Welcome and Setting a Context

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Genomics Landscape

Past → Present → Future
October, 1990

Human Genome Project Begins

Human Genome Project

The Lewis and Clark Expedition of the 21st Century

Jim Borgman, The Cincinnati Enquirer, 12/17/99
First Eukaryotic Genome Sequence

First Animal Genome Sequence

Genome Sequence of the Nematode C. elegans: A Platform for Investigating Biology

The Genome Sequence of *Drosophila melanogaster*

First Mammalian Genome Sequence

Draft Human Genome Sequence Published
Second Mammalian Genome Sequence

Nature 420:520-562, 2002

Initial sequencing and comparative analysis of the mouse genome
Human Genome Project Ends

April, 2003
Myriad Applications of Genomics

Health, Disease, & Medicine
Genomic Medicine

Healthcare tailored to the individual based on genomic information
A vision for the future of genomics research

A blueprint for the genomic era

Nature 2003
Function of the Human Genome Sequence
The ENCODE Portfolio:
Elucidating Genome Function
New NHGRI Vision for Genomics Published

February, 2011

Charting a course for genomic medicine from base pairs to bedside

Understanding the biology of genomes

Substantial progress in understanding the structure of genomes has revealed much about the complexity of genomic biology. Continued acquisition of basic knowledge about genome structure and function will be needed to illuminate further these accomplishments (Fig. 2). The overall benefits of genomics will include more comprehensive (i.e. integrative) 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 of genomic sequence, such as simple collections of oligo- and, ultimately, functional elements, RNAs, proteins, and other biological molecules, for both human and model organisms. Genomic studies of the gene and pathways associated with disease-related traits require comprehensive catalogues of genetic variation, which provide focal markers for association studies and sequence-based functional, linkage and candidate gene analysis. Developing detailed catalogues of variation in the human genome has been an intensive effort that began with the HapMap Consortium and the International HapMap Project (http://hapmap.ncbi.nlm.nih.gov), and is ongoing with the 1000 Genomes Project (http://www.1000genomes.org).

The past decade saw genome sequencing transform our knowledge about biology and its perturbation to disease. Further deepening this understanding will require continued improvement in genomic medicine (i.e. the use of care based on genomic information). But significant change needs continued development of genomic medicine is challenging. Today, we may have the central armamentarium for clinically relevant genetic testing and treatment (Fig. 1). However, this is not enough. Significant advances will be needed to move from the bed of genomic medicine to the bedside.
Five Domains of Genomics Research

Understanding the Structure of Genomes

Understanding the Biology of Genomes

Understanding the Biology of Disease

Advancing the Science of Medicine

Improving the Effectiveness of Healthcare
Alternate Routes Among Domains

Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
Genomic Accomplishments Across Domains

1990-2003
*Human Genome Project*

2004-2010

2011-2020

Beyond 2020
<table>
<thead>
<tr>
<th></th>
<th>HGP (1st Sequence)</th>
<th>Immediate Post-HGP</th>
<th>Today</th>
</tr>
</thead>
<tbody>
<tr>
<td>Time</td>
<td>~6-8 years</td>
<td>~3-4 months</td>
<td>~2-3 days</td>
</tr>
<tr>
<td>Cost</td>
<td>~$1B</td>
<td>~$10-50M</td>
<td>~$4-8K</td>
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Continued Key Role of Model Systems
Special Thanks!

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Improving human health through genomics research