

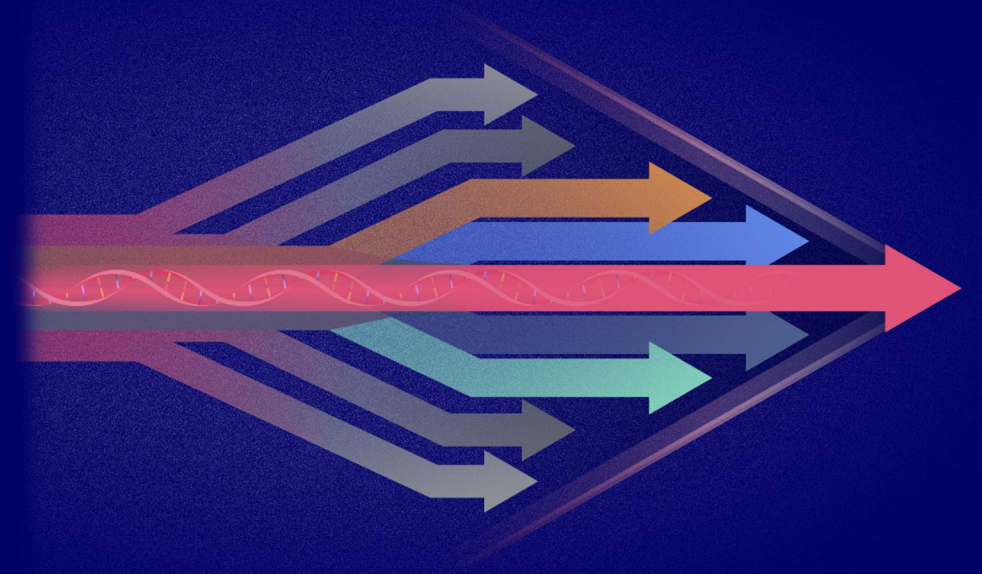


The **Forefront**
of **Genomics**[®]

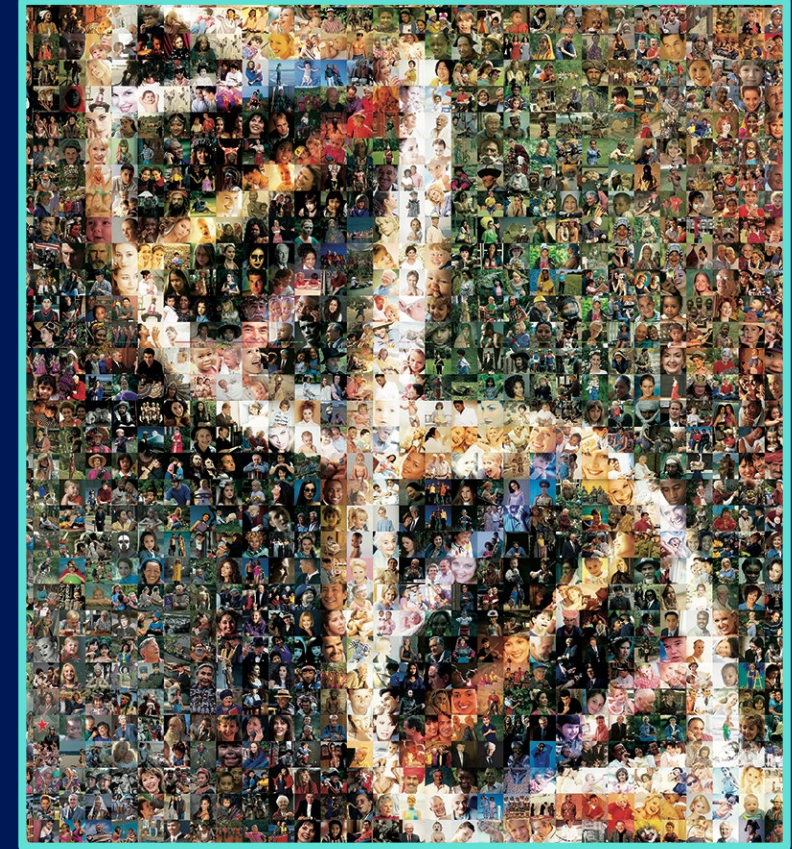
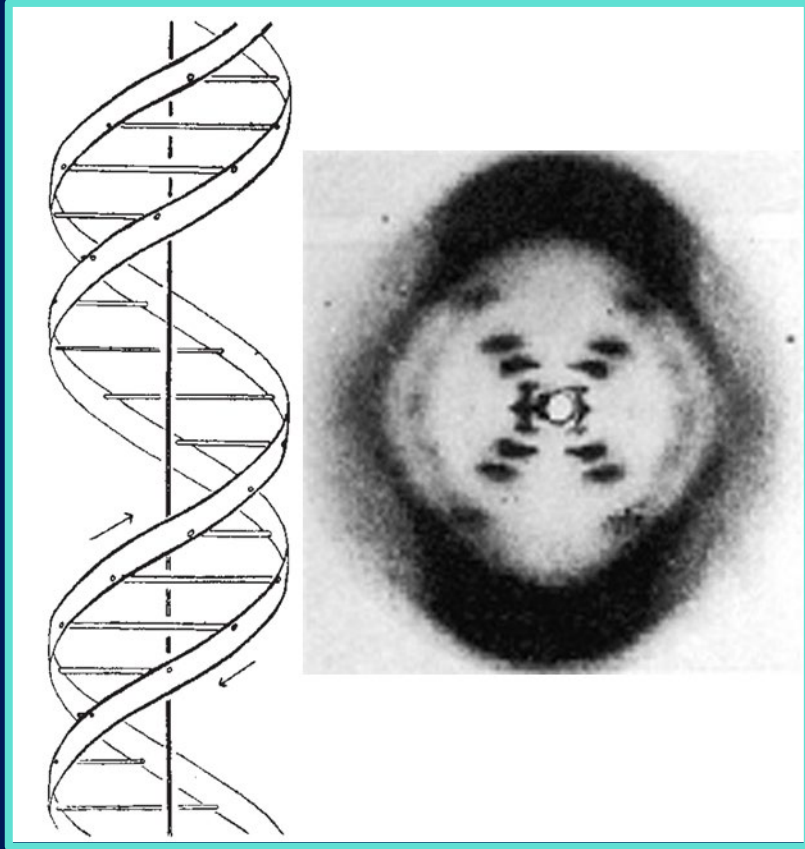
NHGRI Highlights

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

ISCC-PEG Annual Meeting
February 15, 2023



Genomics Anniversaries in 2023



Celebrating 3+ Decades of Genomics



**Human Genome Sequenced for First Time
by the Human Genome Project**



**Cost of Sequencing a Human Genome
Reduced >1 Million-Fold**



**Millions of Human
Genomes Sequenced**



**Profound Advances in Understanding
How the Human Genome Functions**



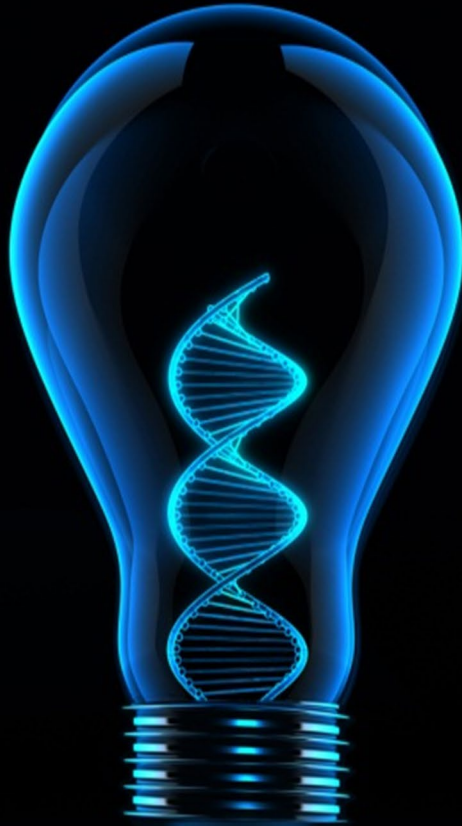
**Significant Advances in Unraveling the
Genomic Bases of Human Disease**



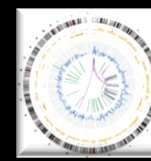
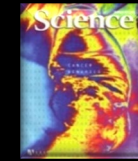
**Vivid Examples of Genomic Medicine
Now Emerging**



Genomic Medicine Implementation



Cancer Genomics



**Rare Genetic Disease
Diagnostics**



Prenatal Genomic Testing



Pharmacogenomics



**Genomic Risk Predictions
for Common Diseases**



???

But Genomics Still Faces Big Challenges

Clinically Understanding a Patient's Genome Sequence Remains Difficult

Genomics is Becoming Profoundly Relevant in Society

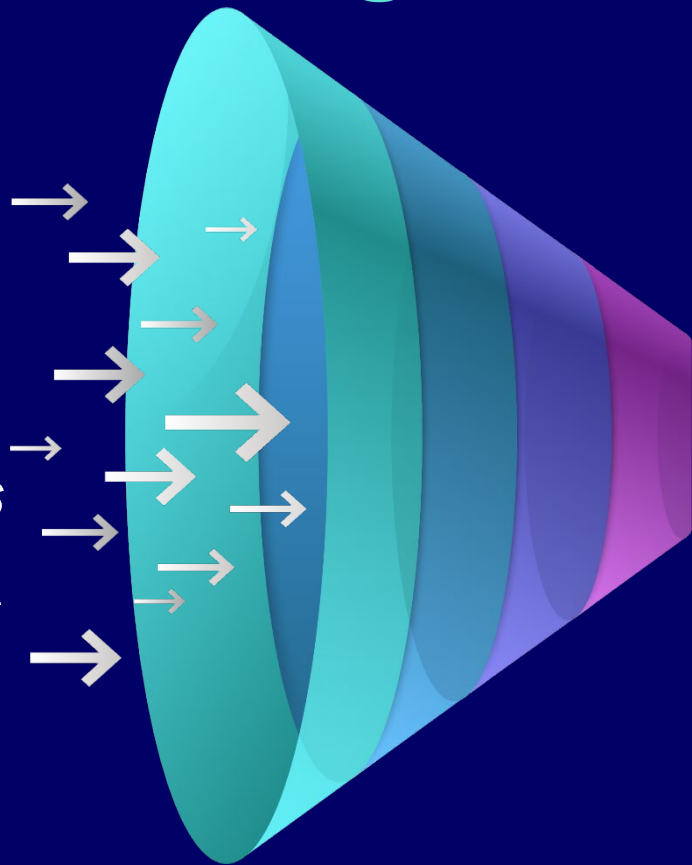
There is a Lack of Diversity in Genomics

Must Understand Roles of Both Genomic and Non-Genomic Factors in Human Health and Disease



From Strategic Planning to Vision Implementation

Ideas
Needs
Proposals
Obstacles



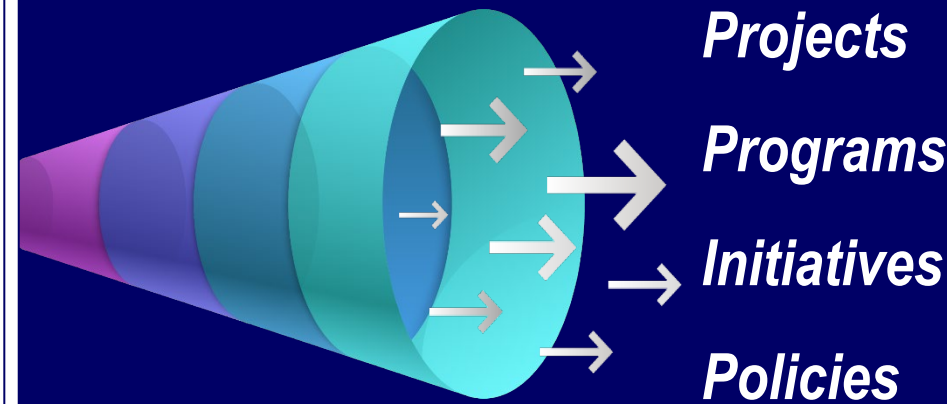
Strategic Planning Process

Feb 2018 to Sept 2020



2020 NHGRI Strategic Vision

October 2020



Implementation of Strategic Vision

2020 and Beyond

Projects
Programs
Initiatives
Policies

Building a Diverse Genomics Workforce: An NHGRI Action Agenda

The **Forefront**
of **Genomics**



The genomics workforce must become more diverse: a strategic imperative

Vence L. Bonham^{1,*} and Eric D. Green^{1,*}

AJHG (2021)



genome.gov/workforcediversity

Training, Diversity, and Health Equity (TiDHE) Office



- **Develop, support, and coordinate NHGRI programs that foster careers in genomics**
- **Advise divisions and branches about genomics workforce diversity and health equity**
- **Provide strategic programmatic leadership to reduce health disparities and advance health equity**

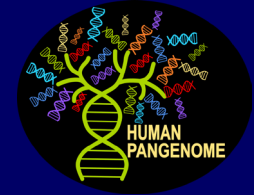
TiDHE Activities



- **Two Industry Roundtables on diversifying the genomics workforce**
- **Diversity, Equity, Inclusion, and Accessibility (DEIA) in the Genomics Workforce Award**
- **Several Genomics Workforce Diversity Funding Opportunities, including "Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Enhance Diversity (GREAT)" (RFA-HG-22-004)**

2020 NHGRI Strategic Vision: New Initiatives

- ‘Action Agenda’ for Enhancing Diversity of Genomics Workforce
- New Training Initiatives (e.g., genetic counselors & data scientists)
- Human Genome Reference Program
- Impact of Genomic Variation on Function (IGVF) Consortium
- Novel Synthetic Nucleic Acid Technology Development
- Developmental Genotype-Tissue Expression (dGTEx) Program
- eMERGE Genomics Risk Assessment and Management Network
- Polygenic Risk Scores in Populations of Diverse Ancestry
- More to come ...



NHGRI Funding for Genomic Medicine Training Modules for Healthcare Professionals

- **Duke University:** *Health Professional Rapid Personalized Learning Platform for Genomic Medicine*, Geoff Ginsburg, Susanne Haga
- **Columbia University:** *Preparing providers to integrate polygenic risk scores into clinical care*, Wendy Chung, M.D., Ph.D.
- **Mayo Clinic:** *Genetics and Genomics Essentials for Nursing Practice*, Timothy Curry, M.D., Ph.D.
- **University of Cincinnati/Cincinnati Children's Hospital Medical Center:** *Reduce or Prolong the Diagnostic Odyssey: You Decide*, Laura Ramsey, Ph.D., Cindy Prows, M.S.N., APRN, FAAN, Loren Pena, M.D., Heide Aungst, M.A.

NHGRI Funding for Genomic Medicine Training Modules for Healthcare Professionals

- **Indiana University/University of Pittsburgh:** *Test2Learn: Modular multidisciplinary pharmacogenomics education*, [Todd Skaar/Philip Empey](#)
- **The University of Texas Health Science Center at Houston:** *UTHealth Adult Cardiovascular Genomics Certificate Program*, [Siddharth Prakash](#), M.D., Ph.D.



The screenshot shows the top portion of a web page for 'Future Medicine'. The header includes the 'Future Medicine' logo and a navigation bar with links for 'JOURNALS', 'BOOKS', 'ABOUT', 'AUTHOR GUIDE', and 'SUBMIT'. Below the navigation bar, the article title 'Development of Competency-based Online Genomic Medicine Training (COGENT)' is displayed in red. The authors listed are Susanne B Haga, Wendy K Chung, Luis A Cubano, Timothy B Curry, Philip E Empey, Geoffrey S Ginsburg, Kara Mangold, Christina Y Miyake, Siddharth K Prakash, Laura B Ramsey, and Robb Rowley. A 'See all authors' link is provided. At the bottom, the publication date is 'Published Online: 23 Nov 2022' and the DOI is 'https://doi.org/10.2217/pme-2022-0101'.

Future Medicine 

JOURNALS ▾ BOOKS ABOUT ▾ AUTHOR GUIDE ▾ SUBMIT

PERSONALIZED MEDICINE, AHEAD OF PRINT | SPECIAL REPORT

Development of Competency-based Online Genomic Medicine Training (COGENT)

Susanne B Haga  , Wendy K Chung , Luis A Cubano, Timothy B Curry ,

Philip E Empey , Geoffrey S Ginsburg , Kara Mangold, Christina Y Miyake, Siddharth K Prakash , Laura B Ramsey , Robb Rowley, ... See all authors ▾

Published Online: 23 Nov 2022 | <https://doi.org/10.2217/pme-2022-0101>

Training and Career Development

Response to Training Task Force Recommendations

- **Mentored Research Experiences for Genetic Counselors (R25)**
- **Research Experience in Genomic Research for Data Scientists (R25)**
- **NHGRI Predoctoral to Postdoctoral Transition Award to Promote Diversity (F99/K00)**
- **K18 – Short-term Mentored Research Career Enhancement Award to Promote Diversity (K18)**

Talking Glossary of Genomic and Genetic Terms

National Human Genome Research Institute

Begin your search here

About Genomics Research Funding Research at NHGRI Health Careers & Training News & Events About NHGRI

Talking Glossary of Genomic & Genetic Terms

The glossary features nearly 250 terms explained in an easy-to-understand way by leading scientists and professionals at the National Human Genome Research Institute.

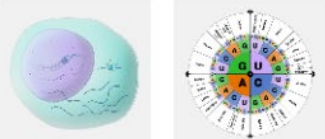
type to search

Browse Alphabetically | En Español

A

- A**
- B** Adenine
- C** Allele
- D** Amino Acid
- E** Aneuploidy
- F**

Popular



National Human Genome Research Institute

Begin your search here

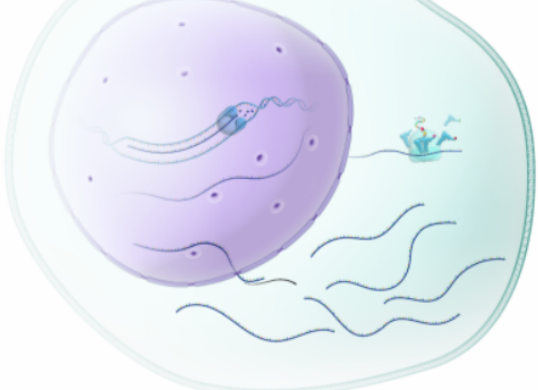
En Español

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Home / About Genomics / Talking Glossary of Genetic Terms / Gene Expression

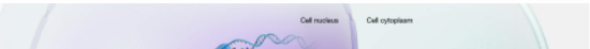
GENE EXPRESSION

updated: April 27, 2022



Definition

Gene expression is the process by which the information encoded in a gene is used to either make RNA molecules that code for proteins or to make non-coding RNA molecules that serve other functions. Gene expression acts as an "on/off switch" to control when and where RNA molecules and proteins are made and as a "volume control" to determine how much of those products are made. The process of gene expression is carefully regulated, changing substantially under different conditions. The RNA and protein products of many genes serve to regulate the expression of other genes.



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

New and Refreshed NHGRI Fact Sheets

Fact Sheet

Human Genomic Variation

NHGRI recently released a fact sheet on human genomic variation, which provides explanatory information and graphics about variation in human genome sequences. Human genomic variation is highly relevant in genomic medicine as healthcare professionals are increasingly learning how to use information about patients' genomic variants to manage their medical care.

[Learn More](#)



NHGRI History of Genomics Program

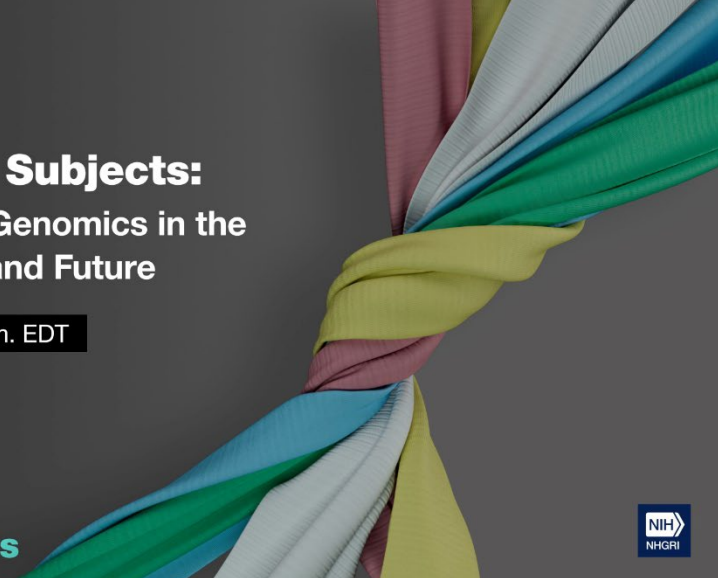

Recent Events



Irreducible Subjects:
Disability and Genomics in the
Past, Present and Future

Oct. 6-7, 2022 10 a.m. EDT
Webinar Day 1

#NHGRIevents

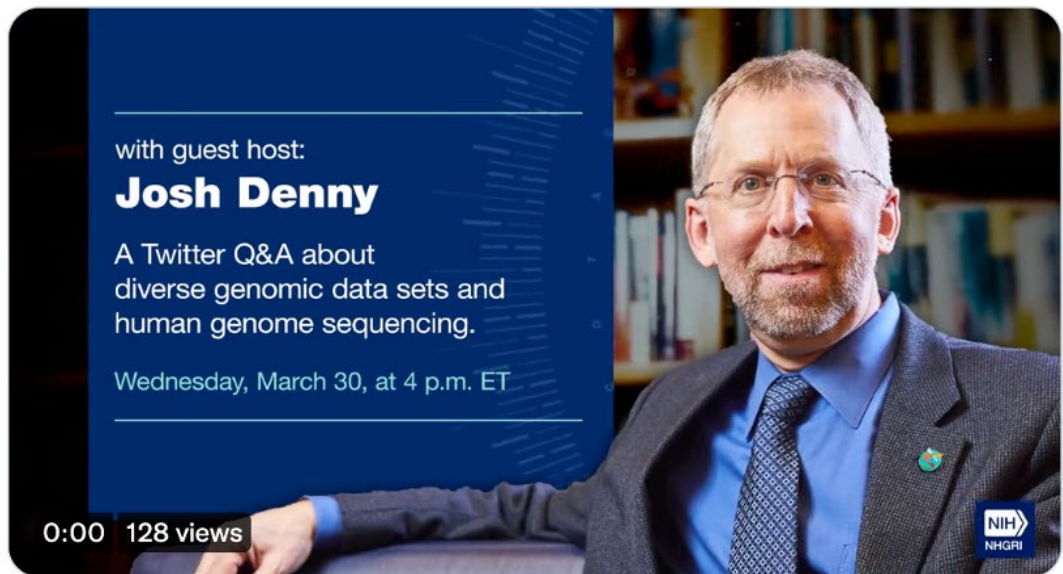


Ask Dr. Genome



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!



1 21 50

'One-Stop-Shop' to Stay Connected

genome.gov/stayconnected

NIH National Human Genome Research Institute

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Home / About NHGRI / NHGRI Director / Stay Connected with Eric & NHGRI

Stay Connected with Eric & NHGRI!

There are many ways to stay connected with NHGRI and its Director, Dr. Eric Green. Below are convenient links to some of the major sites that NHGRI has developed for staying connected and learning more about genomics!



Genome.gov

Explore the official website of the National Human Genome Research Institute.



The Genomics Landscape

Subscribe to Dr. Eric Green's monthly newsletter summarizing NHGRI and genomics highlights.



Dr. Eric Green on Twitter

Follow Dr. Eric Green on Twitter @NHGRI_Director.



2020 NHGRI Strategic Vision

Read NHGRI's strategic vision for improving human health at The Forefront of Genomics.



NHGRI Brochure

Learn more about NHGRI and the institute's history, core principles and values, research portfolio, and various other programs.



Talking Glossary of Genetic and Genomic Terms

Gain a better understanding of hundreds of genetics and genomics terms and concepts using this educational NHGRI resource.



Building a Diverse Genomics Workforce

Read about NHGRI's plans to increase the number of individuals from diverse backgrounds in the genomics workforce.



GenomeTV

Watch GenomeTV, a vast collection of NHGRI videos, including live event coverage, scientific seminars, and educational films.



NHGRI's Oral History Collection

View video interviews of those with perspectives and experiences in genomics programs and other NHGRI endeavors.

Among the Best of Eric Green

Videos

Scale of the Human Genome (with animations) - Eric Green
December 2, 2019

GM XIII: 2020 NHGRI Strategic Vision Genomics 2020 Strategic Planning Process
February 9, 2021

Human Genome Project 30th Anniversary Selfie: Eric Green
October 1, 2020

Webinar By MOHAP: Dr. Eric Green On The Human Genome Project
March 25, 2021

Podcasts

DNA TODAY PODCAST (APRIL 26, 2022)
Eric Green on the Complete Human Genome Project

AMERICAN METAPHORPODCAST (MARCH 30, 2021)
The Butterfly Effect

TRAILBLAZERS WITH WALTER ISAACSON (DECEMBER 16, 2020)
Genomics: Rewriting Our Health

GENETICS LANDSCAPED (OCTOBER 22, 2020)
The Past, Present and Future of the Human Genome Project

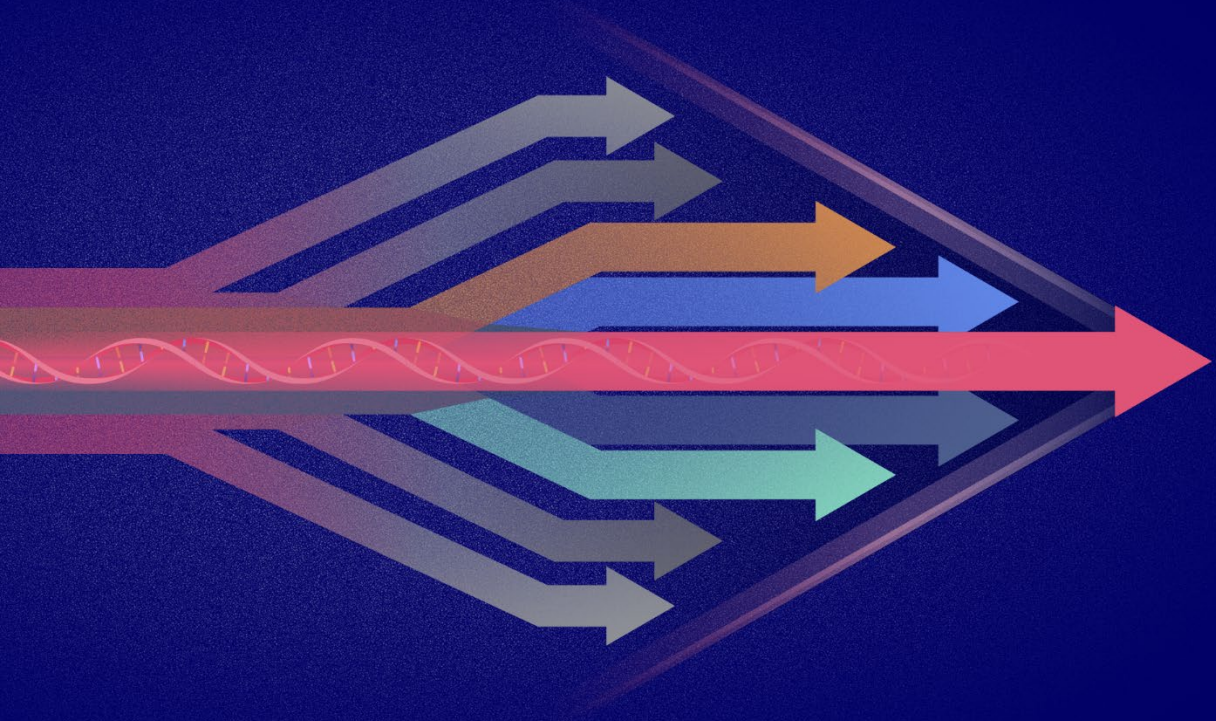
Op-Eds/Commentaries

Completing the Human Genome Sequence (Again)
Scientific American, March 31, 2022

An Anti-racist Action Plan for Studying the Human Genome
The Hill, September 16, 2021

A Vision for the Next Decade of Human Genomics Research
Scientific American, October 28, 2020

Questions/Comments?





The **Forefront**
of **Genomics**[®]