Genomic Medicine and NIH

Francis S. Collins, M.D., Ph.D.
Director, National Institutes of Health

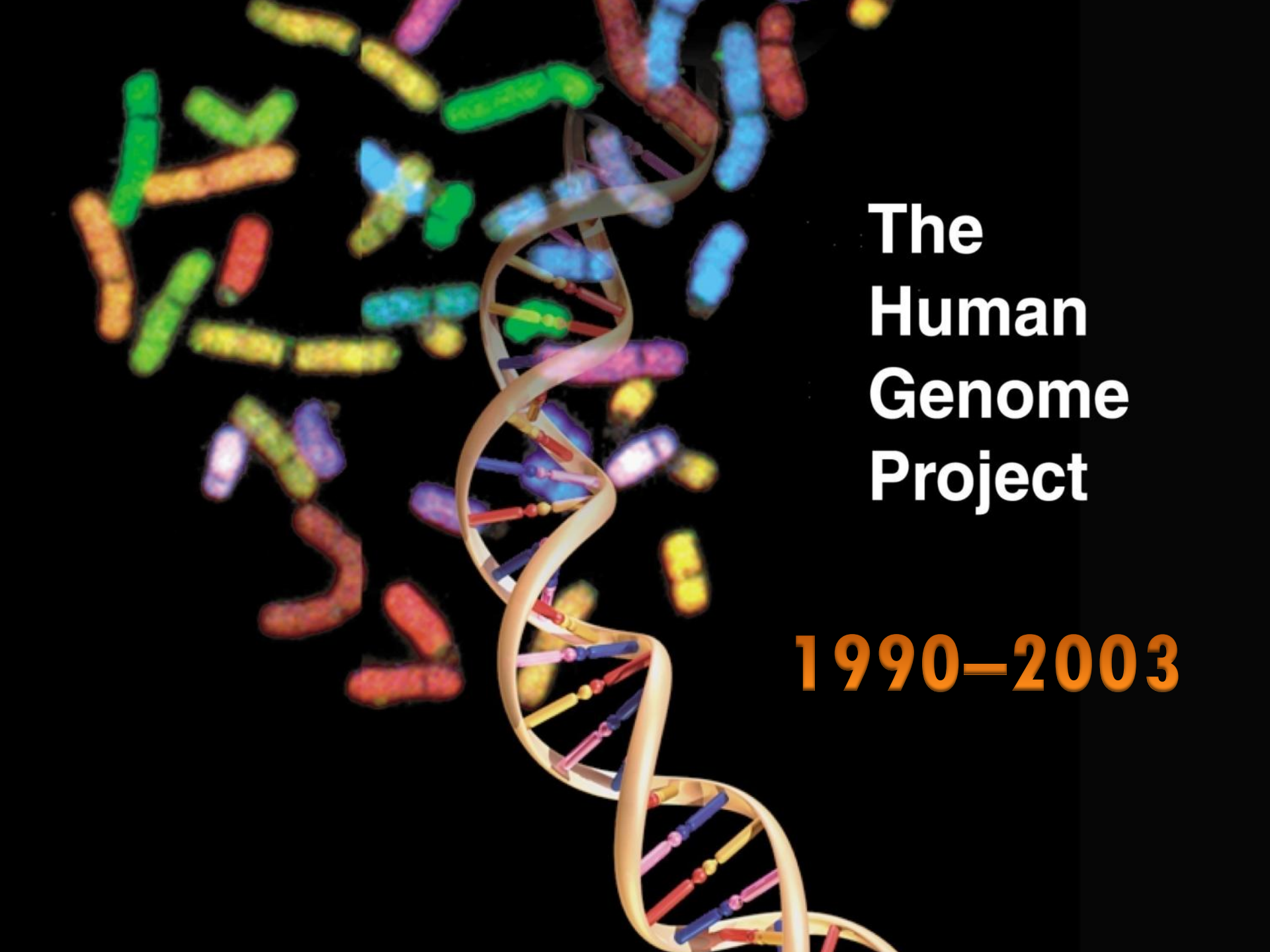
Global Leaders in Genomic Medicine
January 8, 2014

NIH

DEPARTMENT OF HEALTH & HUMAN SERVICES · USA

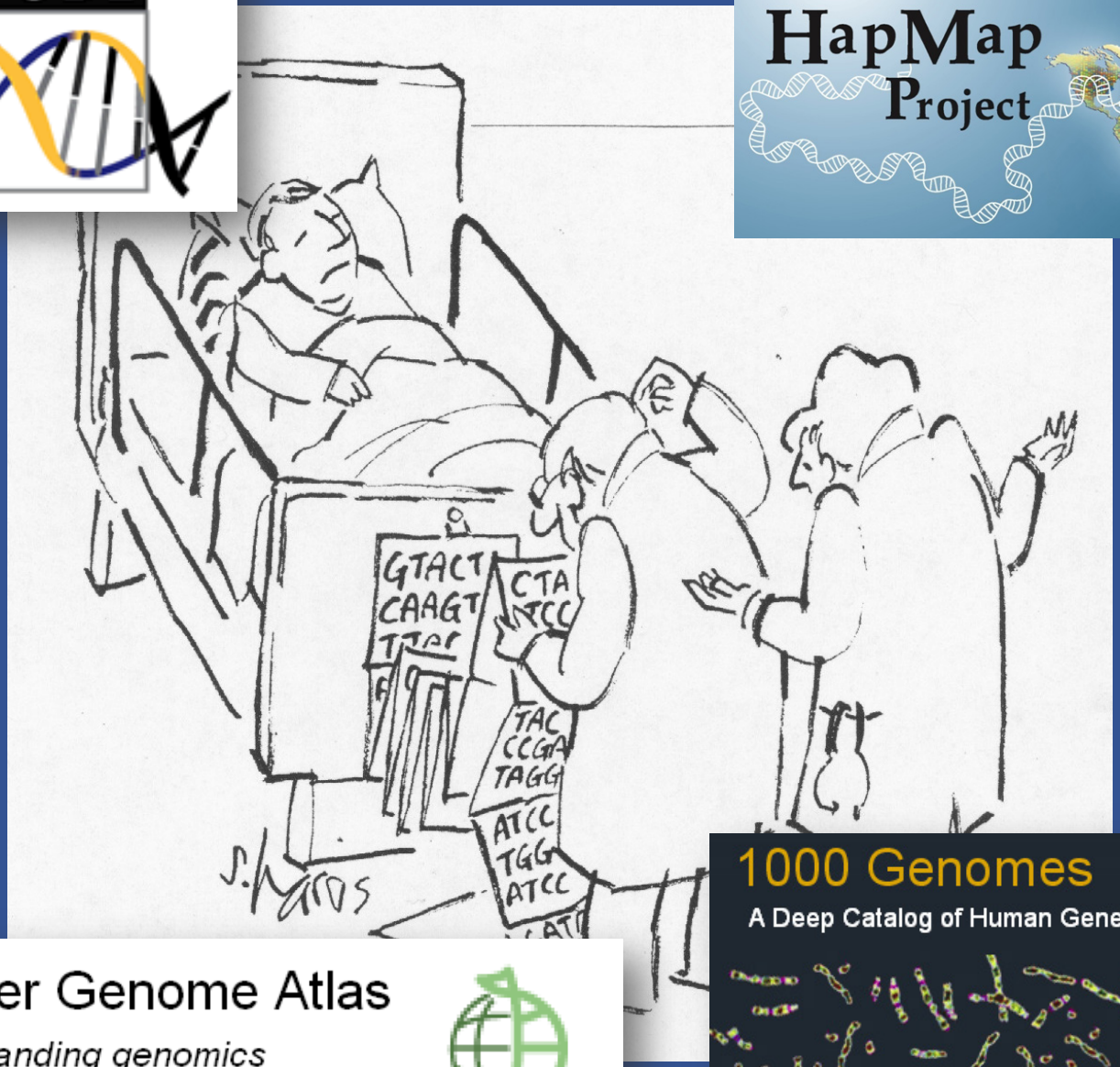
NIH: Steward of Medical and Behavioral Research for the Nation

"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

ENCODE

International HapMap Project



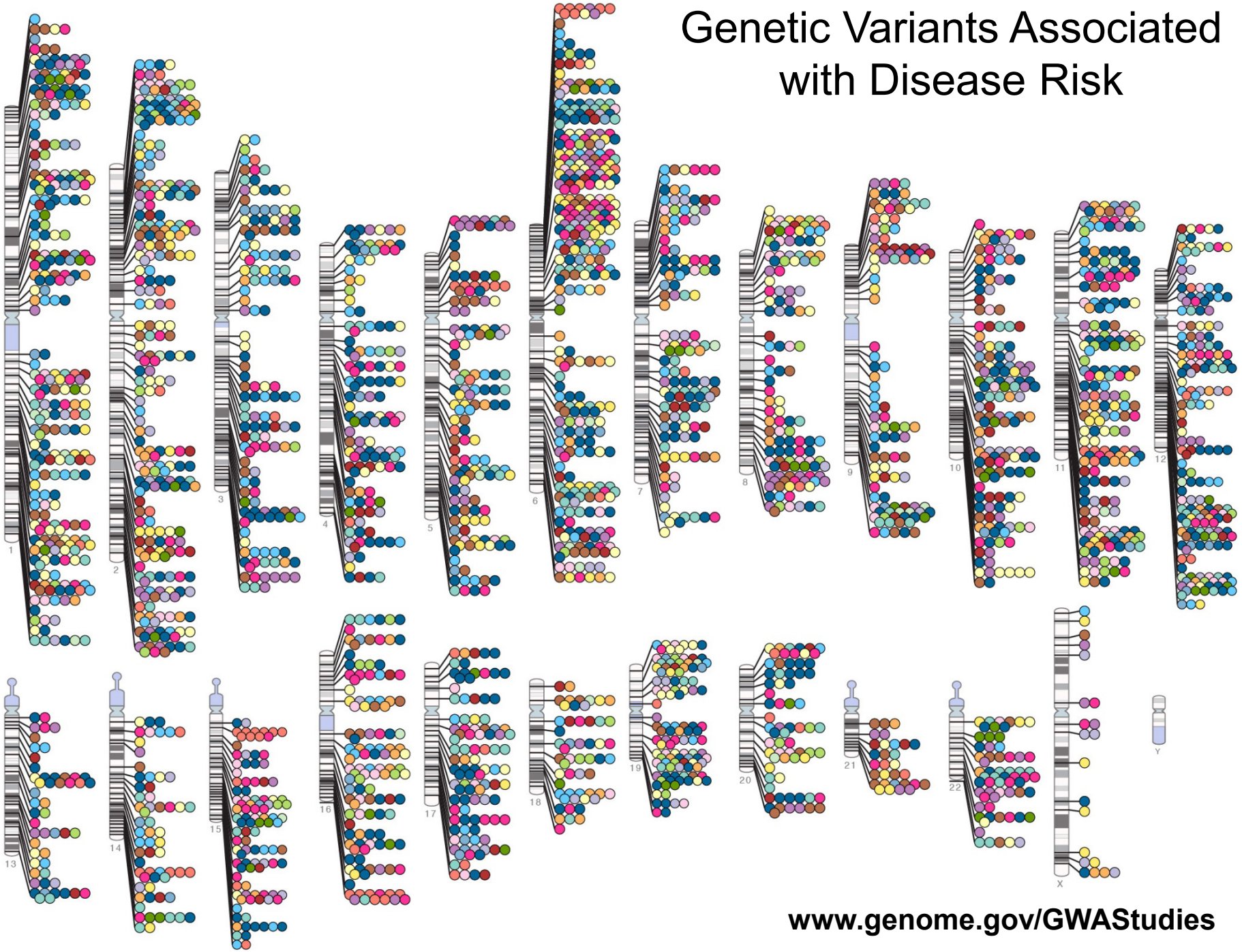
1000 Genomes

A Deep Catalog of Human Genetic Variation

The Cancer Genome Atlas

Understanding genomics to improve cancer care

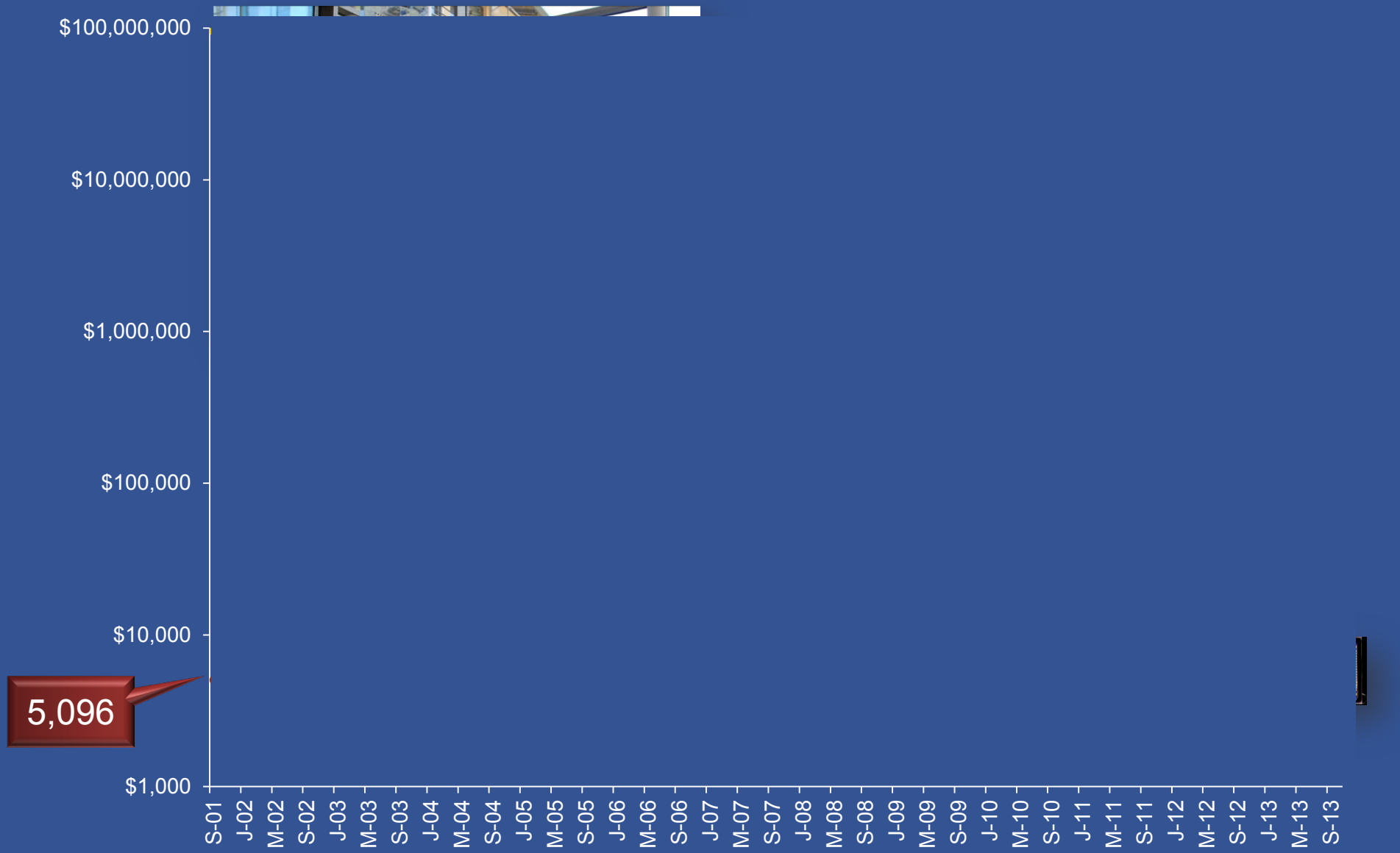
Genetic Variants Associated with Disease Risk



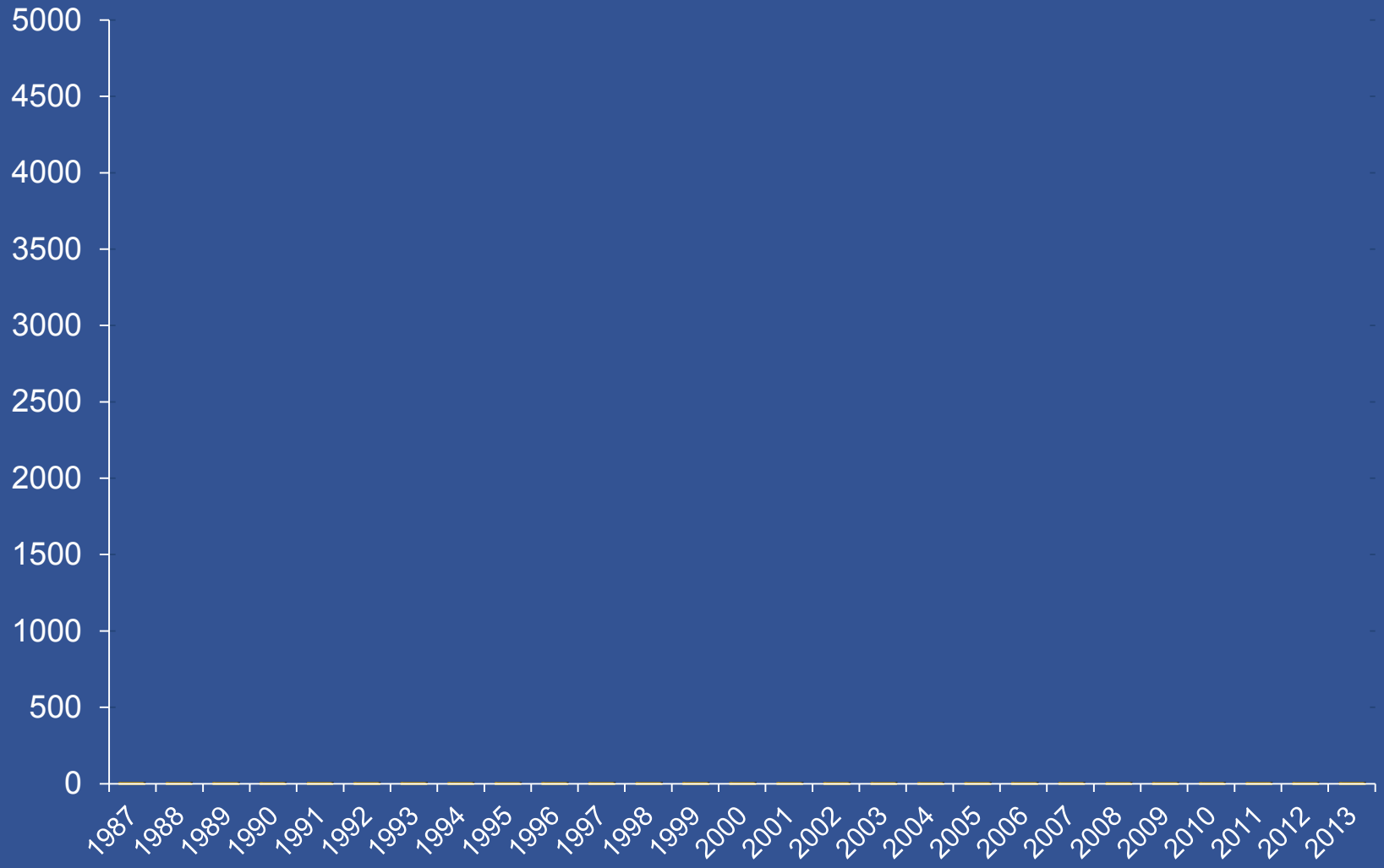
www.genome.gov/GWAStudies

Cost of Sequencing a Human Genome

September 2001–October 2013

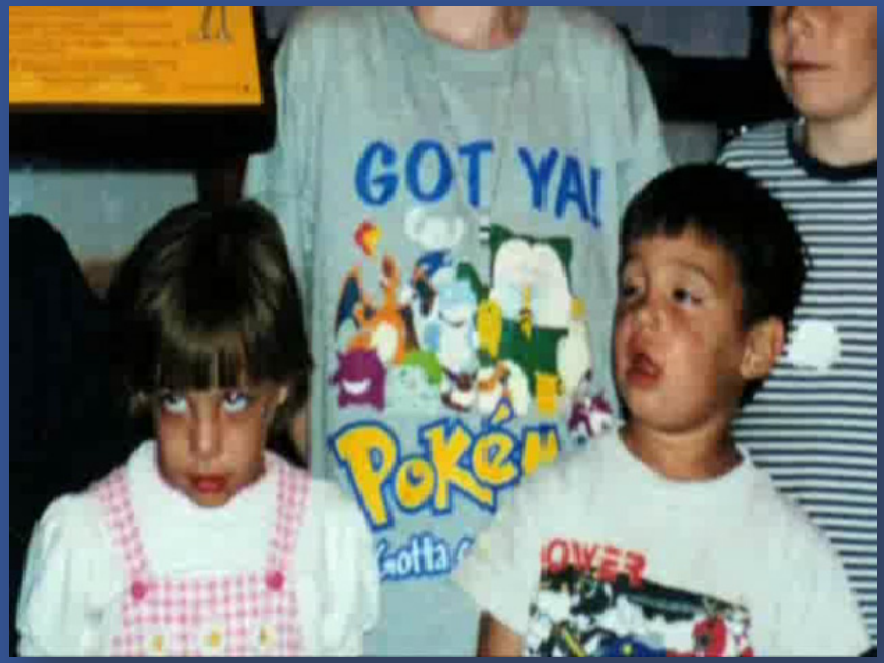


Disorders with Known Molecular Basis



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

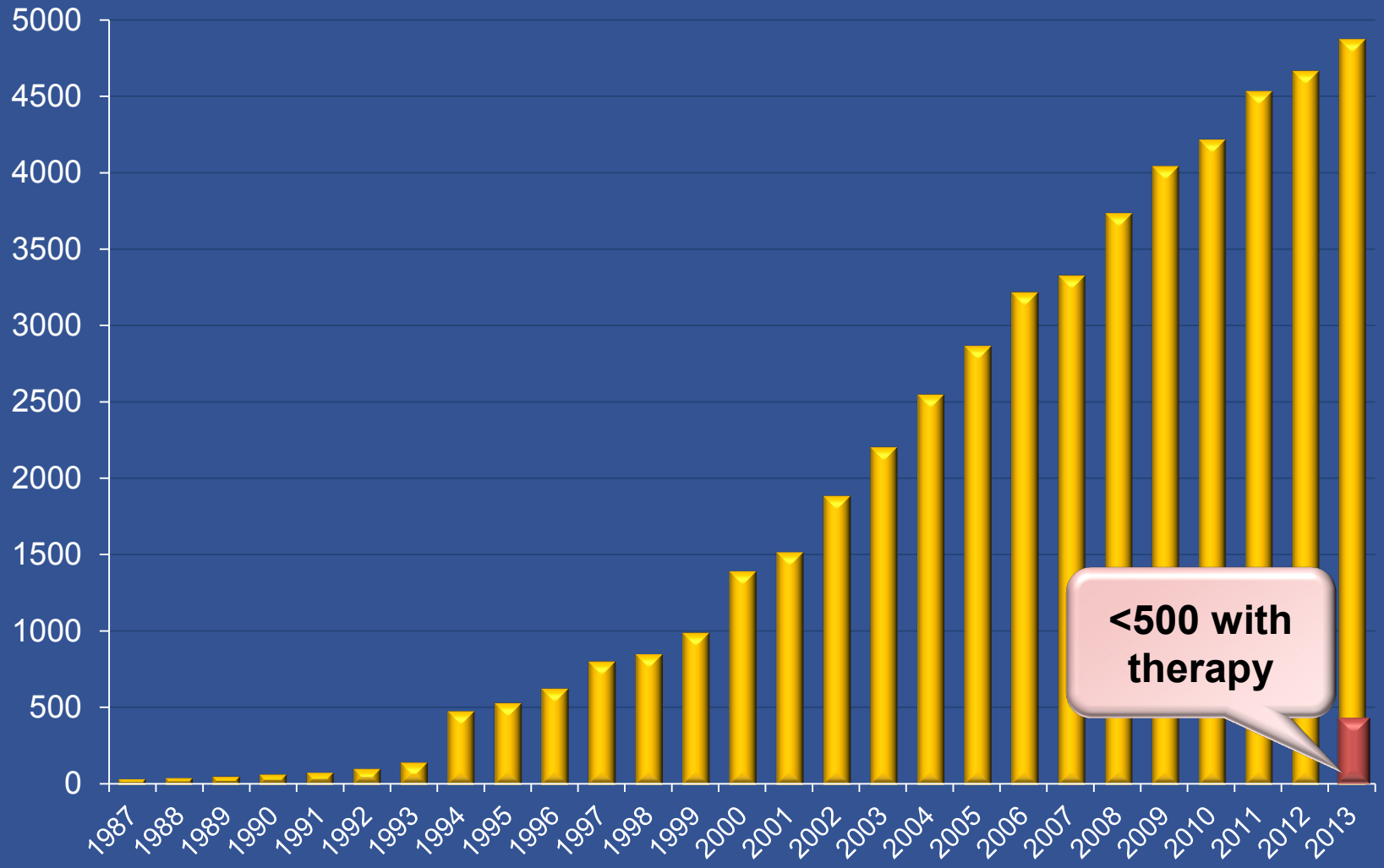
Sequencing found a new genetic disease; readily treated with the addition of an amino acid to diet


A
CELEBRATION

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%

Disorders with Known Molecular Basis



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

NIH: Genomics and Drug Discovery

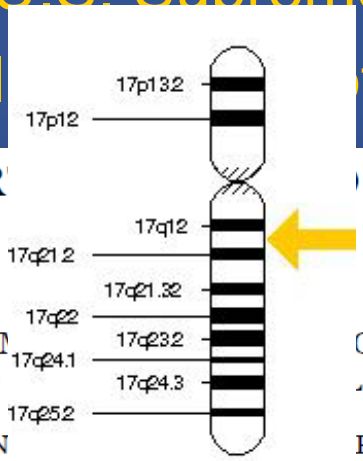
- Therapeutics for Rare and Neglected Diseases (TRND)
  - Collaborations with expert outside labs to speed development of new drugs for rare and neglected diseases
- Discovering New Therapeutic Uses for Existing Molecules
  - Pharma partners have made 58 compounds, biologics, and associated data available 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 [illegible] patentable

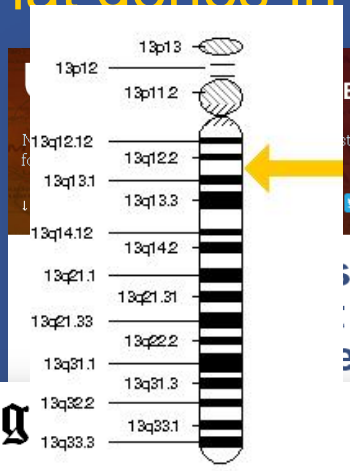


SUPREME COUR[illegible] STATES

ASSOCIATION FOR M[illegible] OGY ET AL.
v. MYRIAD

CERTIORARI TO THE UN[illegible] PPEALS THE

No. 12–398. Argued April 15, 2013—Decided June 13, 2013

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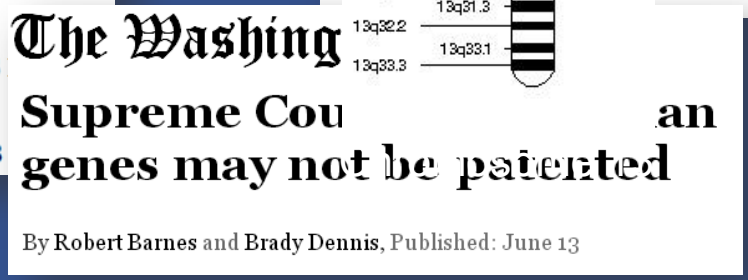
says genes can't be
advocates and
er


The Washing[illegible]

Supreme Cou[illegible]an genes may not be patented

By Robert Barnes and Brady Dennis, Published: June 13

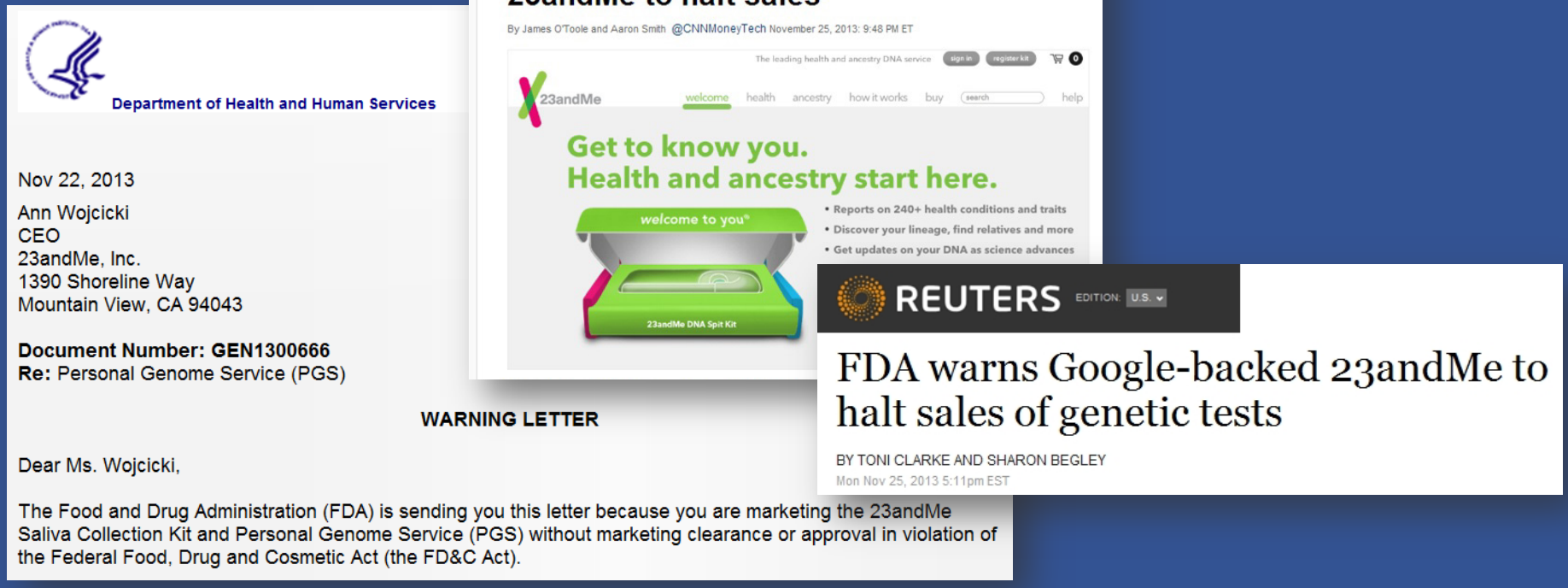
Regulatory Challenges: Preparing the Pathway to Effective Personalized Medicine

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:

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



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FDA orders genetic testing firm 23andMe to halt sales

By James O'Toole and Aaron Smith @CNNMoneyTech November 25, 2013: 9:48 PM ET

23andMe
welcome health ancestry how it works buy search help

Get to know you.
Health and ancestry start here.

- Reports on 240+ health conditions and traits
- Discover your lineage, find relatives and more
- Get updates on your DNA as science advances

welcome to you
23andMe DNA Spit Kit

Department of Health and Human Services

Nov 22, 2013

Ann Wojcicki
CEO
23andMe, Inc.
1390 Shoreline Way
Mountain View, CA 94043

Document Number: GEN1300666
Re: Personal Genome Service (PGS)

WARNING LETTER

Dear Ms. Wojcicki,

The Food and Drug Administration (FDA) is sending you this letter because you are marketing the 23andMe Saliva Collection Kit and Personal Genome Service (PGS) without marketing clearance or approval in violation of the Federal Food, Drug and Cosmetic Act (the FD&C Act).

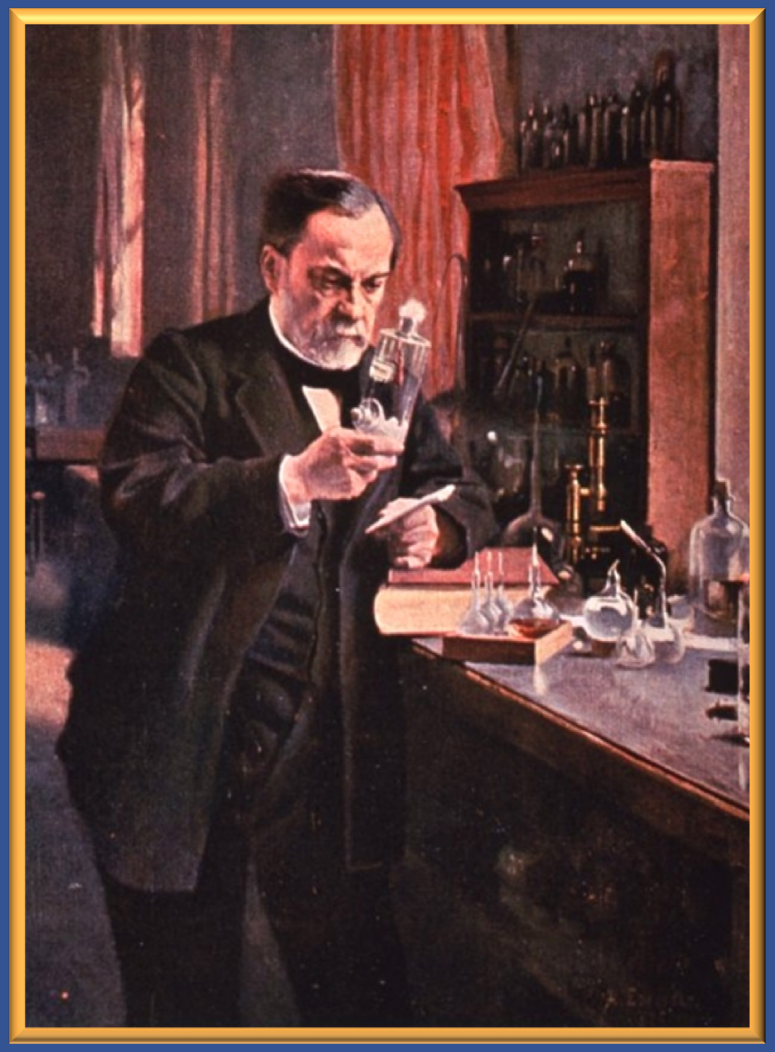
REUTERS EDITION: U.S.

FDA warns Google-backed 23andMe to halt sales of genetic tests

BY TONI CLARKE AND SHARON BEGLEY
Mon Nov 25, 2013 5:11pm EST

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
- Sharin Policy
  - Pror ata
  - Publ
- Modern
-



Science knows no country, because knowledge belongs to humanity and is the torch which illuminates the world.

– *Louis Pasteur*




NIH…

Turning Discovery Into Health