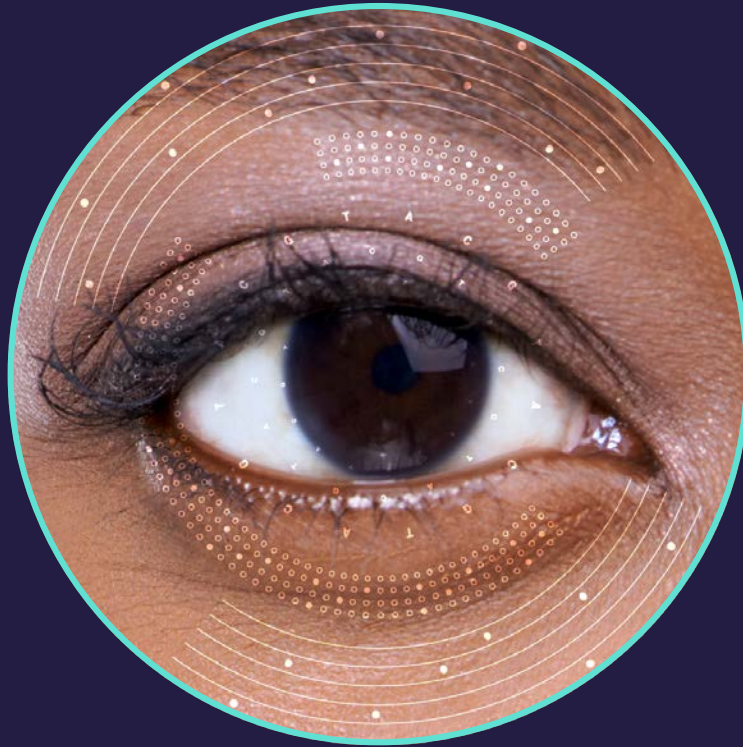


# En Route to a “2020 Vision for Genomics”



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



National Human Genome  
Research Institute

# Official Launch: February 12, 2018



U.S. DEPARTMENT OF HEALTH & HUMAN SERVICES  
NATIONAL INSTITUTES OF HEALTH

National Human Genome Research Institute  
[www.genome.gov](http://www.genome.gov)

## **FOR IMMEDIATE RELEASE**

Monday, February 12, 2018  
11 A.M. Eastern

**Contact:**  
NHGRI Communications  
(301) 402-0911  
[NHGRIPress@mail.nih.gov](mailto:NHGRIPress@mail.nih.gov)

### **Developing a 2020 vision for genomics: NHGRI launches new round of strategic planning**

The National Human Genome Research Institute (NHGRI) today launched a new round of strategic planning that will establish a 2020 vision for genomics research aimed at accelerating scientific and medical breakthroughs. In developing the strategic plan, the institute will engage experts and diverse public communities to identify paradigm-shifting areas of genomics that will expand the field into new frontiers and enable novel applications to human health and disease.

# Video of Announcement Available at...

<https://youtu.be/cCazVxD2isl>





# The **Forefront** of **Genomics**<sup>®</sup>



Understanding  
Our Genetic  
Inheritance

The U.S.  
Human Genome  
Project:

The First  
Five Years  
FY 1991-1995

U.S. DEPARTMENT OF  
HEALTH AND HUMAN SERVICES  
Public Health Service  
National Institutes of Health

U.S. DEPARTMENT OF ENERGY  
Office of Energy Research  
Office of Health and  
Environmental Research

## POLICY FORUM

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

Francis Collins and David Galas\*

The U.S. Human Genome Project is an international effort to map and sequence the human genome. The project has made significant progress in the past five years, and we are now entering a new phase of the project. Thanks to advances in technology, we are able to track with respect to the genome. Because 3 years have gone by, and the goals were set, and the sophisticated and detailed work needs to be done now available, the goal and extended to cover (through September 1995) the genome initiative.

In 1990, the Human Genome Project and the Department of Energy developed a joint research plan for the first 5 years (1991-95) of the U.S. Human Genome Project (1). It has a guide for both the research and the agencies' administrative and assessing its progress. Great strides have been made in the achievement of the project, particularly with respect to the physical maps of the human genome and the genomes of certain model organisms. The project has also developed improved sequencing and information technologies, and social issues associated with the project, and defining the most urgent and important issues in the field.

Progress toward achieving the goals of the project has been on schedule or, in some cases, ahead of schedule. Biological improvements have been anticipated in 1995, and the project has changed the scope of the project. The project has lowered more ambitious goals for the year, it was therefore, and extend the initial scope of genome research.

F. Collins is the director of the Human Genome Research Institute, Bethesda, MD 20892. D. Galas was associate director of the Environmental Research Laboratory, Washington, DC 20585.

\* Present address: Daresbury Laboratory, Warrington, WA 96033.

## SPECIAL SECTION

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## A vision for the future of genomics research

A blueprint for

## PERSPECTIVE

doi:10.1038/nature09764

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

### Charting a course for genomic medicine from base pairs to bedside

Eric D. Green<sup>1</sup>, Mark S. Guyer<sup>1</sup> & National Human Genome Research Institute\*

There has been much progress in genomics in the ten years since a draft sequence of the human genome was published. Opportunities for understanding health and disease are now unprecedented, as advances in genomics are harnessed to obtain robust foundational knowledge about the structure and function of the human genome and about the genetic contributions to human health and disease. Here we articulate a 2011 vision for the future of genomics research and describe the path towards an era of genomic medicine.

Since the end of the Human Genome Project (HGP) in 2003 and the publication of a reference human genome sequence<sup>1,2</sup>, genomics has become a mainstay of biomedical research. The scientific community's foresight in launching this ambitious project<sup>3</sup> is evident in the broad range of scientific advances that the HGP has enabled, as shown in Fig. 1 (see ref. 4). Optimism about the potential contributions of genomics to improving human health has been fuelled by new insights about cancer<sup>5,6</sup>, the molecular basis of inherited diseases (http://www.ncbi.nlm.nih.gov/omim and http://www.genome.gov/GWAStudies) and the role of structural variation in disease<sup>7</sup>, some of which have already led to new therapies<sup>8,9</sup>. Other advances have already changed medical practice (for example, microarrays are now used for clinical detection of genomic imbalances<sup>10</sup>) and pharmacogenomic testing is routinely performed before administration of certain medications<sup>11</sup>). Together, these achievements (see accompanying paper<sup>12</sup>) document that genomics is contributing to a better understanding of human biology and to improving human health.

As it did eight years ago<sup>3</sup>, the National Human Genome Research Institute (NHGRI) has engaged the scientific community (http://www.genome.gov/Planning) to reflect on the key attributes of genomics (Box 1) and explore future directions and challenges for the field. These discussions have led to an update vision that focuses on understanding human biology and the diagnosis, prevention and treatment of human disease, including consideration of the implications of those advances for society (but these discussions, intentionally did not address the role of genomics in agriculture, energy and other areas). Like the HGP, achieving this vision is broader than what any single organization or country can achieve—realizing the full benefits of genomics will be a global effort.

This 2011 vision for genomics is organized around five domains extending from basic research to health applications (Fig. 2). It reflects the view that, over time, the most effective way to improve human health is to understand normal biology (in this case, genome biology) as a basis for understanding disease biology, which then becomes the basis for improving health. At the same time, there are other connections among these domains. Genomics offers opportunities for improving health without a thorough understanding of disease (for example, cancer therapies can be selected based on genomic profiles that identify tumour subtypes<sup>13,14</sup>), and clinical discoveries can lead back to understanding disease or even basic biology. The past decade has seen genomics contribute fundamental knowledge about biology and its perturbation in disease. Further deepening this understanding will accelerate the transition to genomic medicine (clinical care based on genomic information). But significant change rarely comes

quickly. Although genomics has already begun to improve diagnostics and treatments in a few circumstances, profound improvements in the effectiveness of healthcare cannot realistically be expected for many years (Fig. 2). Achieving such progress will depend not only on research, but also on new policies, practices and other developments. We have illustrated the kinds of achievements that can be anticipated with a few examples (Box 2) where a confluence of need and opportunities should lead to major accomplishments in genomic medicine in the coming decade. Similarly, we note three cross-cutting areas that are broadly relevant and fundamental across the entire spectrum of genomics and genomic medicine: bioinformatics and computational biology (Box 3), education and training (Box 4), and genomics and society (Box 5).

#### Understanding the biology of genomes

Substantial progress in understanding the structure of genomes has revealed much about the complexity of genome biology. Continued acquisition of basic knowledge about genome structure and function will be needed to illuminate further those complexities (Fig. 2). The contribution of genomics will include more comprehensive sets (catalogues) of data and new research tools, which will enhance the capabilities of all researchers to reveal fundamental principles of biology.

#### Comprehensive catalogues of genomic data

Comprehensive genomic catalogues have been uniquely valuable and widely used. There is a compelling need to improve existing catalogues and to generate new ones, such as complete collections of genetic variation, functional genomic elements, RNAs, proteins, and other biological molecules, for both human and model organisms.

Genomic studies of the genes and pathways associated with disease-related traits require comprehensive catalogues of genetic variation, which provide both genetic markers for association studies and variants for identifying candidate genes. Developing a detailed catalogue of variation in the human genome has been an international effort that began with The SNP Consortium<sup>15</sup> and the International HapMap Project<sup>16</sup> (http://hapmap.ncbi.nlm.nih.gov), and is ongoing with the 1000 Genomes Project<sup>17</sup> (http://www.1000genomes.org).

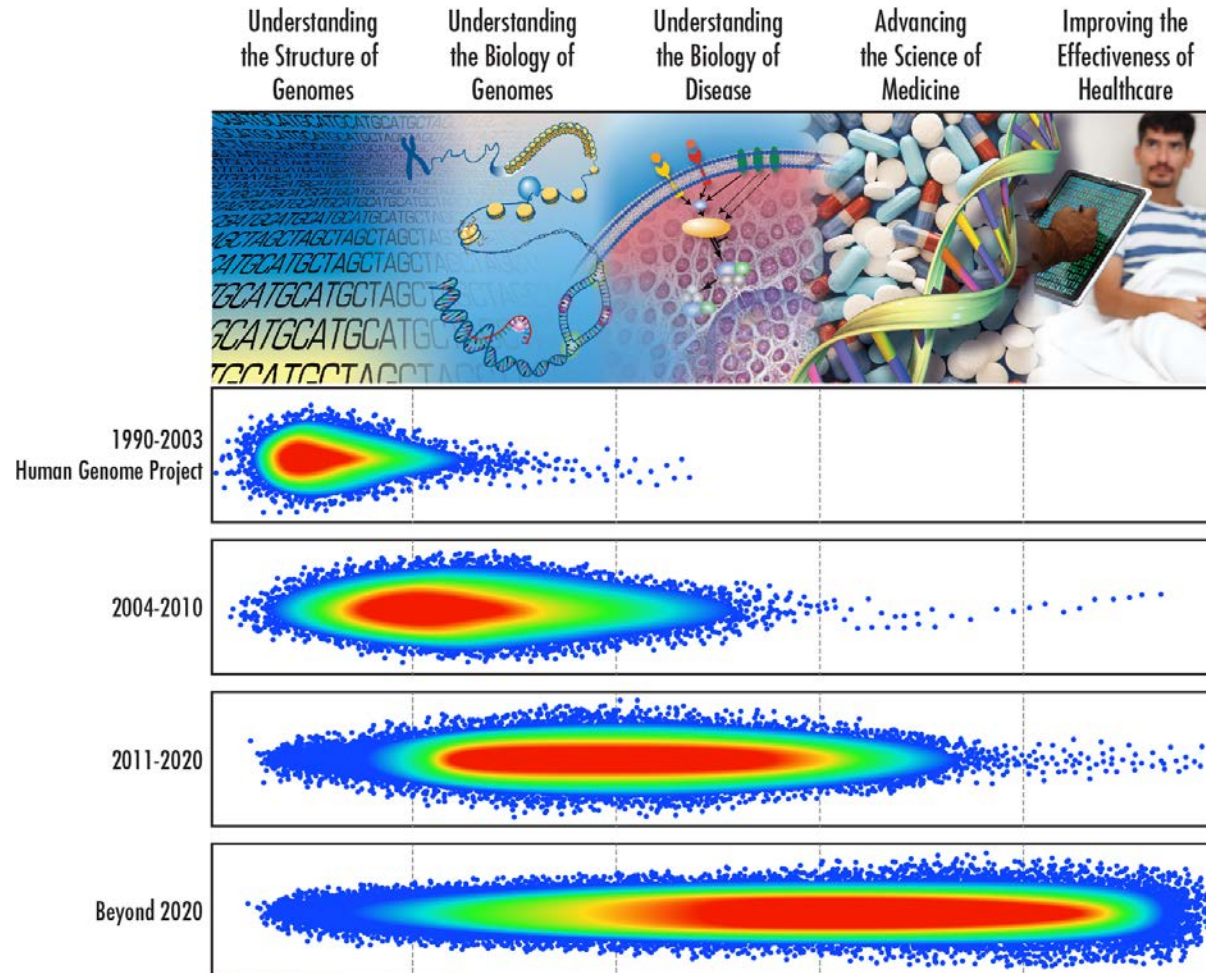
Over the past decade, these catalogues have been critical in the discovery of the specific genes for roughly 3,000 Mendelian (monogenic) diseases

Figure 1 | Genomic achievements since the Human Genome Project (see accompanying ref. 4). ▶

\*National Human Genome Research Institute, National Institutes of Health, 31 Center Dr., Bethesda, Maryland 20892-2152, USA.  
†List of participants and their affiliations appear at the end of the paper.

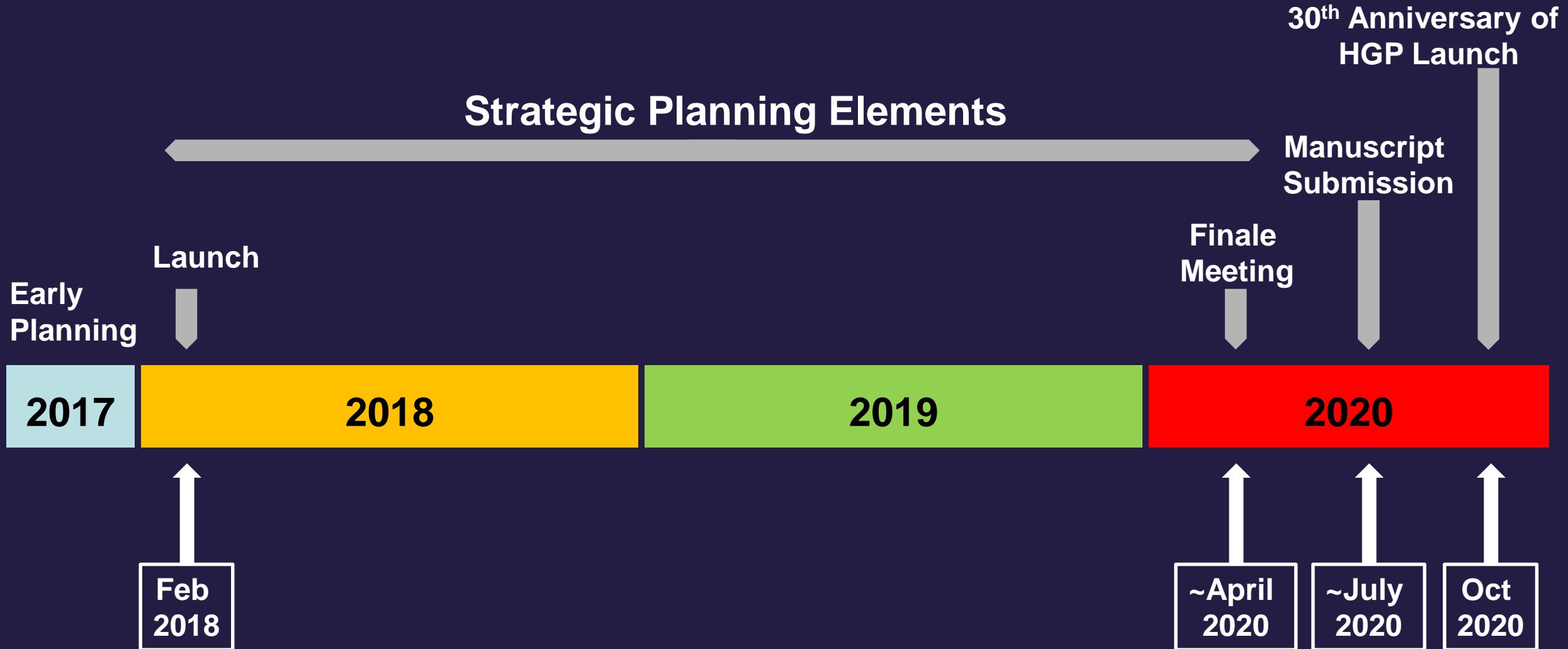
# 2011-Present

## *En Route to Genomic Medicine*



**It is hard to claim (or believe) that a  
2011 strategic plan represents a  
suitable blueprint for genomics or  
NHGRI in 2020 (and beyond)**

# “Genomics 2020” Strategic Planning Timeline

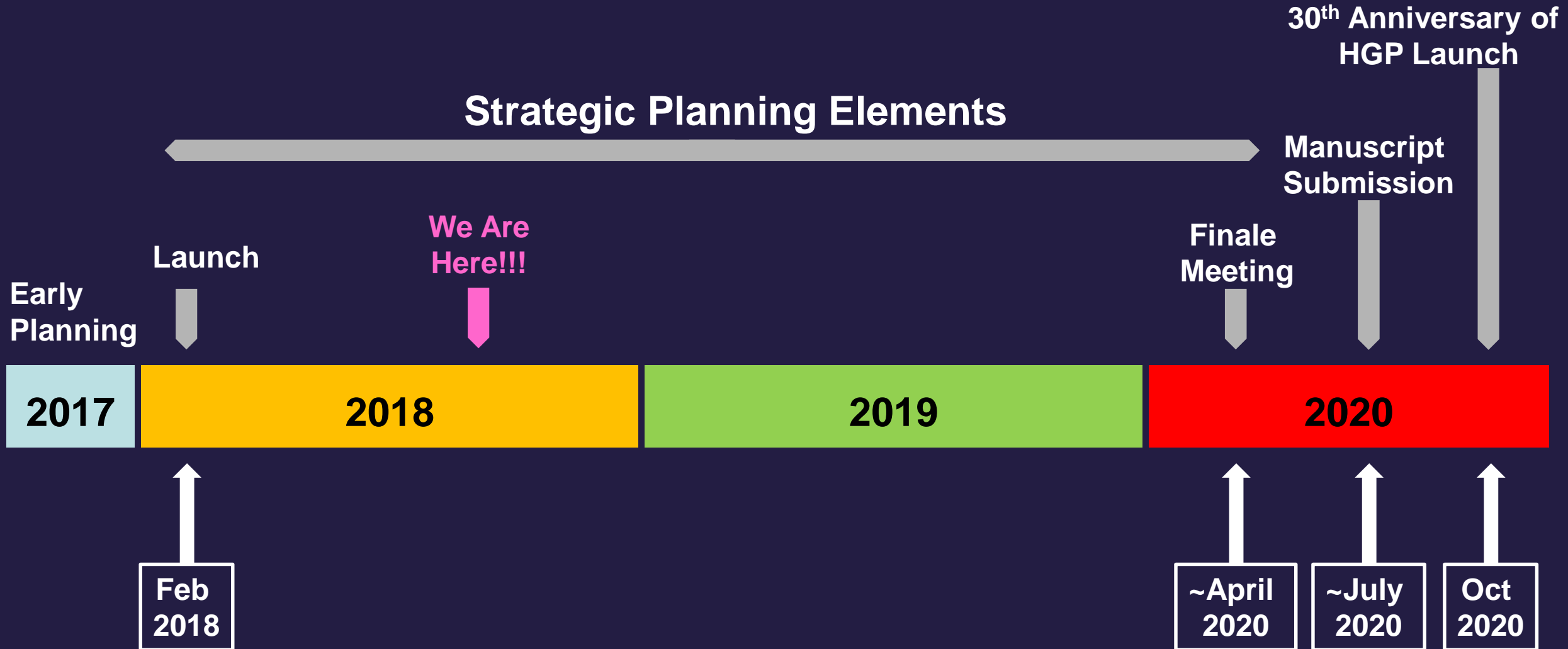


# “Genomics 2020” Strategic Planning Elements

- Workshops
- Town Halls
- Gatherings at Existing Meetings
- Dedicated Web Page
- Social Media Tools (e.g., #genomics2020)
- Engagement of Advisory Groups
- Finale Meeting



# “Genomics 2020” Strategic Planning Timeline



# **Overarching (and Difficult) Issue: Scope**

**Is the “Genomics 2020” strategic planning  
process about the future of genomics as a field**

**– OR –**

**the future of NHGRI-supported genomics research?**

# 2020 and Beyond

## *NHGRI-led Genomics Efforts Starting Next Decade*

Cancer Genomics

Microbial Genomics

### Prototypic Mainstream Areas:

Technology development  
Genome function and variation  
Epigenomics  
Genome-environment interactions  
Genomic medicine implementation  
Genomics workforce development  
Policy development/implementation  
ELSI of genomic advances

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Microbiome Research

Other???

# Overarching (and Difficult) Issue: Scope

Is the “Genomics 2020” strategic planning process about the future of genomics as a field

– OR –

the future of NHGRI-supported genomics research?

## Evolving Answer:

The “Genomics 2020” strategic planning process will focus on *The Forefront of Genomics* – as it pertains to human health and disease

# The 2020 NHGRI Strategic Plan will Aim to...

- Be the driving force for much of genomics at NIH and around the world
  - Provide a clear (i.e., 2020) vision for using genomics to advance human health
  - Guide NHGRI's scientific priorities and shape our research portfolio
  - Foster partnerships with research, healthcare, education, policy, and various general-public communities
- 
- Help to define NHGRI's position at:

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# Seeking Your Ideas



**Website:** [genome.gov/genomics2020](https://genome.gov/genomics2020)

**Email:** [genomics2020@mail.nih.gov](mailto:genomics2020@mail.nih.gov)

**Hashtag:** [#genomics2020](https://twitter.com/genomics2020)



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