub 2021 Dec 8.

Leukocyte methylomic imprints of exposure to the genocide against the Tutsi in Rwanda: a pilot epigenome-wide analysis

Clarisse Musanabaganwa ¹ ² ³ ⁴, Agaz H Wani ³, Janelle Donglasan ³, Segun Fatumo ⁵ ⁶, Stefan Jansen ⁷, Jean Mutabaruka ², Eugene Rutembesa ², Annette Uwineza ¹, Erno J Hermans ⁴, Benno Roozendaal ⁴, Derek E Wildman ³, Leon Mutesa ¹, Monica Uddin ³

> BMJ Glob Health. 2022 Jan;7(1):e007184. doi: 10.1136/bmjgh-2021-007184.

Guideline for feedback of individual genetic research findings for genomics research in Africa

Alice Matimba ¹, Stuart Ali ², Katherine Littler ³, Ebony Madden ⁴, Patricia Marshall ⁵, Sheryl McCurdy ⁶, Victoria Nembaware ⁷, Laura Rodriguez ⁴, Janet Seeley ⁸, Paulina Tindana ⁹, Aminu Yakubu ¹⁰ ¹¹ ¹², Jantina de Vries ¹³, H3Africa Ethics and Community Engagement Working Group

- 600+ publications to date
- Uptake of Consortium recommendations into policies
- Final Consortium meeting in Nigeria



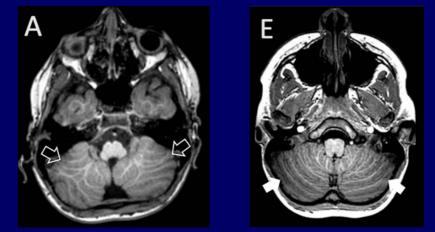
Undiagnosed Diseases Network (UDN)



Image from: https://www.modelmatcher.net/

Online investigator matchmaking tool

 Cross-disciplinary collaboration in rare disease research

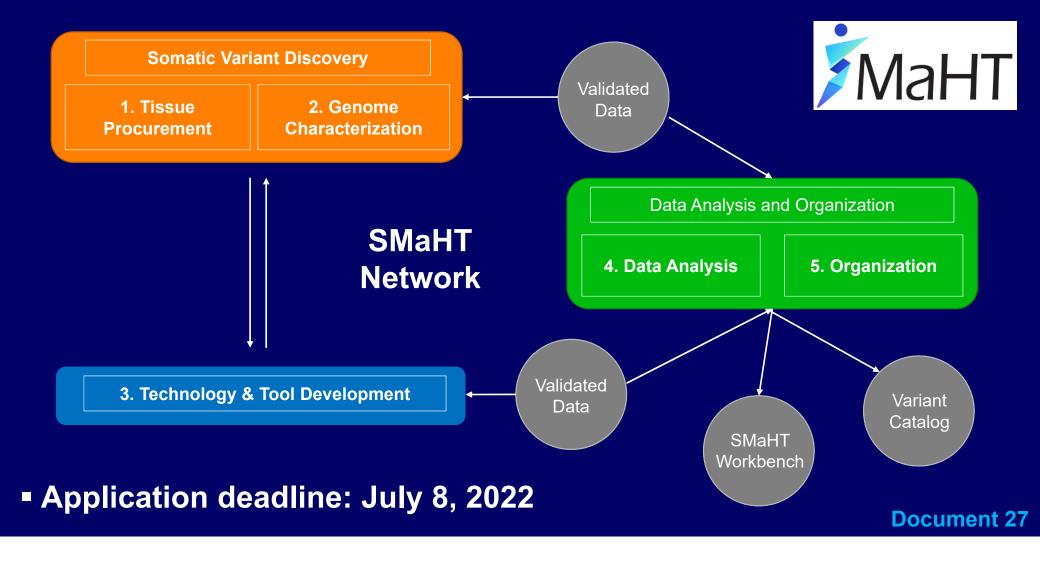


Neuroimaging features of the NAV2-related neurodevelopmental disorder (Fig. A) vs. age-matched control subject (Fig. E)

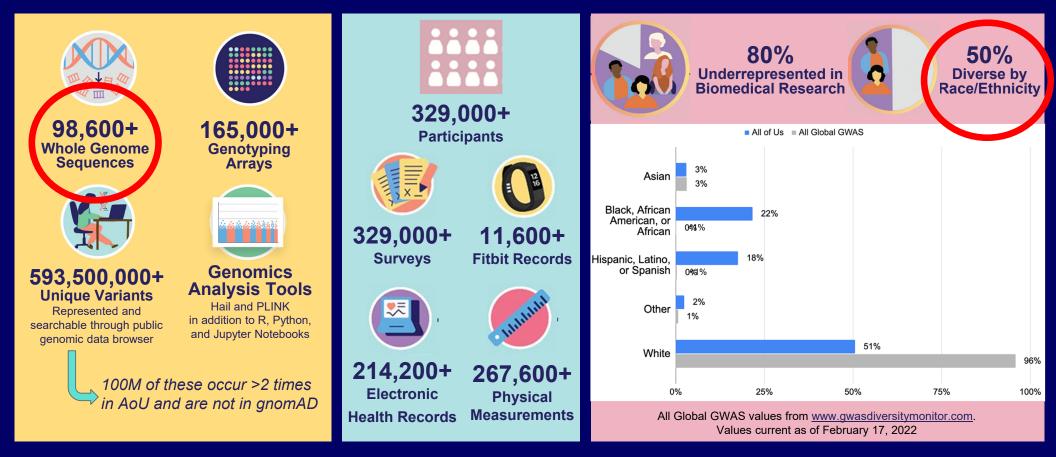
First human case of Neuron Navigator 2 (NAV2) gene mutation

 Functional analyses to identify crucial role of NAV2 in central nervous system development

Somatic Mosaicism Across Human Tissues (SMaHT)



RESEARCH PROGRAM



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NHGRI History of Genomics Program

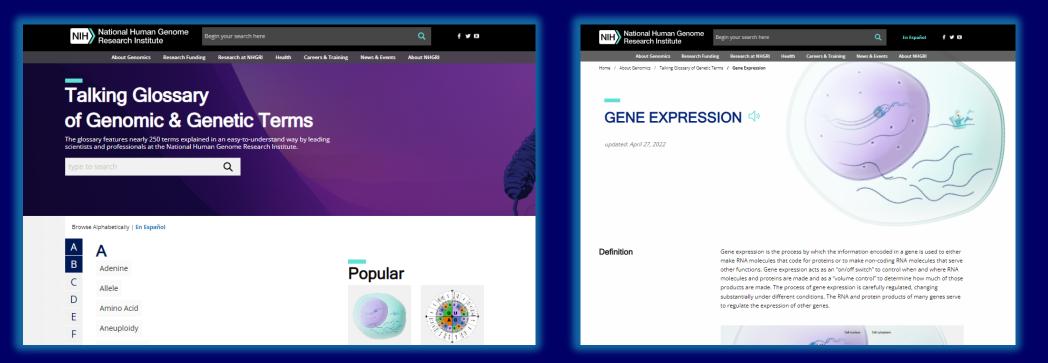
Roundtable Meetings



 Does genetic and genomic screening keep open the door to eugenics? May 25, 2022

The promise and perils of social and behavioral genomics December 2022

Talking Glossary of Genomic and Genetic Terms



- Audio recordings from 37 NHGRI experts
- 222 terms 142 with new illustrations and 15 with new animations

Genomics and the Media



Magdalena Skipper Editor in Chief, Nature;

Chief Editorial Advisor, Nature Research

June 30, 2022

Conversations with trailblazing science communicators



Apoorva Mandavilli **Reporter (Science and** Global Health), The New York Times

May 20, 2021



John Inglis Executive Director, Cold Spring Harbor Laboratory Press: Co-founder.

July 28, 2021



September 20, 2021



Dorothy Roberts Professor of Law and Sociology, Civil Rights, University of Pennsylvania; Author, Fatal Invention

November 4, 2021

Elizabeth Wayne

Assistant Professor of Chemistry, Carnegie Mellon University: Co-host. PhDivas Podcast

January 20, 2022



Joe Palca Science Correspondent, NPR

March 8, 2022

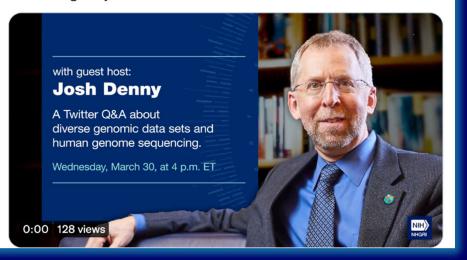
Ask Dr. Genome

...



Josh Denny 🤣 @AllofUsCEO · Mar 30

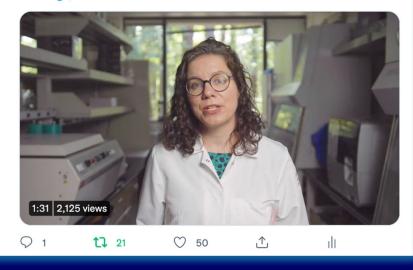
What questions do you have about diverse genomic datasets and sequencing research for precision medicine? The @NHGRI_Director and I are answering your questions now! Use the **#AskDrGenome** hashtag to follow along and join the conversation.





Eric Green 🤣 @NHGRI_Director · Apr 11

It has been very exciting to discuss how the **#T2T** Consortium completed the human genome sequence. I am having so much fun that I decided it is time for another **#AskDrGenome** Twitter Q&A! This time I will be joined by @KHMiga, co-leader of the **#T2T** Consortium!



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Communications Award Nominations



SHORTY AWARDS

HONORING THE BEST OF SOCIAL MEDIA AND DIGITAL

The Shorty Awards is an international awards competition honoring brands, agencies and organizations producing great work across digital and social platforms.

Best Twitter Presence finalists

- Maricopa County, AZ
- National Human Genome Research Institute
- Oklahoma Department of Wildlife Conservation
- Salt Lake Health Department, UT
- Washington State Department of Transportation

GOVERNMENT & POLITICS

NASA's #SolarTour: A Journey to Touch the Sun NASA

Reckoning with the complex history of genetics National Human Genome Research Institute

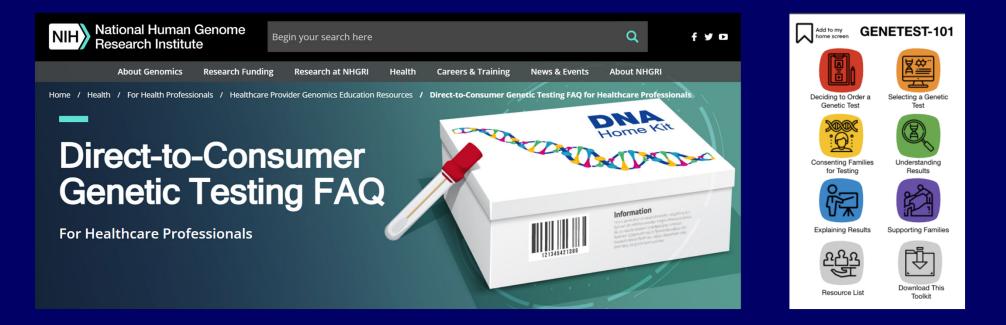
National DNA Day 2022







Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)



ISCC-PEG 11th Annual Meeting

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EURORDIS Lifetime Achievement Award



EURORDIS LIFETIME ACHIEVEMENT AWARD

William Gahl, M.D., Ph.D.

Election into American Society for Clinical Investigation





Neil Hanchard, D.Phil.

Samuel J. Heyman Service to America Medal Finalist



FOR PUBLIC SERVICE

SAMUEL J. HEYMAN SERVICE TO AMERICA MEDALS

Diana Bianchi, M.D.

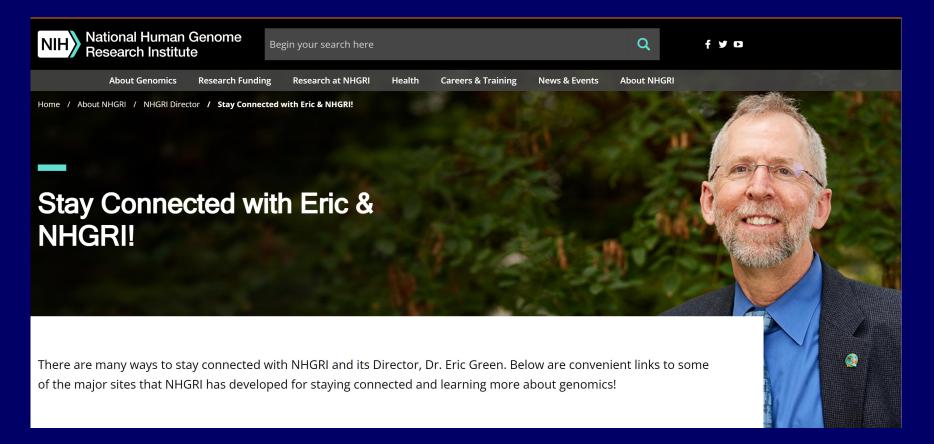
Intramural Research Program Blue Ribbon Panel

2022 – 2023 Panel Members:

Gail Jarvik, M.D., Ph.D. (co-chair) Lynn Jorde, Ph.D. (co-chair) Greg Barsh, M.D., Ph.D. Lon Cardon, Ph.D. Chanita Hughes-Halbert, Ph.D. Guillermina (Gigi) Lozano, Ph.D. Carole Ober, Ph.D. Neil Risch, Ph.D.



'One-Stop-Shop' to Stay Connected genome.gov/stayconnected





Thanks!



Special Thanks!

