Genomic Medicine and NIH
Francis S. Collins, M.D., Ph.D.
Director, National Institutes of Health
Global Leaders in Genomic Medicine
January 8, 2014
NIH: Steward of Medical and Behavioral Research for the Nation

“Science in pursuit of fundamental knowledge about the nature and behavior of living systems ... and the application of that knowledge to extend healthy life and reduce illness and disability.”
The Human Genome Project

1990–2003
Genetic Variants Associated with Disease Risk
Disorders with Known Molecular Basis

Source: Online *Mendelian Inheritance in Man*, Morbid Anatomy of the Human Genome
The Beery Twins

Alexis and Noah diagnosed with cerebral palsy (CP) at age two

Sequencing found a new genetic disease; readily treated with the addition of an amino acid to diet
Undiagnosed Diseases and NIH

- Undiagnosed Diseases Program (launched 2008)
  - Patients have longstanding medical conditions that have eluded diagnosis; seen at NIH Clinical Center
  - Trans-NIH staff, led by Dr. William Gahl, has:
    - Evaluated ~3,000 medical records
    - Accepted ~700 cases
    - Determined a diagnosis in ~25%

- Undiagnosed Diseases Network (being implemented)
  - Expands Program, analysis, across U.S. (~$145M over 7 years)
  - Harvard University named UDN Coordinating Center
  - 5-7 additional clinical sites to be added this summer
  - Focus on patient phenotyping; gene function research; DNA sequencing; data sharing
Disorders with Known Molecular Basis

Source: Online Mendelian Inheritance in Man, Morbid Anatomy of the Human Genome
NIH: Genomics and Drug Discovery

- **Therapeutics for Rare and Neglected Diseases (TRND)**
  - Collaborations with expert outside labs to speed development of new drugs for rare and neglected diseases

- **Discovering New Therapeutic Uses for Existing Molecules**
  - Pharma partners have made 58 compounds, biologics, and associated data available to select NIH grantees

- **High tech human biochips to test for toxicity** – for all 10 major organ systems
  - Partnership with DARPA; FDA
Our laws and institutions must go hand-in-hand with progress of the human mind.

Thomas Jefferson
Major Legal Decision for Genomic Medicine: U.S. Supreme Court Ruling on Patenting Human Genes

- **1997/1998:** U.S. patents result in exclusive rights for diagnostic testing to Myriad for *BRCA1* and *BRCA2*
- **2009:** AMP/ACLU lawsuit filed asserting that patents on these genes should never have been allowed
- **2013:** U.S. Supreme Court rules that genes in their natural state are not patentable
November 19, 2013: FDA announced first regulatory clearance of high-throughput DNA sequencing device

- Authorized broad clinical use of Illumina MiSeqDx
  - Allows fast, full, accurate sequencing of patient’s genome
- Potential applications include:

  - Assessing disease risk
  - Uncovering drivers of disease process
  - Determining possible responses to drugs
Regulatory Challenges: Preparing the Pathway to Effective Personalized Medicine

- November 22, 2013: FDA warns 23andMe to discontinue marketing its Personal Genome Service until it receives clearance.
Critical Ethical and Policy Considerations for Genomic Medicine

- **GINA** (Genetic Information Nondiscrimination Act, 2008)
  - Preceded by, expands on, **HIPAA** (Health Insurance Portability and Accountability Act, 1996)
  - Extended by Affordable Care Act of 2010

- **Sharing data**: expanded Genomic Data Sharing Policy
  - Promotes sharing of large-scale human, non-human data
  - Public has commented; final policy coming

- **Modernizing human subjects rules**: revisions to the Common Rule are coming
  - Increase protections for participants; decrease unnecessary burdens

- **Incidental findings**: Presidential Commission reports Critical Ethical and Policy Considerations for Genomic Medicine
Science knows no country, because knowledge belongs to humanity and is the torch which illuminates the world.

– Louis Pasteur
NIH... Turning Discovery Into Health