NHGRI’s Genomic Medicine Definition

An emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use.

• Purposefully narrow definition

• By ‘genomic,’ NHGRI means direct information about DNA or RNA; downstream products outside the immediate view

• Metaphorically viewed as a key ‘destination’ for attaining NHGRI’s mission of improving health through genomics research
The Path to Genomic Medicine

Human Genome Project → ? → Realization of Genomic Medicine
A vision for the future of genomics research

Nature 2003

Charting a course for genomic medicine from base pairs to bedside

Nature 2011
Perspective

Charting a course for genomic medicine from base pairs to bedside

Erik D. Groves1, Mark S. Goyer2,3 & National Human Genome Research Institute4

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. New advances in genomics are harmonized to obtain robust longitudinal knowledge about the structure and function of the human genome and to advance the potential benefits of genomic medicine.

Since the end of the Human Genome Project (HGP) in 2003 and the publication of a draft sequence of the human genome, genomic medicine has become a priority of biomedical research. A scientific community is emerging that is leveraging this architectural project to make medical advances at a rapid and unprecedented pace.

In this Perspective, we explore the current status of genomic medicine in the context of advances in basic biological research, clinical practice, and public health. We identify three core areas of genomic medicine that are expected to drive further progress and discuss the challenges and opportunities associated with them.

1. Understanding the biology of genomes

The central theme of genomic medicine is the understanding of the biology of genomes. This includes the identification of genetic factors that contribute to health and disease, the development of diagnostic tests for genetic disorders, and the identification of therapeutic targets for the treatment of diseases.

2. Applications of genomic data

The use of genomic data in clinical practice is rapidly expanding. This includes the use of genomics to guide medical decisions, to personalize treatment plans, and to predict patient outcomes.

3. The future of genomic medicine

The future of genomic medicine is bright. With advances in technology and computational methods, we can expect to see continued progress in understanding the biology of genomes and in developing new applications of genomic data.

References


Image credit: Nature Publishing Group
Five Domains of Genomics Research

1. Understanding the Structure of Genomes
2. Understanding the Biology of Genomes
3. Understanding the Biology of Disease
4. Advancing the Science of Medicine
5. Improving the Effectiveness of Healthcare
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<tr>
<th>Year Period</th>
<th>Understanding the Structure of Genomes</th>
<th>Understanding the Biology of Genomes</th>
<th>Understanding the Biology of Disease</th>
<th>Advancing the Science of Medicine</th>
<th>Improving the Effectiveness of Healthcare</th>
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Green et al. 2011
## Domain 3
**Discovery Research**

- Establish genotype-phenotype associations for human diseases
  - Identify persons at increased risk of disease based on their genomic variants
  - Find all variants related to a given phenotype or disease
  - Characterize variants known to be related to disease or treatment response

## Domain 4
**Clinical Validation**

- Assess outcomes from using genomic information for clinical care
  - Assess impact on health outcomes and care utilization
  - Identify causes of rare or undiagnosed diseases
  - Validate drug targets and develop improved therapeutics

## Domain 5
**Clinical Implementation**

- Develop processes for using genomic information for clinical care
  - Develop clinical informatics systems for reporting genomic information
  - Educate clinicians and patients about use of genomic information
  - Define and disseminate information on actionable clinical variants and relevant evidence base

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### Genomic Medicine

- Disease-Related Genomics Research
NHGRI Programs in Genomic Medicine

Cancer Genomics

Pharmacogenomics

eMERGE Network & eMERGE-PGRN

Dan Roden
NHGRI Programs in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Genomic Medicine ‘Test Drive’ Programs

Clinical Sequencing
Exploratory Research (CSER)
Lucia Hindorff

Implementing Genomics in Practice (IGNITE)
Geoff Ginsburg
NHGRI Programs in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Genomic Medicine ‘Test Drive’ Programs
- Newborn Genomic Analysis

Newborn Sequencing Program

Anastasia Wise
NHGRI Programs in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Genomic Medicine ‘Test Drive’ Programs
- Newborn Genomic Analysis
- Clinical Genomics Information Systems
New NIH-funded resource focuses on use of genomic variants in medical care

Bethesda, Md., Wed., Sept. 25, 2013 - Three grants totaling more than $25 million over four years will help three research groups to develop authoritative information on the millions of genomic variants relevant to human disease and the hundreds that are expected to be useful for clinical practice. The awards are from the National Institutes of Health.

More and more medical and research centers are sequencing the DNA of whole genomes (the body's entire genetic blueprint) or exomes (the genome's protein-coding region) of patients. Each time, millions of DNA differences in genes and the regions between the genes are detected. But doctors struggle to know which of those differences, called variants, are relevant to disease and for a patient's medical care. As a result, information on few genomic variants is used in clinical practice.

The grants will support a consortium of research groups to develop the Clinical Genome Resource (ClinGen). The investigators will design and implement a framework for evaluating which variants play a role in disease and those that are relevant to patient care, and will work closely with the National Center for Biotechnology Information (NCBI) of the National Library of Medicine (NLM), which will distribute this information through its ClinVar database. The grants are funded by the National Human Genome Research Institute (NHGRI) and the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), which, along with NCBI and NLM, are part of NIH. ClinGen was developed from NHGRI's Clinically Relevant Variants Resource program.
NHGRI Programs in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Genomic Medicine ‘Test Drive’ Programs
- Newborn Genomic Analysis
- Clinical Genomics Information Systems
- Ultra-Rare Genetic Disease Diagnostics
Ultra-Rare Genetic Disease Diagnostics

Exome Sequencing: Dual Role as a Discovery and Diagnostic Tool

Clinical application of exome sequencing in undiagnosed genetic conditions

Next-Generation Sequencing for Clinical Diagnostics

Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders

Genomics in Clinical Practice: Lessons from the Front Lines

Howard J. Jacob,¹,5,6* Kelly Abrams,¹² David P. Bick,¹,5,10 Kent Brodie,¹ David P. Dimmock,¹,5,10 Michael Farrell,³ Jennifer Geurts,¹,⁷ Jeremy Harris,¹,⁵ Daniel Helbling,¹,⁵ Barbara J. Joers,¹² Robert Kliegman,⁵ George Kowalski,¹ Jozef Lazar,¹,² David A. Margolis,⁵ Paula North,⁴,⁹,¹¹ Jill Northup,¹ Altheia Roquemore-Goins,¹¹ Gunter Scharer,¹,5,10 Mary Shimoyama,¹,⁷ Kimberly Strong,¹,⁸ Bradley Taylor,¹ Shirng-Wern Tsaih,¹ Michael R. Tschannen,¹ Regan L. Veith,¹,¹⁰ Jaime Wendt-Andrae,¹ Brandon Wilk,¹,⁵ Elizabeth A. Worthey¹,5,⁹

Sci Transl Med (2013)
Undiagnosed Diseases Network (UDN)

- Build upon the successful experience with the NIH Undiagnosed Diseases Program to improve the diagnosis and care of patients with undiagnosed diseases
- Facilitate research into the etiology of undiagnosed diseases
- Create a highly collaborative research community to identify best practices for the diagnosis and management of undiagnosed diseases
October 21, 2013

I am pleased to debut a new means of communicating information from the National Human Genome Research Institute (NHGRI) — The Genomics Landscape. In response to encouragement that I have received from various stakeholders to provide more regular personal updates about topics of interest, I am starting a monthly email message that aims to disseminate information from the NHGRI Director to the broader genomics community and other interested recipients. Each month, I will endeavor to highlight two to four topics, typically featuring one in greater detail.

To subscribe, follow link from:

 genome.gov/Director
• Opened June 14, 2013
• ~4400 square foot exhibition
• Hall 23 (adjacent to Hope Diamond)
• Resident in Smithsonian NMNH for ~1 year
• Subsequently will tour North America for 4-5 years
Exhibition Opening June 14, 2013

GENOME
UNLOCKING
LIFE'S
CODE

unlockinglifescodencode.org
Advancing human health through genomics research