







# NHGRI Strategic Planning 'Virtual' Town Hall

May 4, 2018





### Instructions for Participating in the Discussion

- To be placed in the queue to ask a question after the presentation, press \* (star) and 1
- State your name and organization
- To remove yourself from the queue, press the # (pound) key



### Welcome!





Director, Division of Policy, Communications, and Education



### Today's Speaker



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

**Director, National Human Genome Research Institute** 



### Also in the Room for Our Discussion



Leslie Biesecker, M.D. Division of Intramural Research



Carla Easter, Ph.D.

Division of Policy,

Communications and Education



Elise Feingold, Ph.D. Division of Genome Sciences



Adam Felsenfeld, Ph.D.

Division of Genome Sciences



Elaine Ostrander, Ph.D. Division of Intramural Research



**Bill Pavan, Ph.D.**Division of Intramural Research



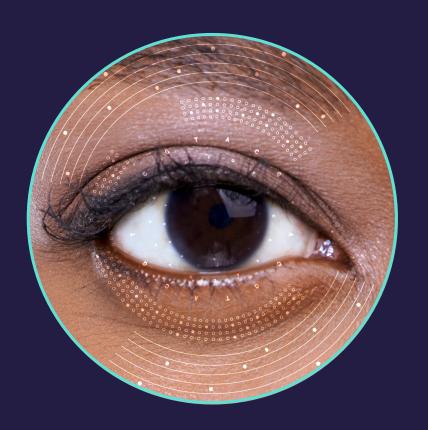
Adam Phillippy, Ph.D. Division of Intramural Research



Anastasia Wise, Ph.D. Division of Genomic Medicine



### En Route to a "2020 Vision for Genomics"



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

Director, NHGRI



### Official Launch: February 12, 2018



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

National Human Genome Research Institute www.genome.gov

#### FOR IMMEDIATE RELEASE

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

#### Contact:

NHGRI Communications (301) 402-0911 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/cCazVxD2isI



Understanding **Our Genetic** Inheritance

The U.S. Human Genor Project:

The First Five Years FY 1991-1995

U.S. DEPARTMENT OF

Public Health Service

HEALTH AND HUMAN SER

National Institutes of Healt U.S. DEPARTMENT OF ENER Office of Health and

1991-1995

POLICY FORUM ===

The U.S. Human G

of an international

netic and physical m DNA sequence of th

the genomes of sev tightly focused effort track with respect to

Because 3 years have

goals were set, and sophisticated and de

what needs to be do

now available, the g and extended to

(through Septembe

genome initiative. In 1990, the Hun of the National Inst and the Department

veloped a joint research

goals for the first 5 1991-95] of the

Project (1), It has

guide for both the res

he agencies' admir

oping and executing

and assessing its pr

vears. Great strides h the achievement of

particularly with rest

tailed human ger

physical maps of the

the genomes of cert

developing improved

equencing and info

defining the most u gal, and social issues

guisition and use o

Progress toward a

goals for the genome

on schedule or, in

ahead of schedule.

logical improvemen

been anticipated in

lowed more ambi-

this year, it was the

scope of genome

and extend the initi

eas changed the scope

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

physical maps; (iii) the definition of the sequence tagged site (STS) (5) as a common unit of physical mapping; and (iv) improved technology and automation for DNA sequencing. Further substantial im-

A vision for the future of

1993-1998

#### SPECIAL SECTION

G. An, B. D. Watson, C. C. Chiang, Plant Physiol. 81, 301 (1986); A. M. Lloyd et al., Science 234, 464 (1986); K. A. Feldmann and M. D. Marks, Mol. Gen. Genet. 208, 1

1996). 9. For example, see S. A. Kerns 0. N. Bechtold, J. Ellis, G. Pellet et al., Plant J. 5, 551 (1994) 1. V. Sundaresan, Trends Plant

D. Meinke and M. Koomnee P. Fransz et el., ibid, 13, 86 C. Lister and C. Dean, ibid.

nsearch project, progress rej 97-131, Arlangton, VA, 1997 10. M. Bevan et al., Hant Cell 9 11. H. Hofte et al., Hant J. 4, 10

New

The Human Genon

Human Genome Rese Health, Bethesda, MD 20 D. Galas was associate Environmental Researc The Human Genome F Washington, DC 2058 Present address: Da Point, Kirkland, WA 980

G. An, e. D. Western, Science 234, 464 (1986); K. A. Feldmann and M. D. Marks, Mo. (1987). E. M. Meyerowitz and R. E. Pruitt, Science 229, 1214 (1985).

r. u. R. Rick, Genetics 149, 473 (1998).
8. C. Konz, N.-H. Chua, J. Schell, Eds., Methods in Arabidopsis Research (World Scientific, River 1696, HJ.) 1992). J. M. Martiner-Zapater and J. Salinas, Eds., Arabidopsis Protocols, vol. 82 of Methods in Modernia's Rivlane Humann. Totowa. HJ. 1998).

C. Lister and C. Dean, Ibid. (1996).
 E. J. Finnegar, R. K. Genger, V. Mol. Biol. 49, 223 (1998).
 D. Preuss, S. Y. Rhee, R. W. D. Browne, D. Preuss, Proc. No. 17. For recent examples of ident

al., Plant J. 8, 613 (1995); P. Acad. Sci. U.S.A. 99, 8145 ( . For information, contact ht . D. Meinke et al., Eds., "M

Francis S. Collin

the major goals in its 1993-98. A new plan human DNA sequene bitious schedule has by the end of 2003, 2 the course of compl the human sequence plan also includes or ment; for studying h developing technolo ing the sequence of the ethical, legal, and for bioinformatics and of genome scientists

single most important pr one that will perma

F. S. Collins and E. Jordan are National Institutes of Health, B of Biological and Environment 20585, USA, A. Chakravarti isw Genetics, Case Western Reso Cleveland, OH 44106, USA, R University of Utah, Salt Lake Institute of Ethics, Georget \*To whom correspondence

A blueprint fo

rancis S. Collins, I

uyer on behalf of t

ıman Genome l

The completion of a h

comprehensive sequ

ıman genome, in

anniversary year of the double-helical structur

andmark event. The

future of genomics resea

ate to consider the remar

has brought us here.

(Figure 1) shows a time

mark accomplishments

and genomics, beginnin Mendel's discovery of the

and their rediscovery in twentiethcentury. Recog

ereditary material<sup>2</sup>.

structure<sup>5</sup>, elucidation

nologies<sup>5,6</sup>, and establish

utomatable methods

ing7-10 set the stage for th

oject (HGP) to begin

ww.nature.com/nature

ne creativity and deterr

f talented scientists v

the initial objectives of

search has begun.

is project their overa

en achieved at least t

pectation, and a revol

The project's new re-

perimental technolog

adv stream of ever-la

lex genomic data sets th

momic approach of te

public databases and ha

study of virtually all

ment and large-scale gen

ty resource data sets

nportant new dimensi omedical research.

genetics, comparat

roughput biochemi

the vision of the origi

w a reality. In contemplating a

Alan E. Guttmache

S. Choi, R. A. Creelman, J. E. Mullet, R. A. Wing. Hant Mol. 800, Rep. 13, 124 (1995);
 F. Creurot et al., Hant. J. R. 763 (1995).
 R. Schmidt et al., Science 214, 406 (1995);
 E. A. Zachgo et al., Genome Rec. 6, 19 (1996);
 E. A. Zachgo et al., Genome Rec. 6, 19 (1996);
 E. A. Zachgo et al., Genome Rec. 6, 19 (1996);
 E. A. Zachgo et al., Genome Rec. 6, 19 (1996).
 H. Beran et al., Naturo 291, 148 (1998).

1998-2003

2003-2010

genomics research PERSPECTIVE

#### 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.

arrays are now used for dinical detection of genomic imbalances<sup>14</sup> and pharmacogenomic testing is routinely performed before administration of certain medications 15). Together, these achievements (see accompanying paper16) document that genomics is contributing to a better understanding of human biology and to improving human health.

As it did eight years ago<sup>17</sup>, the National Human Genome Research and explore future directions and challenges for the field. These discussions have led to an updated 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 achieverealizing 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 18,19), and clinical discoveries can lead back to understanding disease or even basic biology.

The past decadeh as seen genomics contribute fun damen tal 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 accompanying rollfold).

🥆 ince the end of the Human Genome Project (HGP) in 2003 and the 📉 quickly. Although genomics has already begun to improve diagnostic: publication of a reference human genome sequence12, genomics has and treatments in a few circumstances, profound improvements in the ecome a mainstay of biomedical research. The scientific commu-effectiveness of healthcare cannot realistically be expected for many years nity's foresight in launching this ambitious project<sup>3</sup> is evident in the broad (Fig. 2). Achieving such progress will depend not only on research, but range of scientific advances that the HGP has enabled, as shown in Fig. 1 also on new policies, practices and other developments. We have illu-(see rollfold). Optimism about the potential contributions of genomics for strated the kinds of achievements that can be anticipated with a few improving human health has been fuelled by new insights about cancer 4-7, examples (Box 2) where a confluence of need and opportunities should the molecular basis of inherited diseases (http://www.ncbi.nlm.nih.gov/ lead to major accomplishments in genomic medicine in the coming omimandhttp://www.genome.gov/GWAStudies) and theroleof structural decade. Similarly, we note three cross-cutting areas that are broadly variation in disease, some of which have already led to new therapies 11. relevant and fundamental across the entire spectrum of genomics and Other advances have already changed medical practice (for example, microgenomic 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 Institute (NHGRI) has engaged the scientific community (http://www. be needed to illuminate further those complexities (Fig. 2). The contrigenome.gov/Planning) to reflect on the key attributes of genomics (Box 1) bution 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.

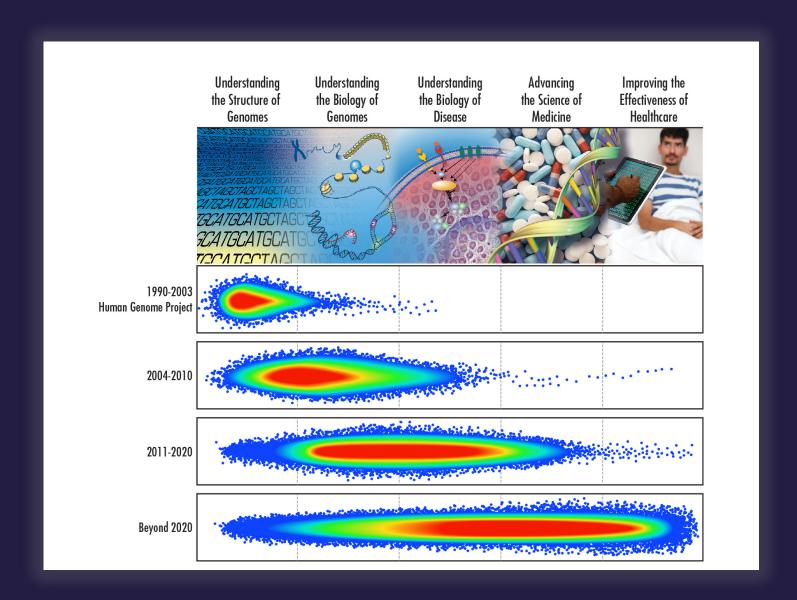
Genomic studies of the genes and pathways associated with disease related traits require comprehensive catalogues of genetic variation, which provide both genetic markers for association studies and variants for identifying candidate genes. Developing a detailed catalogue of variation in the human genome has been an international effort that began with The SNP Consortium<sup>20</sup> and the International HapMap Project<sup>20</sup> (http://hapmap. nchi.nlm.nih.gov), and is ongoing with the 1000 Genomes Project (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

<sup>1</sup>National Human Genome Research Institute, National Institutes of Health, 31 Center Dr., Bethesda, Maryland 20892-2152, USA \*Usts of participants and their affiliations appear at the end of the paper.

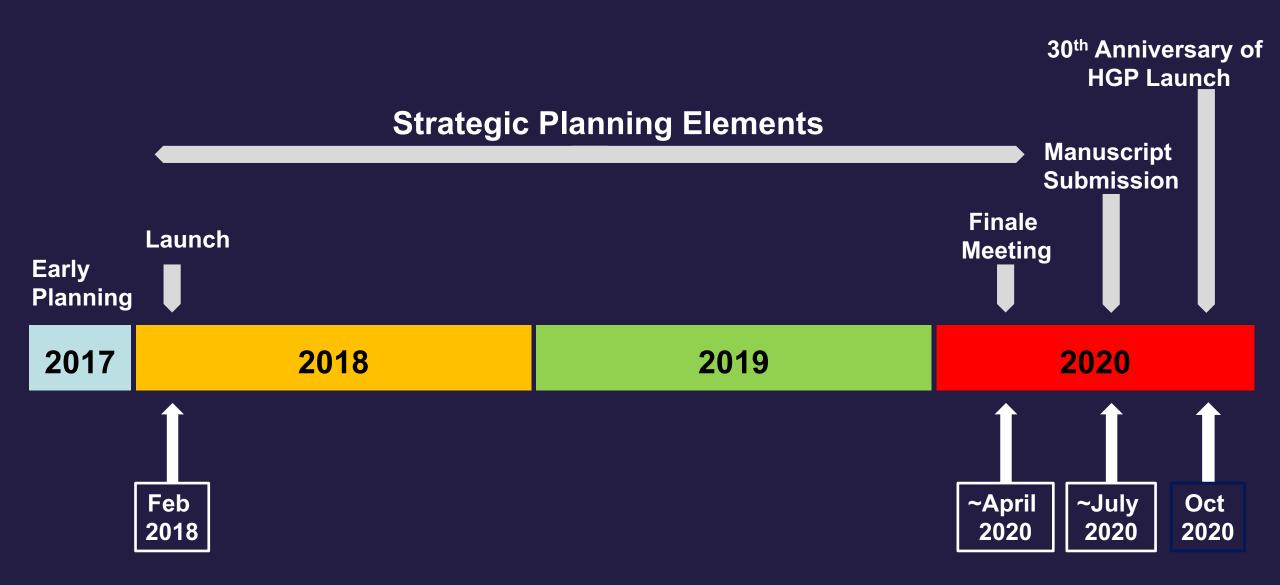
2011-Present

## 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



### Overarching (and Difficult) Issue: Scope

Is the "Genomics 2020" strategic planning process about the future of genomics as a field - OR -

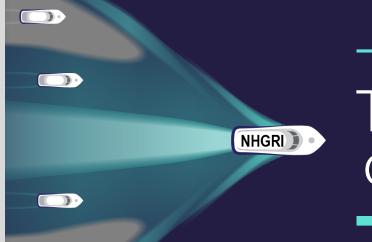
the future of NHGRI-supported genomics research?

## 2020 and Beyond NHGRI-led Genomics Efforts Starting Next Decade

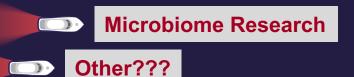


#### **Prototypic Mainstream Areas:**

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



## The Forefront of Genomics®



### 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:

The Forefront of Genomics<sup>®</sup>

### Initial 'Focus Areas' to Organize Brainstorming

- Basic Genomics & Genomic Technologies
- Genomic Data Science
- Genomics of Disease
- Genomics in Medicine & Health
- Society, Education, & Engagement

### **Seeking Your Ideas**



Website: genome.gov/genomics2020

Email: genomics2020@mail.nih.gov

Hashtag: #genomics2020



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

National Human Genome

Research Institute



Laura Lyman Rodriguez, Ph.D.

Division of Policy,

Communications, and Education



Leslie Biesecker, M.D.

Division of Intramural Research



Carla Easter, Ph.D.
Division of Policy,
Communications, and Education



Elise Feingold, Ph.D. Division of Genome Sciences



Adam Felsenfeld, Ph.D. Division of Genome Sciences



Elaine Ostrander, Ph.D. Division of Intramural Research



Bill Pavan, Ph.D.

Division of Intramural Research



Adam Phillippy, Ph.D.

Division of Intramural Research



Anastasia Wise, Ph.D. Division of Genomic Medicine

- To be placed in the queue to ask a question, press \* (star) and 1
- State your name and organization
- To remove yourself from the queue, press the # (pound) key



### Thank you!

Genome.gov/genomics 2020 Genomics2020@nih.gov #genomics2020



