The Human Genome: Unlocking Life’s Code

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The Relevance of Genomics

Biomedical Researchers

Healthcare Professionals

Patients (and Friends & Relatives of Patients)
Human Genome Project
1990-2003
Turning discovery into health

Advancing human health through genomics research
Genomic Medicine

An emerging medical discipline that involves using an individual’s genomic information as part of their clinical care
New NHGRI Vision for Genomics Published

February, 2011

perspective

Charting a course for genomic medicine from base pairs to bedside

Understanding the biology of genomes

Comprehensive catalogues of genomic data

Comparative genomic studies have been completely redefined and validated. There is an increasing need to improve existing catalogues of genome sequences, such as complete collections of genetic variants, functional genomic elements, RNA, proteins, and other biological molecules, for both humans and model organisms.

Genomic studies of the gene networks and pathways associated with disease-related traits require comprehensive catalogues of genetic variation, which provides critical markers for association studies and pharmacogenetics. - The concept of identifying gene-gene and gene-environment interactions in disease is now necessary because of the complexity of the human genome.}

Figure 1 | Genomics and the Human Genome Project (see accompanying article).
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
Your Genome: By the Numbers

- ~6B nucleotides
- ~3-5M single-nucleotide variants
  - ~150K not in databases
  - ~60 not in either parent
- ~10-20K structural variants
- ~100 ‘disruptive’ variants in genes
- ~20 completely inactivated genes (both copies)
Genomic Architecture of Genetic Diseases

Rare, Simple, Monogenic, Mendelian…
Mostly Coding Mutations

Common, Complex, Multigenic, Non-Mendelian…
Mostly Non-Coding Mutations
Cost per Sequenced Human Genome

Moore’s Law

genome.gov/sequencingcosts
Sequencing a Human Genome

HGP (1st Sequence)

~6-8 years
~$1B

Immediate Post-HGP

~3-4 months
~$10-50M

Today

~2-3 days
~$4-6K
From 1988-2012, genomic activities generated $965 billion in economic output.

In 2012, genomic activities generated $65 billion in economic output.

Genomics has a remarkable demonstrated return-on-investment.
The Future
NEXT EXIT
Genomic Accomplishments Across Domains

1990-2003
Human Genome Project

2004-2010

2011-2020

Beyond 2020
‘Hot Areas’ in Genomic Medicine

Cancer Genomics
Genome Sequencing: Cancer

The Cancer Genome Atlas

News Releases and Announcements

The TCGA Research Network has completed two new characterization studies on acute myeloid leukemia (AML) and endometrial cancer. Read the press releases here.

Cancer Genomics
‘Hot Areas’ in Genomic Medicine

Cancer Genomics

Pharmacogenomics
All patients with same disease

- No response
- Bad side effects
- Good response without any side effects
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare & Common Genetic Diseases
Genome Sequencing: Rare Diseases

- Genomic Basis Known (~4800)
- Genomic Basis Unknown (~1800)
- Suspected Mendelian Disease/Trait (~1900)

Mendelian Diseases/Traits
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare & Common Genetic Diseases
- Prenatal & Newborn Genomic Analysis
- Clinical Genomics Information Systems
Clinical Genomic Information Systems
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare & Common Genetic Diseases
- Prenatal & Newborn Genomic Analysis
- Clinical Genomics Information Systems
- Genomic Medicine ‘Test Drive’ Programs
Bringing Genomic Medicine into Focus

~1990  ~2003  ~2011  ~2020
| 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 |

A pessimist sees the difficulty in every opportunity. An optimist sees the opportunity in every difficulty.

---Winston Churchill
Advancing human health through genomics research