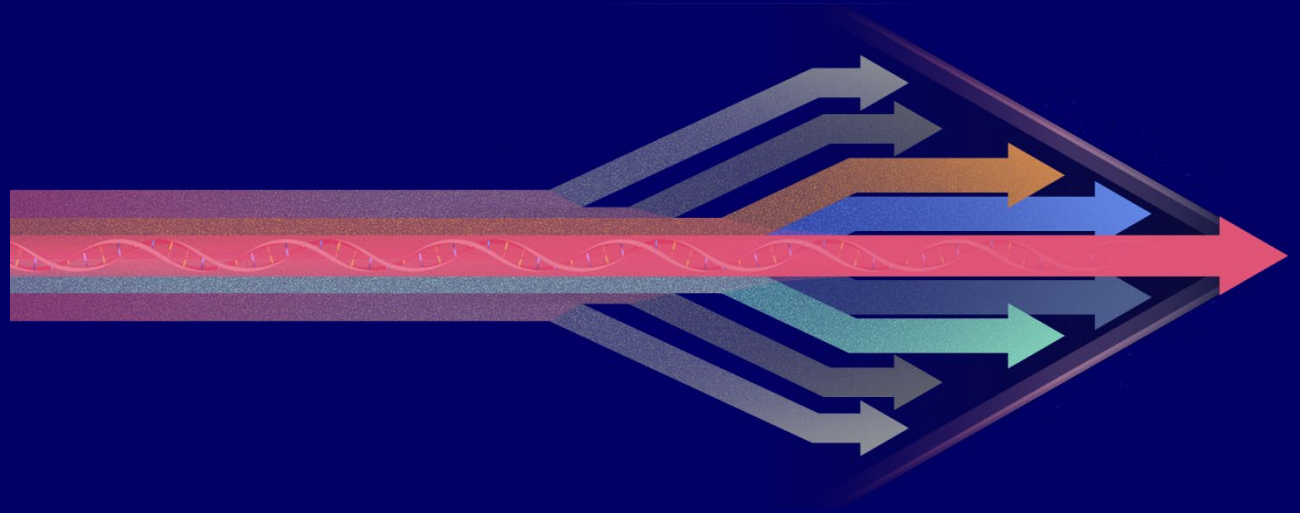


The **Forefront** of **Genomics**®

A 2020 Strategic Vision for Improving Human Health at *The Forefront of Genomics*

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

Director, NHGRI

National Human Genome Research Institute

Strategic Visions for Human Genomics

1991-1995

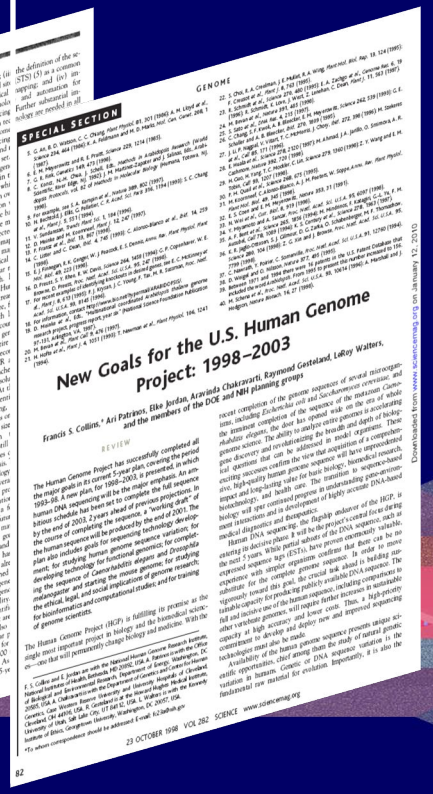
1993-1998


POLICY FORUM

A New Five-Year Plan for the U.S. Human Genome Project

Francis Collins and David Galas*

1998-2003


SPECIAL SECTION

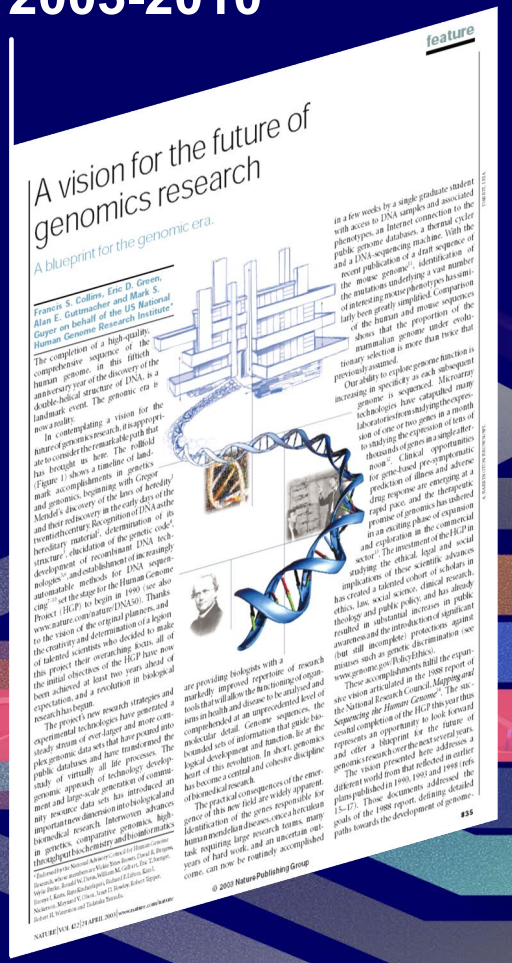
GENOME

New Goals for the U.S. Human Genome Project: 1998–2003

Francis S. Collins,* Ari Patrinos, Elke Jordan, Aravinda Chakravarti, Raymond Gesteland, LeRoy Walters, and the members of the DOE and NIH planning groups

REVIEW

2003-2010


feature

A vision for the future of genomics research

A blueprint for the genomic era.

Francis S. Collins, Eric D. Green, Alan E. Guttmacher and Mark S. Guyer on behalf of the US National Human Genome Research Institute*

2011-2020


PERSPECTIVE

Charting a course for genomic medicine from base pairs to bedside

Eric D. Green*, Mark S. Guyer* & National Human Genome Research Institute*

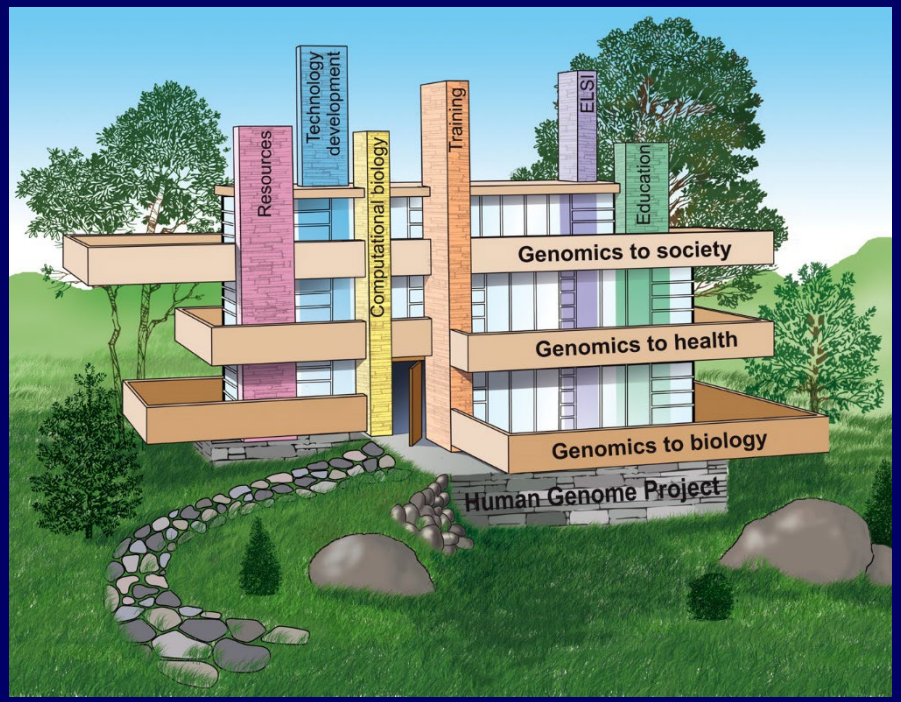
Different Eras for NHGRI

1990-2003

Human Genome Project

2003-2011

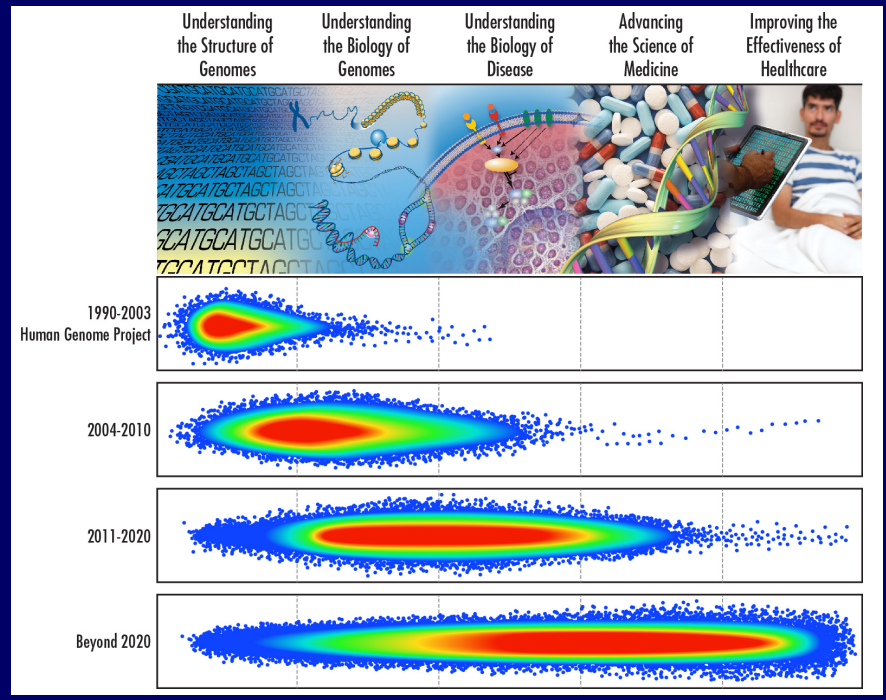
Building Upon the Human Genome Project



Collins et al. (2003)

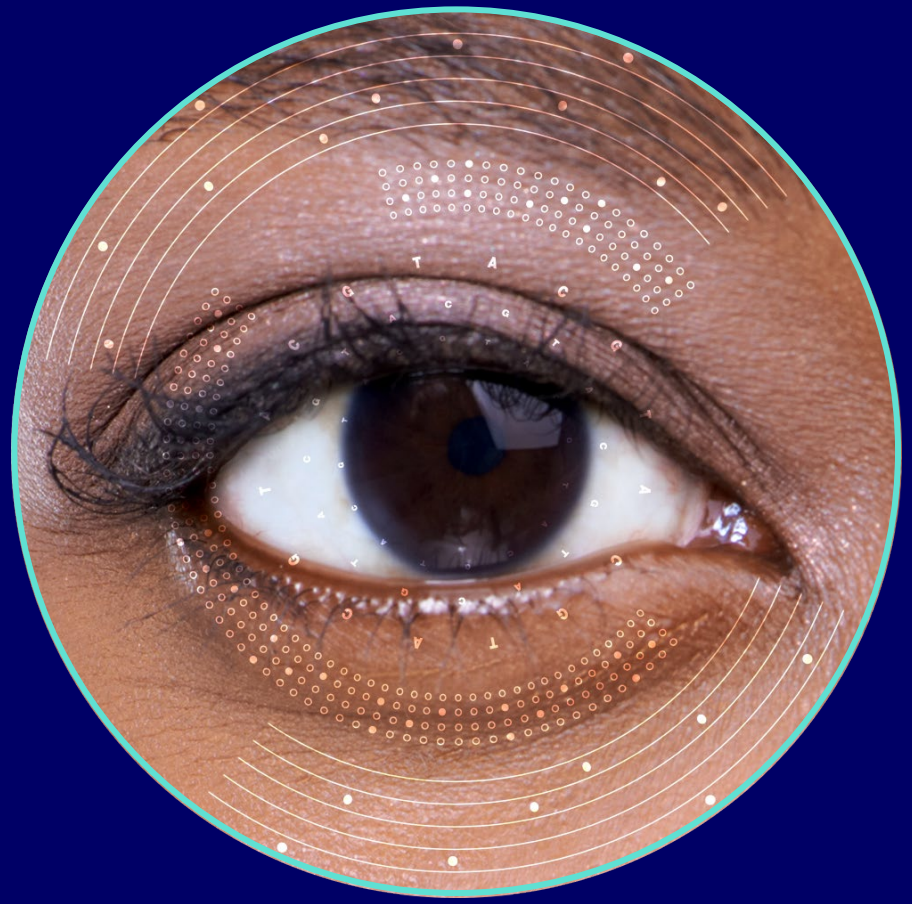
2011-2020

En Route to Genomic Medicine



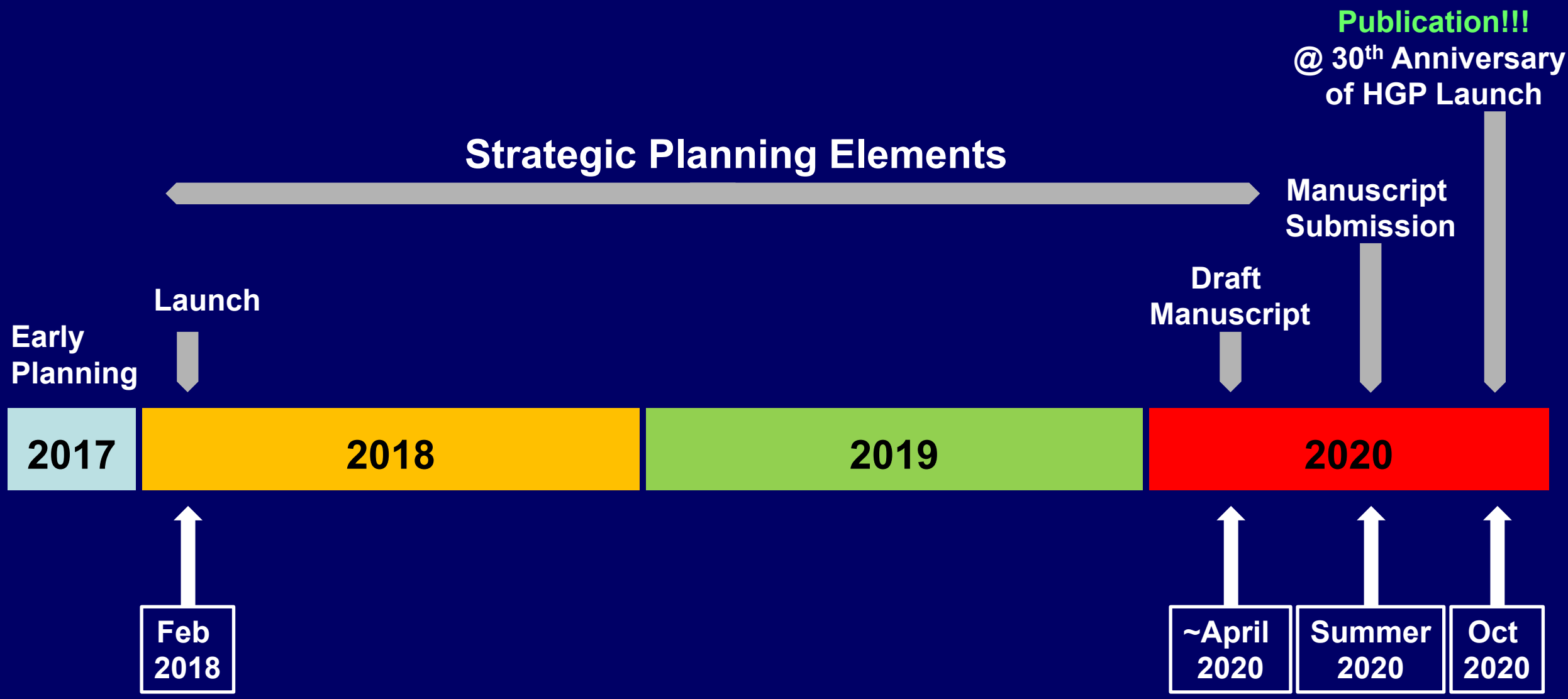
Green et al. (2011)

Genomics2020 Strategic Planning Process



Establishing a “2020 Vision for Human Genomics”

Genomics2020: Timeline



The Reality of Genomics as a Field

The **Forefront** of **Genomics**®

2020 NHGRI Strategic Vision

Perspective

Strategic vision for improving human health at The Forefront of Genomics

https://doi.org/10.1038/s41586-020-2817-4
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Check for updates

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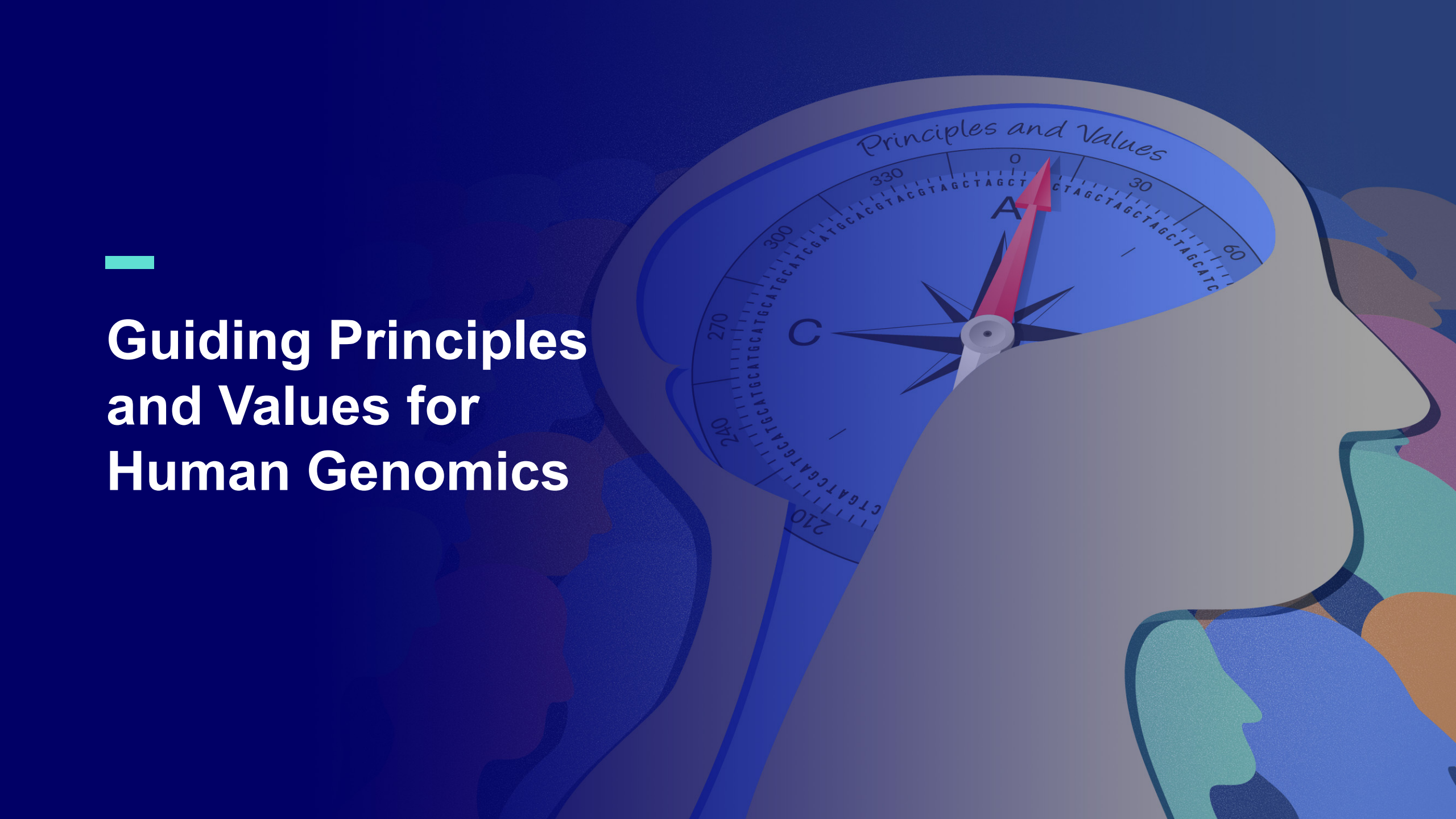
[1]National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA. ✉e-mail: egreen@nhgri.nih.gov

The **Forefront** of **Genomics**®

Nature (2020)

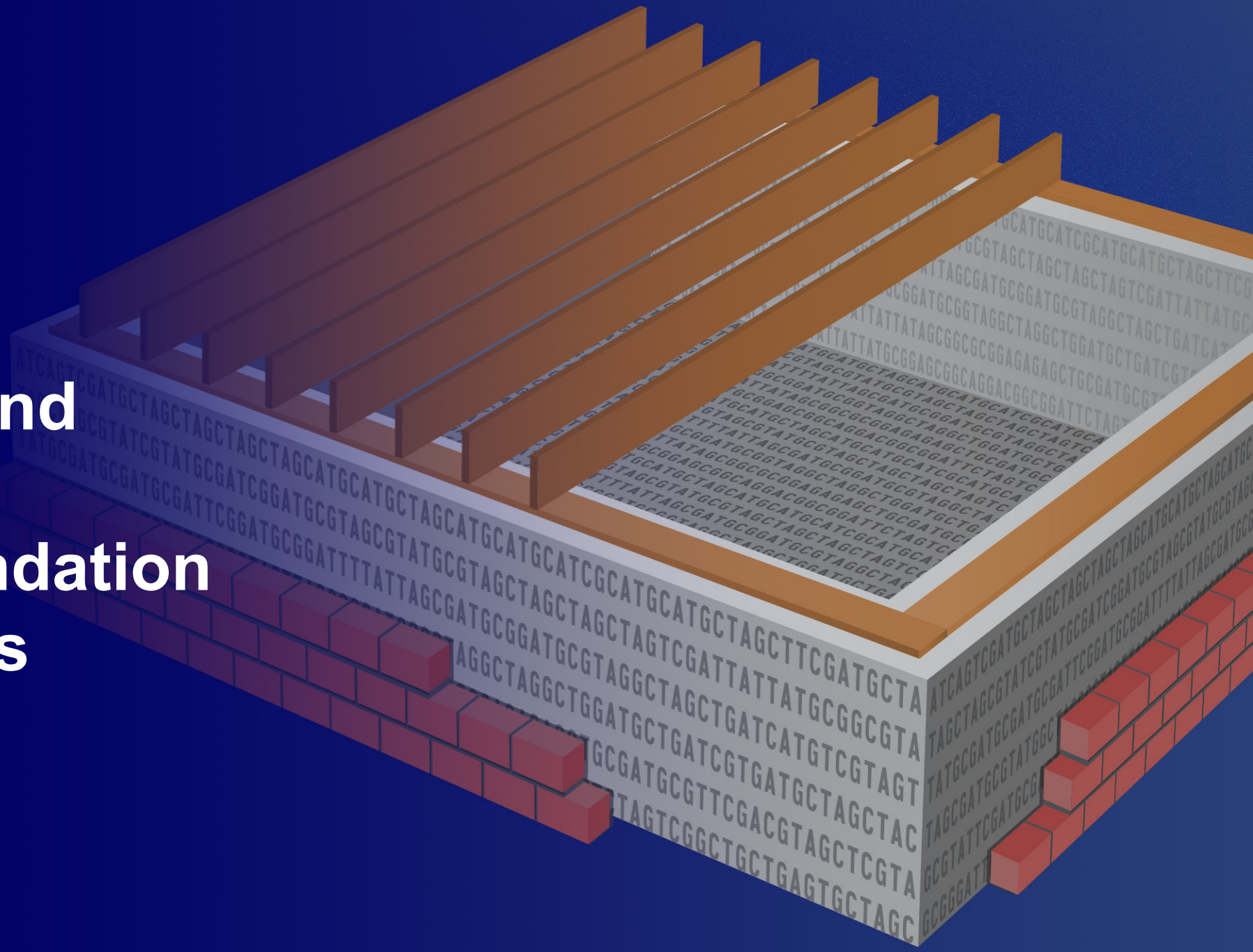
Guiding Principles and Values for Human Genomics



Guiding Principles and Values for Human Genomics

- Diversity
- Equity and social justice
- Consortia-based ‘team science’
- Open science and data sharing
- Data standards

Sustaining and Improving a Robust Foundation for Genomics



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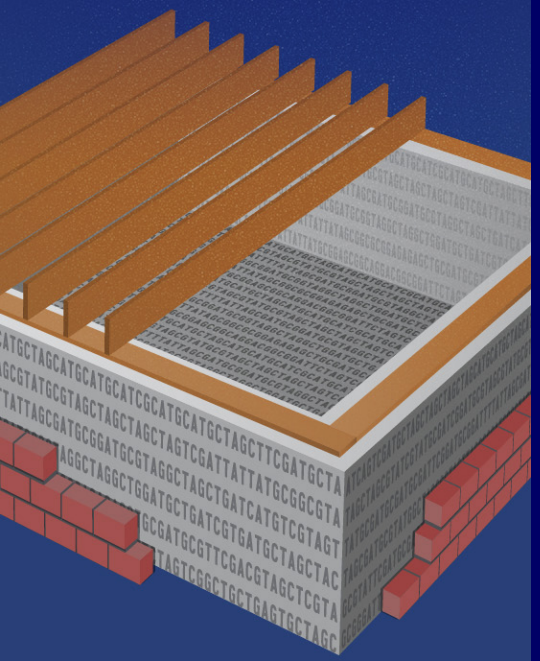
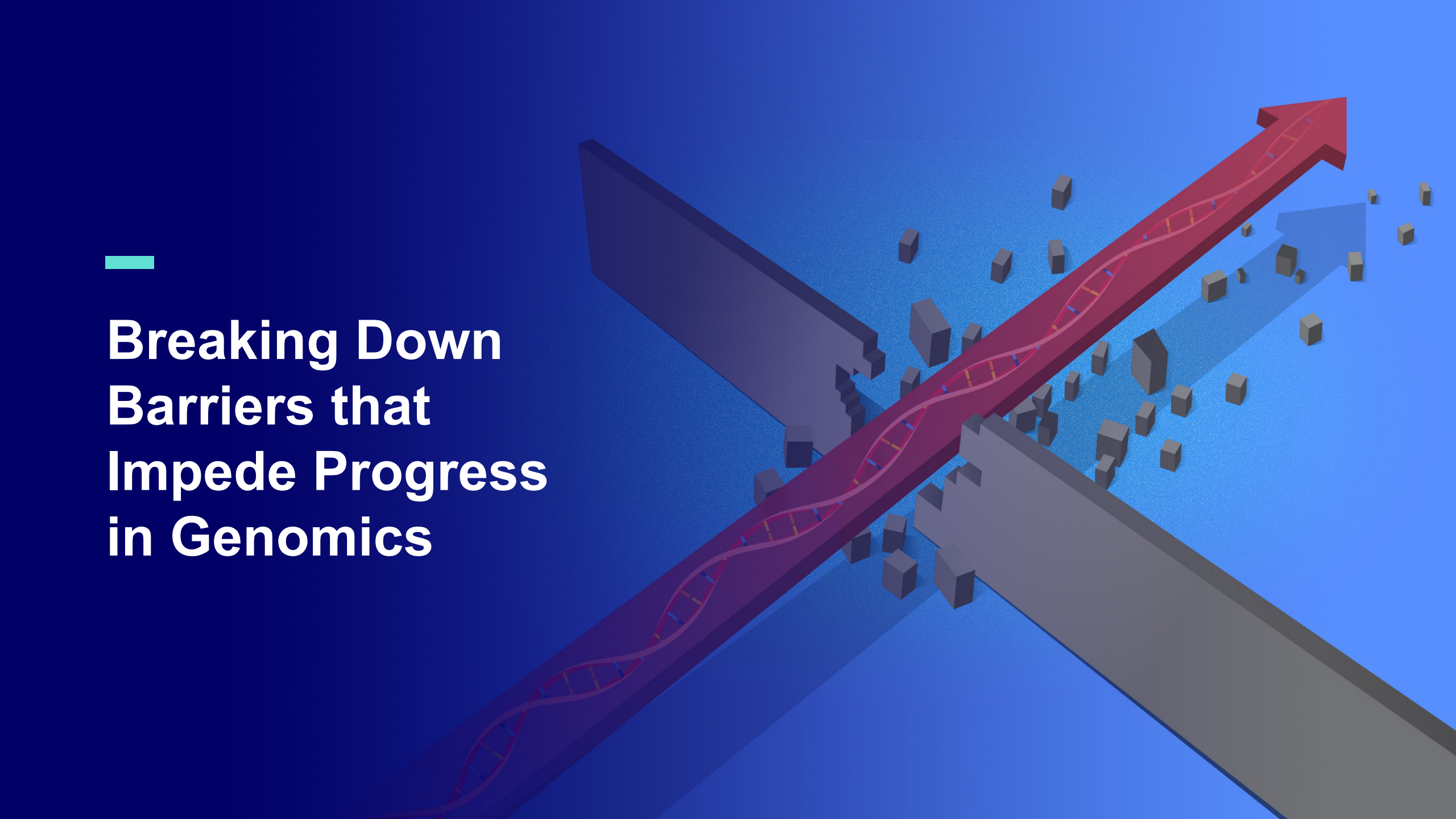


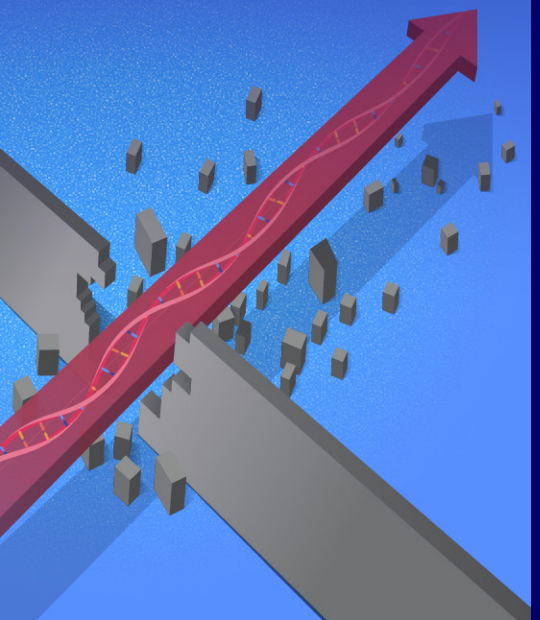
- Genomic data science (data generation, management, resources, analyses, etc.)
- Comparative genomics
- Empowering people to be stewards of their genomic information
- Genomic literacy
- Training healthcare providers in genomics

Breaking Down Barriers that Impede Progress in Genomics



Breaking Down Barriers that Impede Progress in Genomics

- DNA synthesis and editing
- Characterizing genomic variants
- Understanding genomic mosaicism
- Implementation science

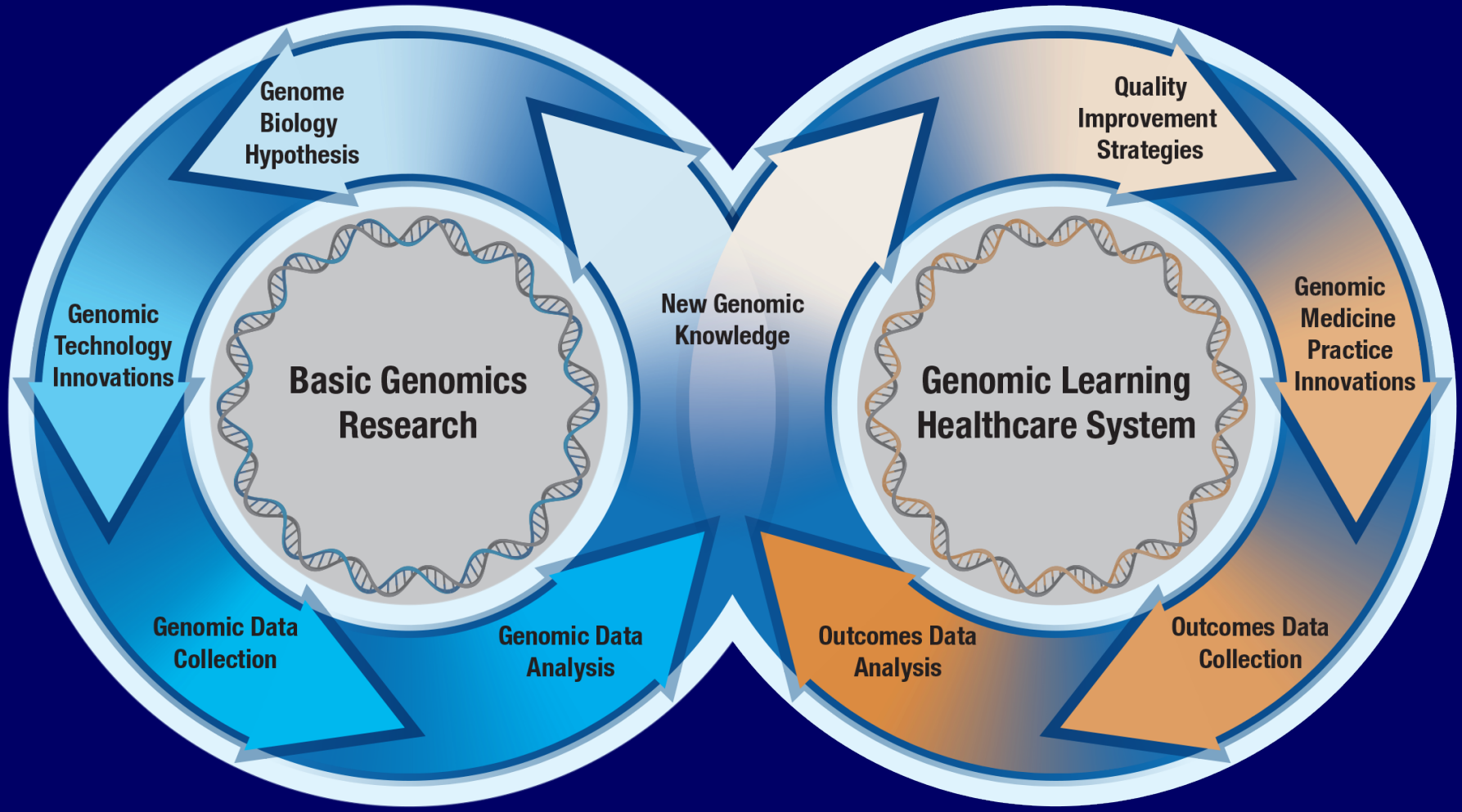


Compelling Genomics Research Projects in Biomedicine

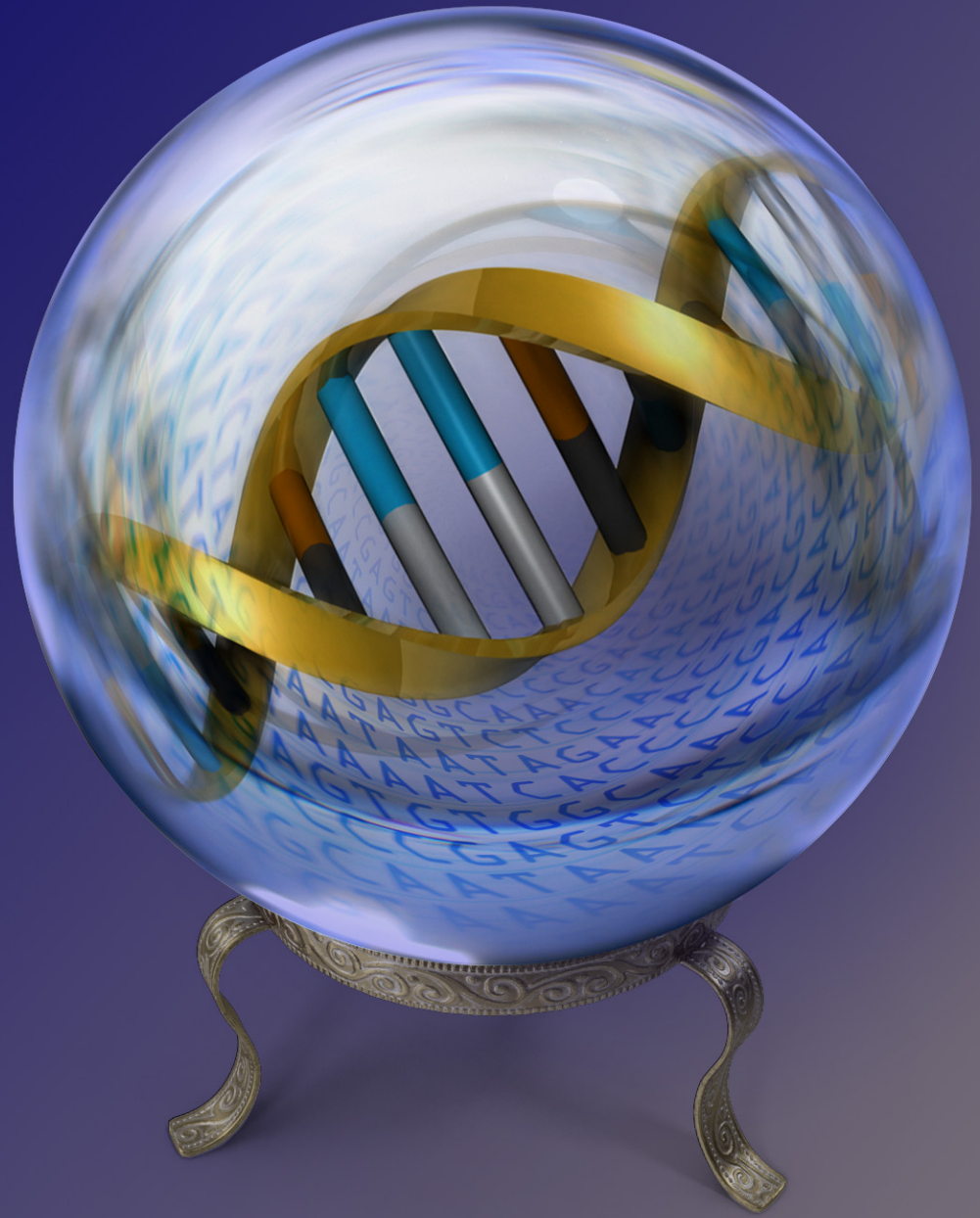
Compelling Genomics Research Projects in Biomedicine

- Comprehensive views of genes and regulatory elements
- Genetic architecture of human diseases and traits
- Enhancing diversity in genomics research
- Multi-omic studies in clinical settings
- Genomic learning healthcare systems

Virtuous Cycles in Human Genomics Research and Clinical Care



Bold Predictions for Human Genomics by 2030



Perspective

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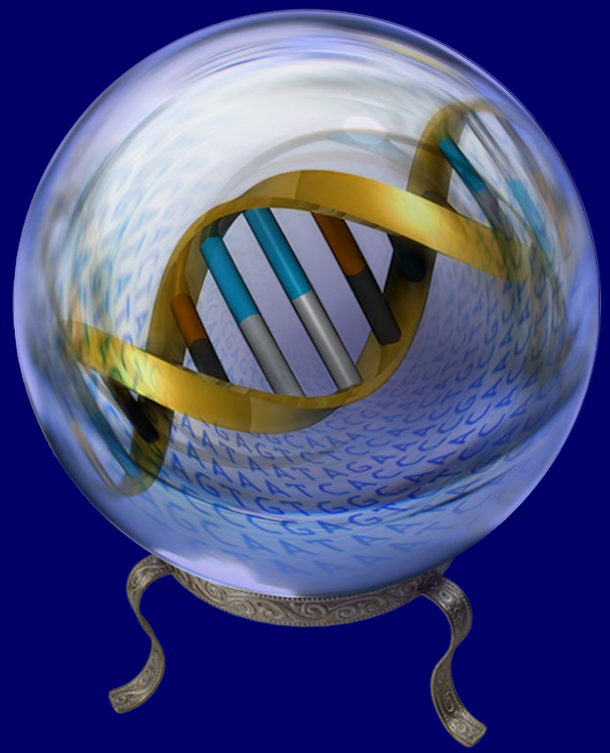
[1]National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA. ✉e-mail: egreen@nhgri.nih.gov

Box 5

Bold predictions for human genomics by 2030

Some of the most impressive genomics achievements, when viewed in retrospect, could hardly have been imagined ten years earlier. Here are ten bold predictions for human genomics that might come true by 2030. Although most are unlikely to be fully attained, achieving one or more of these would require individuals to strive for something that currently seems out of reach. These predictions were crafted to be both inspirational and aspirational in nature, provoking discussions about what might be possible at The Forefront of Genomics in the coming decade.

1. Generating and analysing a complete human genome sequence will be routine for any research laboratory, becoming as straightforward as carrying out a DNA purification.
2. The biological function(s) of every human gene will be known; for non-coding elements in the human genome, such knowledge will be the rule rather than the exception.
3. The general features of the epigenetic landscape and transcriptional output will be routinely incorporated into predictive models of the effect of genotype on phenotype.
4. Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.
5. Studies that involve analyses of genome sequences and associated phenotypic information for millions of human participants will be regularly featured at school science fairs.
6. The regular use of genomic information will have transitioned from boutique to mainstream in all clinical settings, making genomic testing as routine as complete blood counts.
7. The clinical relevance of all encountered genomic variants will be readily predictable, rendering the diagnostic designation 'variant of uncertain significance (VUS)' obsolete.
8. An individual's complete genome sequence along with informative annotations will, if desired, be securely and readily accessible on their smartphone.
9. Individuals from ancestrally diverse backgrounds will benefit equitably from advances in human genomics.
10. Breakthrough discoveries will lead to curative therapies involving genomic modifications for dozens of genetic diseases.



SCIENTIFIC AMERICAN®

BIOLOGY

A Vision for the Next Decade of Human Genomics Research

An article in *Nature* lays out 10 bold predictions for a field whose extraordinary achievements are just the beginning of what could be possible

By Eric D. Green on October 28, 2020

www.scientificamerican.com/article/a-vision-for-the-next-decade-of-human-genomics-research/

2020 NHGRI Strategic Vision Website

genome.gov/2020sv

What the 2020 NHGRI Strategic Vision IS

- A broad vision for human genomics with an emphasis on health applications
- A reflection of *The Forefront of Genomics* – i.e., areas most appropriate for NHGRI's vision, leadership, and responsible stewardship
- A paper intended to illuminate, illustrate, and inspire

What the 2020 NHGRI Strategic Vision IS NOT

- A vision for all of genomics
- An NHGRI-only (or even an NIH-only) vision
- A 5-year vision
- An implementation plan

From Strategic Planning to Vision Implementation

Ideas

Needs

Proposals

Obstacles

Perspective

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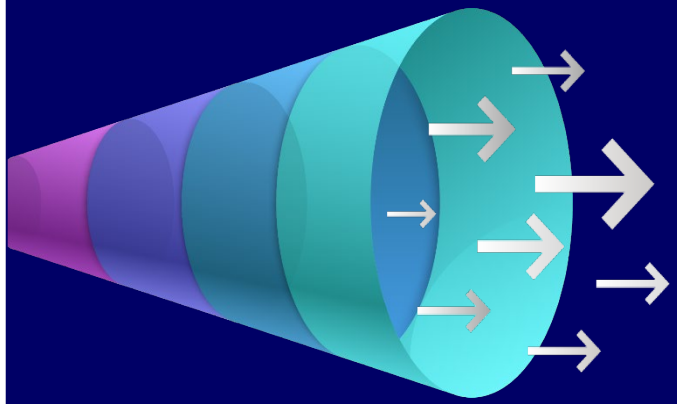


Projects

Programs

Initiatives

Policies

Strategic Planning Process

Feb 2018 to Sept 2020

2020 NHGRI Strategic Vision

October 2020

Implementation of Strategic Vision

2020 and Beyond

Guiding Principles and Values

Box 1

• **Champion a diverse genomics workforce** — the promise of genomics cannot be fully achieved without attracting, developing, and retaining a diverse workforce, which includes individuals from groups that are currently underrepresented in the genomics enterprise.

and underrepresented individuals in major genomic studies — attention to diversity in genomics research is both socially just and scientifically essential, which includes meaningful, sustained partnerships with diverse communities in the design and implementation of research studies, the propagation of research findings, and the development and use of new technologies.

• **Maximize the usability of genomics for all members of the public, including the ability to access genomics in healthcare** — engagement, inclusion, and understanding the needs of diverse and medically underserved groups are required to ensure that all members of society benefit equitably from genomic advances, with particular attention given to the equitable use of genomics in healthcare that avoids exacerbating and strives towards reducing health disparities.

• **Champion a diverse genomics workforce** — the promise of genomics cannot be fully achieved without attracting, developing, and retaining a diverse workforce, which includes individuals from groups that are currently underrepresented in the genomics enterprise.

• **Provide a conceptual research framing that consistently examines the role of both genomic and non-genomic contributors to health and disease** — routinely considering the

— the use of carefully defined standards (for example, those for generating, analysing, storing, and sharing data) has benefited genomics in numerous ways, and this must include appropriate privacy and data-security protections for those participating in genomics research.

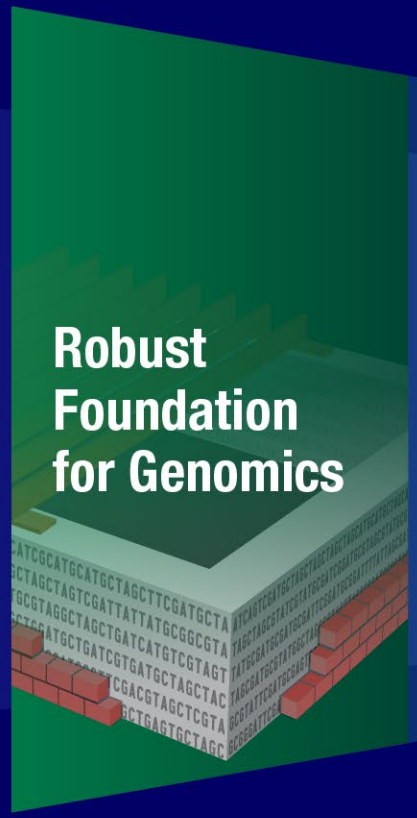
• **Embrace the interdisciplinary and team-oriented nature of genomics research** — starting with the Human Genome Project, some of the most challenging genomics endeavours have benefited from the creation and management of large, interdisciplinary research collaborations.

• **Adhere to the highest expectations and requirements related to open science, responsible data sharing, and rigor and reproducibility in genomics research** — the genomics enterprise has a well-respected history of leading in these areas, and that commitment must be built upon and continually reaffirmed.

• **Pursue advances in genomics as part of a vibrant global community of genomics researchers and funders** — the challenges in genomics require the collective energies and creativity of a collaborative international ecosystem that includes partnerships among researchers, funders, and other stakeholders from academia, government, and the commercial sector.

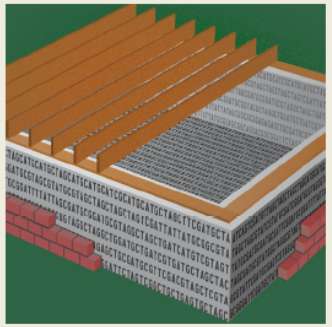
Guiding Principles and Values

Robust Foundation for Genomics



Box 2

Sustaining and improving a robust foundation for genomics



Genome structure and function

- Enable the routine generation and analysis of increasingly complex genomic data
- Use evolutionary and comparative genomic data to maximize understanding of genome function

Genomic data science

- Develop new methods and build sustainable data resources for genomics research
- Ensure facile storing, sharing, and computing on large genomic datasets
- Develop integrated knowledgebases and informatics methods for genomic medicine

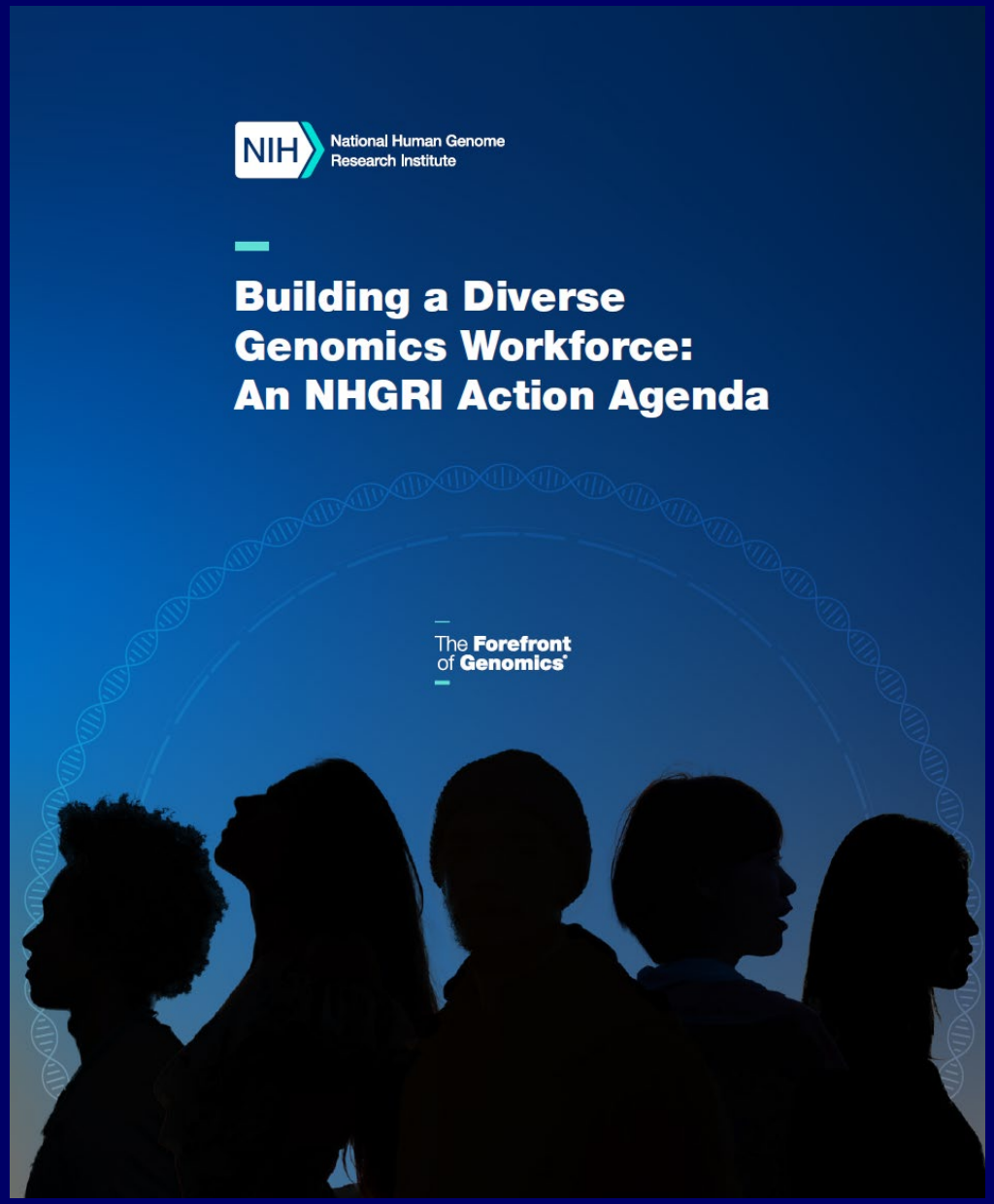
Genomics and society

- Understand the interrelationships between genomics and the social and environmental factors that influence human health
- Empower people to make well-informed decisions about genomic data and develop data-stewardship systems that reinforce their choices
- Increase the genomic literacy of all sectors of society

Training and genomics workforce development

- Ensure that the next generation of genomic scientists are sufficiently trained in data science
- Train healthcare providers to integrate genomics into the clinical workflow
- Foster a diverse genomics workforce

In both research and clinical settings, the global genomics workforce—as with the general biomedical research workforce—falls considerably short of reflecting the diversity of the world's population (a vivid example of this is seen in the United States[72]), which limits the opportunity of those systematically excluded to bring their unique ideas to scientific and clinical research[73]. To attain a diverse genomics workforce, new strategies and programs to reduce impediments to career opportunities in genomics are required, as are creative approaches to promote workforce diversity, leadership in the field, and inclusion practices. Efforts must intentionally include women, underrepresented racial and ethnic groups, disadvantaged populations, and individuals with disabilities. Initiatives should not focus exclusively on early-stage recruitment; instead, they must also include incentives to recruit and retain a diverse workforce at all career stages[74] as well as new approaches for cultivating the next generation of genomics practitioners.

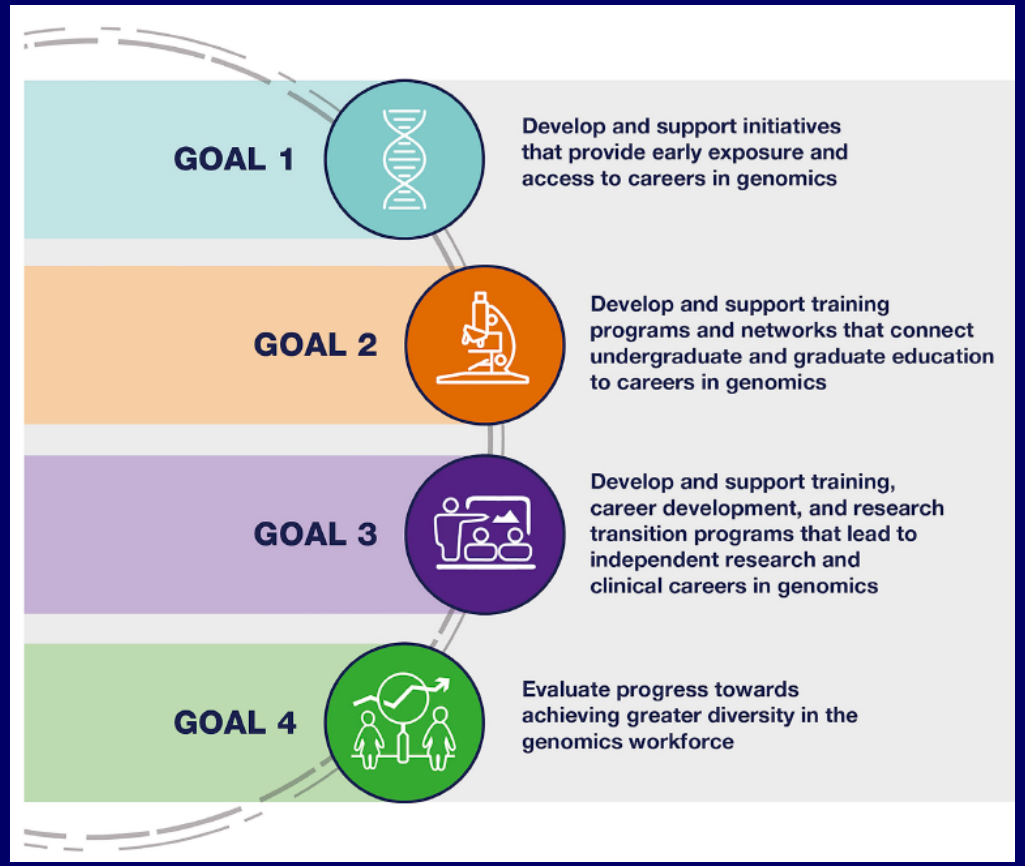


COMMENTARY

The genomics workforce must become more diverse: a strategic imperative

Vence L. Bonham[1,*] and Eric D. Green[1,*]

AJHG (2021)



genome.gov/workforcediversity

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

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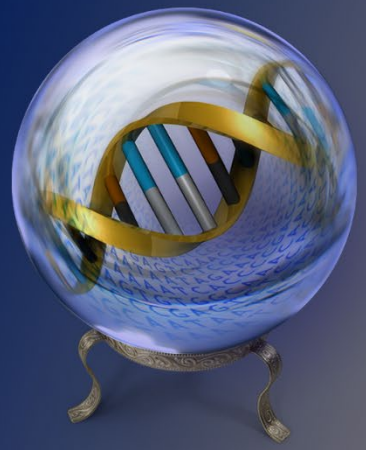
Bold Predictions for Human Genomics by 2030: An NHGRI Seminar Series



Bold Predictions for Human Genomics by 2030

A National Human Genome Research Institute (NHGRI) Seminar Series

Bold Prediction #1
February 1, 2021; 3 p.m. - 4:30 p.m. ET

Generating and analyzing a complete human genome sequence will be routine for any research laboratory, becoming as straightforward as carrying out a DNA purification.

Evan Eichler, Ph.D., University of Washington
Karen Miga, Ph.D., University of California, Santa Cruz
Moderator: **Eric Green, M.D., Ph.D.**, NHGRI

Bold Prediction #2
March 8, 2021; 3 p.m. - 4:30 p.m. ET

The biological function(s) of every human gene will be known; for non-coding elements in the human genome, such knowledge will be the rule rather than the exception.

Nancy Cox, Ph.D., Vanderbilt University
Neville Sanjana, Ph.D., New York Genome Center
Moderator: **Carolyn Hutter, Ph.D.**, NHGRI

Bold Prediction #3
April 12, 2021; 3 p.m. - 4:30 p.m. ET

The general features of the epigenetic landscape and transcriptional output will be routinely incorporated into predictive models of the impact of genotype on phenotype.

Tom Gingeras, Ph.D., Cold Spring Harbor Laboratory
Tuuli Lappalainen, Ph.D., New York Genome Center
Moderator: **Paul Liu, M.D., Ph.D.**, NHGRI

Bold Prediction #4
May 25, 2021; 3 p.m. - 4:30 p.m. ET

Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.

Charmaine Royal, Ph.D., Duke University
Genevieve Wojcik, Ph.D., Johns Hopkins University
Moderator: **Vence Bonham, Jr., J.D.**, NHGRI

Bold Prediction #5
June 7, 2021; 3 p.m. - 4:30 p.m. ET

Studies involving analyses of genome sequences and associated phenotypic information for millions of human participants will be regularly featured at school science fairs.

Neil Lamb, Ph.D., HudsonAlpha Institute for Biotechnology
Chanda Jefferson, Albert Einstein Distinguished Educator Fellow; Office of Mark DeSaulnier, California's 11th Congressional District
Moderator: **Larry Brody, Ph.D.**, NHGRI

The Bold Predictions are part of the 2020 NHGRI "Strategic Vision for Improving Human Health at The Forefront of Genomics." For more information, visit: https://www.genome.gov/2020SV.

Dates and speakers for sessions 6-10 will be announced in April 2021.

NIH NHGRI

See our bold predictions at: genome.gov/bold-predictions

The Forefront of Genomics

Series Webpage:

genome.gov/bold-predictions

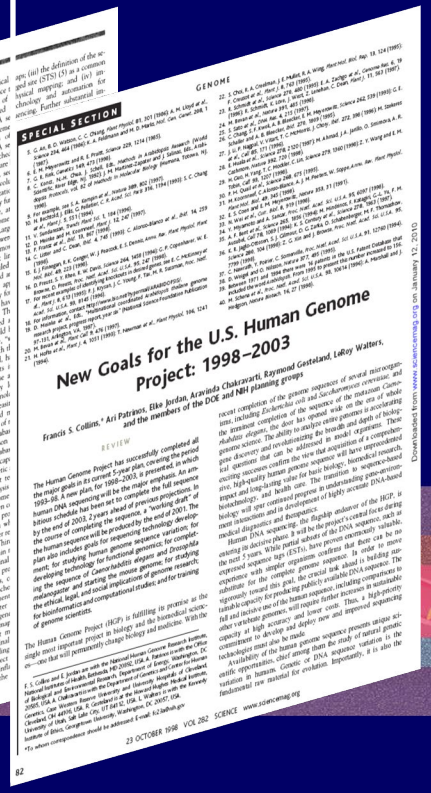
Latest Chapter for Human Genomics

1991-1995 | 1993-1998 | 1998-2003 | 2003-2010 | 2011-2020 | 2020-???


POLICY FORUM

A New Five-Year Plan for the U.S. Human Genome Project

Francis Collins and David Galas*

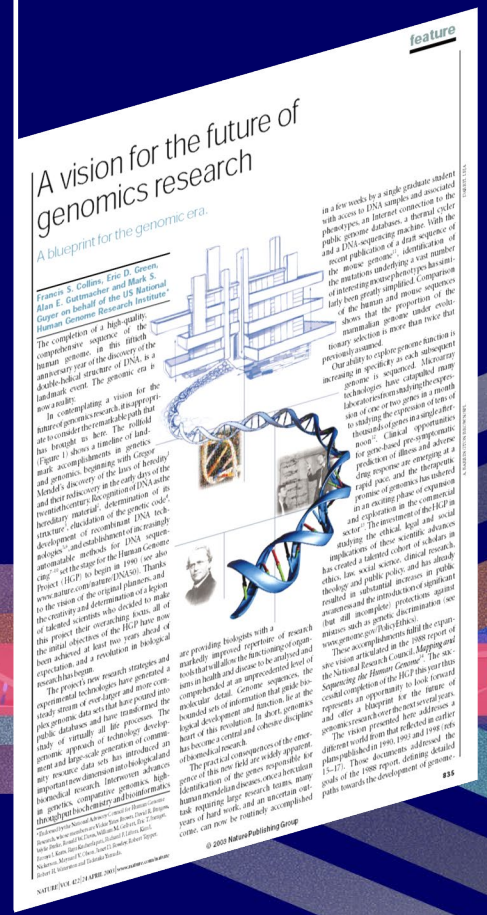

SPECIAL SECTION

GENOME

New Goals for the U.S. Human Genome Project: 1998–2003

Francis S. Collins,* Ari Patrinos, Elke Jordan, Aravinda Chakravarti, Raymond Gesteland, LeRoy Walters, and the members of the DOE and NIH planning groups

REVIEW


feature

A vision for the future of genomics research

A blueprint for the genomic era.

Francis S. Collins, Eric D. Green, Alan E. Guttmacher and Mark S. Guyer on behalf of the National Human Genome Research Institute*


PERSPECTIVE

Charting a course for genomic medicine from base pairs to bedside

Eric D. Green[1] & Mark S. Guyer[2] & National Human Genome Research Institute*


Perspective

Strategic vision for improving human health at The Forefront of Genomics

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Check for updates

Eric D. Green[1,2*], Chris Gunter[3], Leslie D. Biesecker[4], Valentina Di Francesco[5], Carla L. Easter[6], Elise A. Feingold[7], Adam M. Felsenfeld[8], David J. Kaufman[9], Elaine A. Ostrander[1], William J. Pavan[10], Adam L. Phillippy[11], Anastasia L. Wise[1], Jyoti Gupta Dayal[1], Britny J. Kish[1], Allison Mandich[1], Christopher R. Wellington[1], Mark S. Watson[12], Bettie J. Graham[13], Daniel L. Koeller[14], Darryl Leja[1], Susan Vasquez[1], William A. Gahl[1], Bettie J. Graham[13], Daniel L. Koeller[14], Laura Lyman Rodriguez[1], Benjamin D. Solomon[1], Vence L. Bonham[1], Lawrence C. Brody[1], Carolyn M. Hutter[1] & Teri A. Manolio[1]

Starting with the launch of the Human Genome Project three decades ago, and continuing after its completion in 2003, genomics has progressively come to have a central and catalytic role in basic and translational research. In addition, studies increasingly demonstrate how genomic information can be effectively used in clinical care. In the future, the anticipated advances in technology development, biological insights, and clinical applications (among others) will lead to more widespread integration of genomics into almost all areas of biomedical research, the adoption of genomics into mainstream medical and public health practices, and an increasing relevance of genomics for everyday life. On behalf of the research community, the National Human Genome Research Institute recently completed a multi-year process of strategic engagement to identify future research priorities and opportunities in human genomics, with an emphasis on health applications. Here we describe the highest-priority elements envisioned for the cutting-edge of human genomics going forward—that is, at 'The Forefront of Genomics'.

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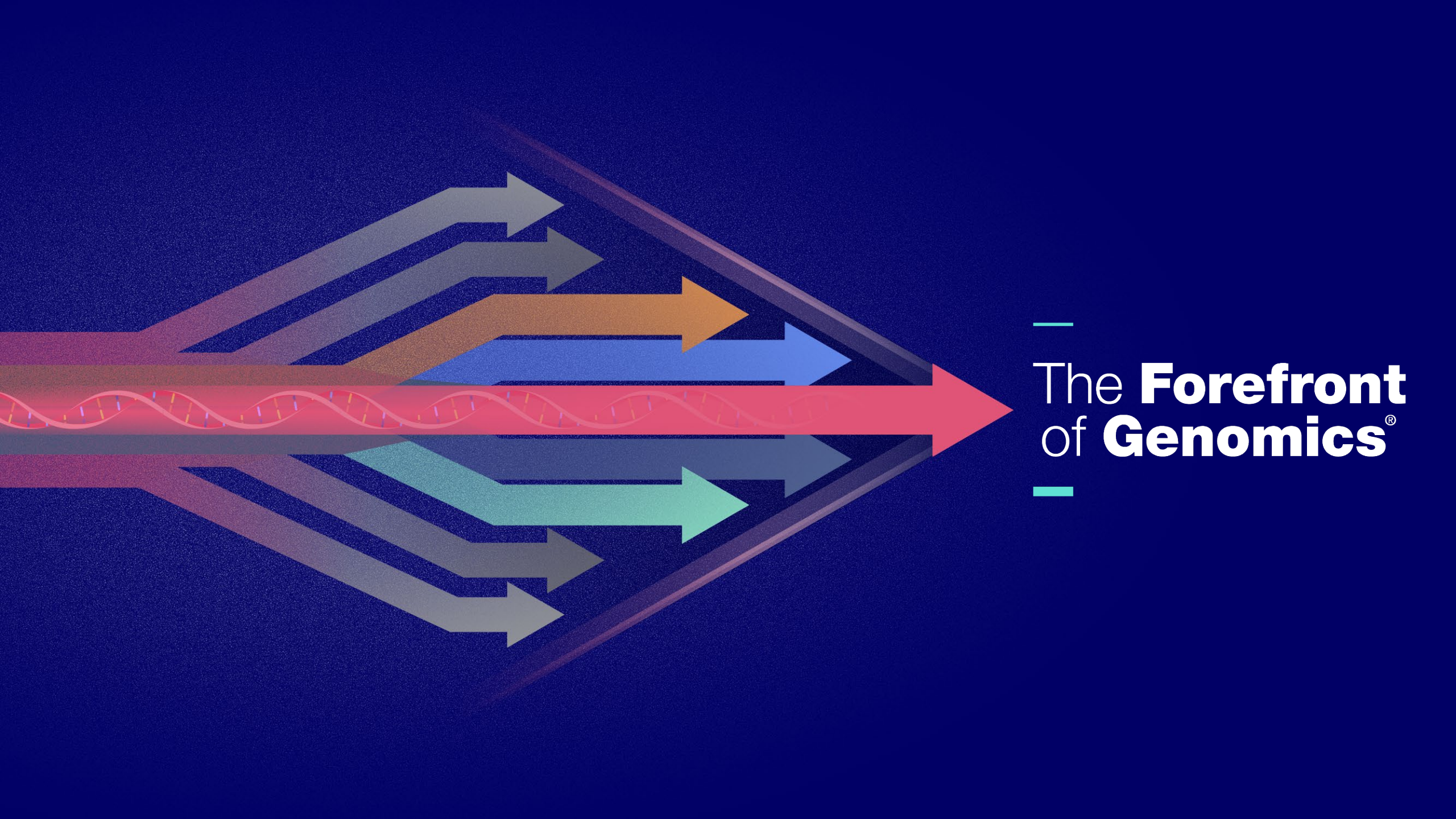
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