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# Building a Diverse Genomics Workforce: An NHGRI Action Agenda

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National Human Genome  
Research Institute

—  
The **Forefront**  
of **Genomics**  
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# 2020 NHGRI Strategic Vision



## Perspective

### Strategic vision for improving human health at The Forefront of Genomics

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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’.

Beginning in October 1990, a pioneering group of international researchers began an audacious journey to generate the first map and sequence of the human genome, marking the start of a 13-year odyssey called the Human Genome Project<sup>1–3</sup>. The successful and early completion of the Project in 2003, which included parallel studies of a set of model organism genomes, catalysed enormous progress in genomics research. Leading the signature advances has been a greater than one million-fold reduction in the cost of DNA sequencing<sup>4</sup>. This decrease has allowed the generation of innumerable genome sequences, including hundreds of thousands of human genome sequences (both in research and clinical settings), and the continuous development of assays to identify and characterize functional genomic elements<sup>5,6</sup>. These new tools, together with increasingly sophisticated statistical and computational methods, have enabled researchers to create rich catalogues of human genomic variants<sup>7,8</sup> to gain an ever-deepening understanding of the functional complexities of the human genome<sup>9</sup>, and to determine the genomic bases of thousands of human diseases<sup>10,11</sup>. In turn, the past decade has brought the initial realization of genomic medicine<sup>12</sup>, as research successes have been converted into powerful tools for use in healthcare, including somatic genome analysis for cancer (enabling development of targeted therapeutic agents)<sup>13</sup>, non-invasive prenatal genetic screening<sup>14</sup>, and genomics-based tests for a growing set of paediatric conditions and rare disorders<sup>15</sup>, among others.

In essence, with growing insights about the structure and function of the human genome and ever-improving laboratory and computational technologies, genomics has become increasingly woven into the fabric

of biomedical research, medical practice, and society. The scope, scale, and pace of genomic advances so far were nearly unimaginable when the Human Genome Project began; even today, such advances are yielding scientific and clinical opportunities beyond our initial expectations, with many more anticipated in the next decade.

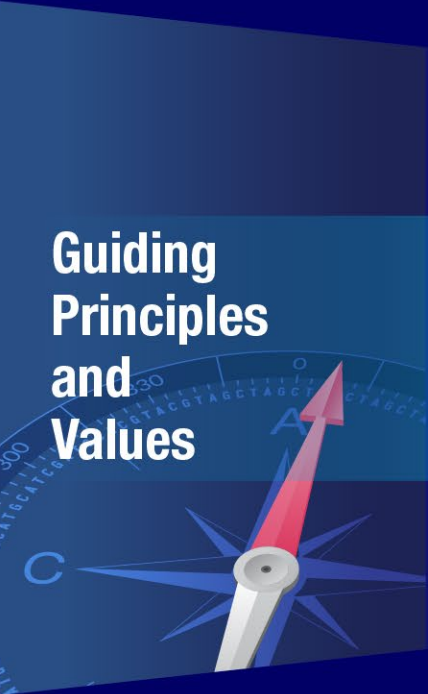
Embracing its leadership role in genomics, the National Human Genome Research Institute (NHGRI) has developed strategic visions for the field at key inflection points, in particular at the end of the Human Genome Project in 2003<sup>16</sup> and then again at the beginning of the last decade in 2011<sup>17</sup>. These visions outlined the most compelling opportunities for human genomics research, in each case informed by a multi-year engagement process. NHGRI endeavoured to start the new decade with an updated strategic vision for human genomics research. Through a planning process that involved more than 50 events (such as dedicated workshops, conference sessions, and webinars) over the past two years (see <http://genome.gov/genomics2020>), the institute collected input from a large number of stakeholders, with the resulting input catalogued and synthesized using the framework depicted in Fig. 1.

Unlike the past, this round of strategic planning was greatly influenced by the now widely disseminated nature of genomics across biomedicine. A representative glimpse into this historic phenomenon is illustrated in Fig. 2. During the Human Genome Project, NHGRI was the primary funder of human genomics research at the US National Institutes of Health (NIH), but the past two decades have brought a greater than tenfold increase in the relative fraction of funding coming from other parts of the NIH.

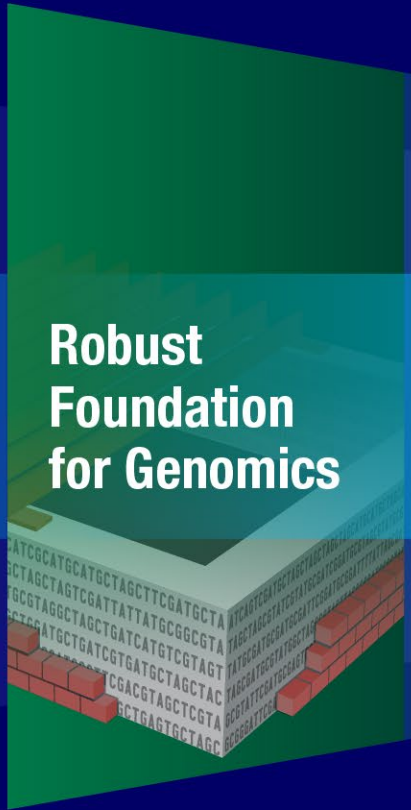
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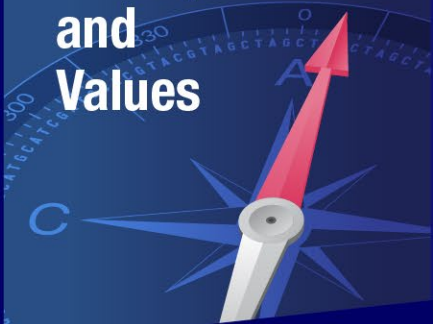


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# Guiding Principles and Values





## 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.

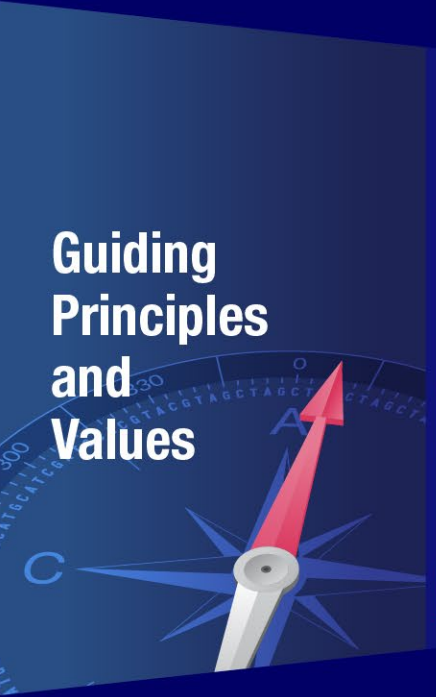
• **Provide a conceptual research framing that consistently examines the role of both genomic and non-genomic contributors to health and disease** — routinely considering the

— 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.

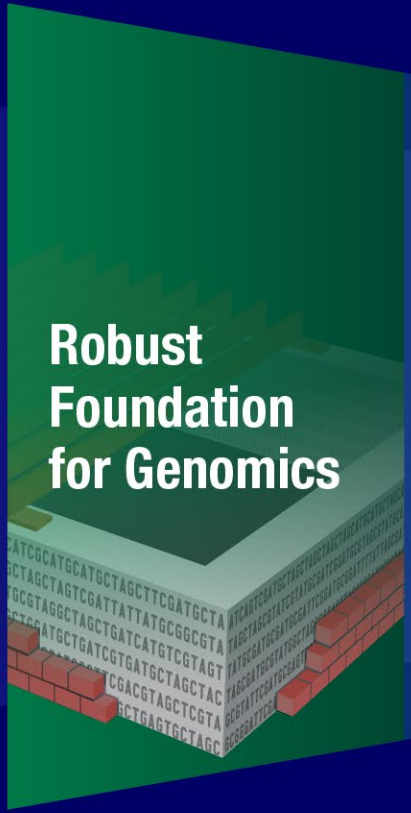
• **Embrace the interdisciplinary and team-oriented nature of genomics research** — starting with the Human Genome Project, some of the most challenging genomics endeavours have benefited from the creation and management of large, interdisciplinary research collaborations.

• **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



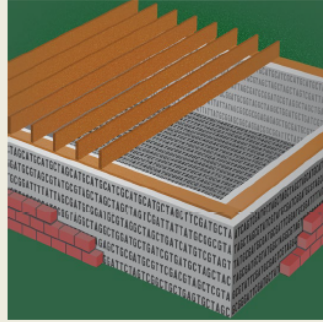
## Robust Foundation for Genomics

## 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<sup>72</sup>), which limits the opportunity of those systematically excluded to bring their unique ideas to scientific and clinical research<sup>73</sup>. 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<sup>74</sup> as well as new approaches for cultivating the next generation of genomics practitioners.



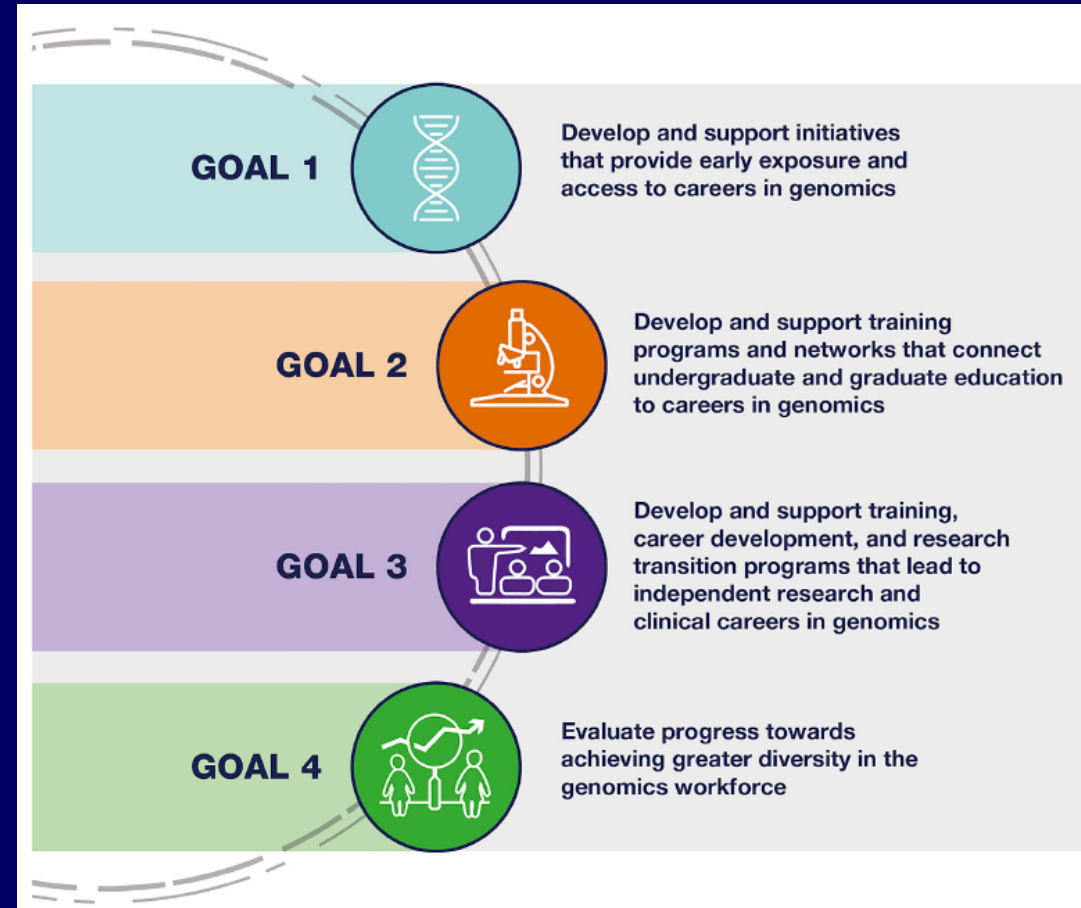
## Building a Diverse Genomics Workforce: An NHGRI Action Agenda

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### The genomics workforce must become more diverse: a strategic imperative

Vence L. Bonham<sup>1,\*</sup> and Eric D. Green<sup>1,\*</sup>

*Am J Hum Genet.* 2021 7;108(1):3-7.



[genome.gov/workforcediversity](https://genome.gov/workforcediversity)



## GOAL 1



Develop and support initiatives that provide early exposure and access to careers in genomics.

## GOAL 2



Develop and support training programs and networks that connect undergraduate and graduate education to careers in genomics.

## GOAL 3



Develop and support training, career development, and research transition programs that lead to independent research and clinical careers in genomics.

## GOAL 4



Evaluate progress towards achieving greater diversity in the genomics workforce.



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## Building a Diverse Genomics Workforce

NHGRI articulates goals and objectives to increase the number of individuals from diverse backgrounds in genomics.

[Read the Action Agenda](#)

[genome.gov/workforcediversity](https://genome.gov/workforcediversity)