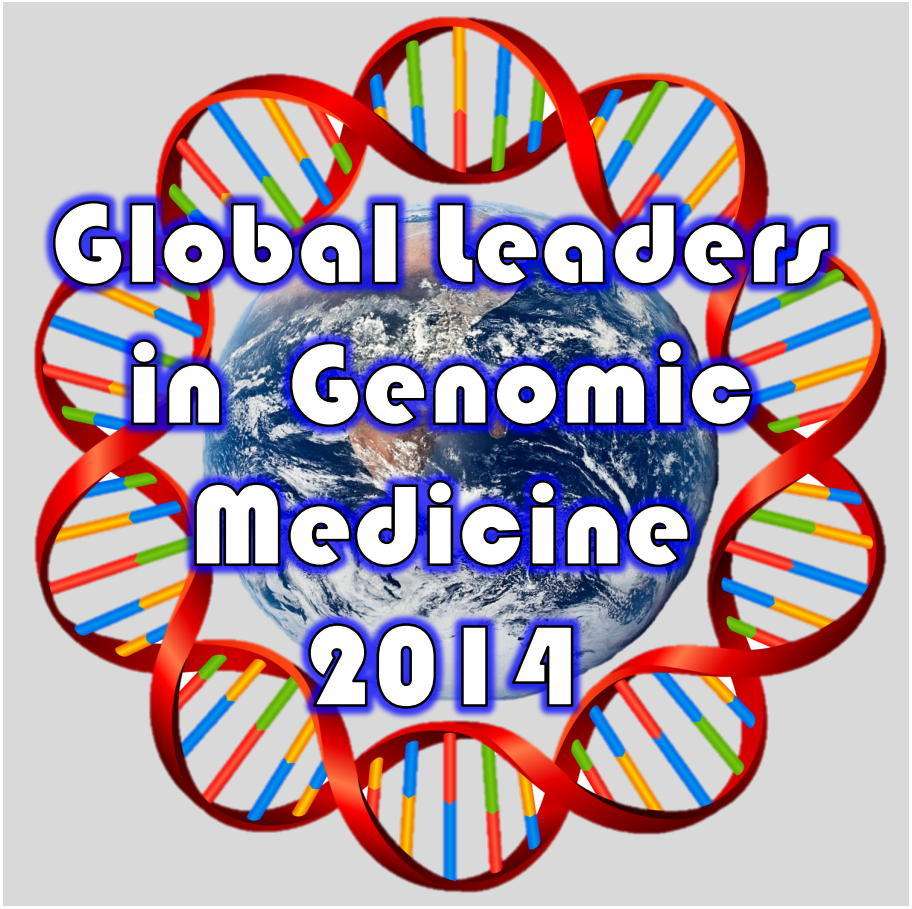


January 8-9, 2014

Washington, DC, USA

April 14, 2003

National Human Genome Research Institute
National Institutes of Health
Department of Health and Human Services
and
Office of Science
U.S. Department of Energy

International Consortium Completes Human Genome Project

All Goals Achieved; New Vision for Genome Research Unveiled

BETHESDA, Md., April 14, 2003 - The International Human Genome Sequencing Consortium, led in the United States by the National Human Genome Research Institute (NHGRI) and the Department of Energy (DOE), today announced the successful completion of the Human Genome Project more than two years ahead of schedule.

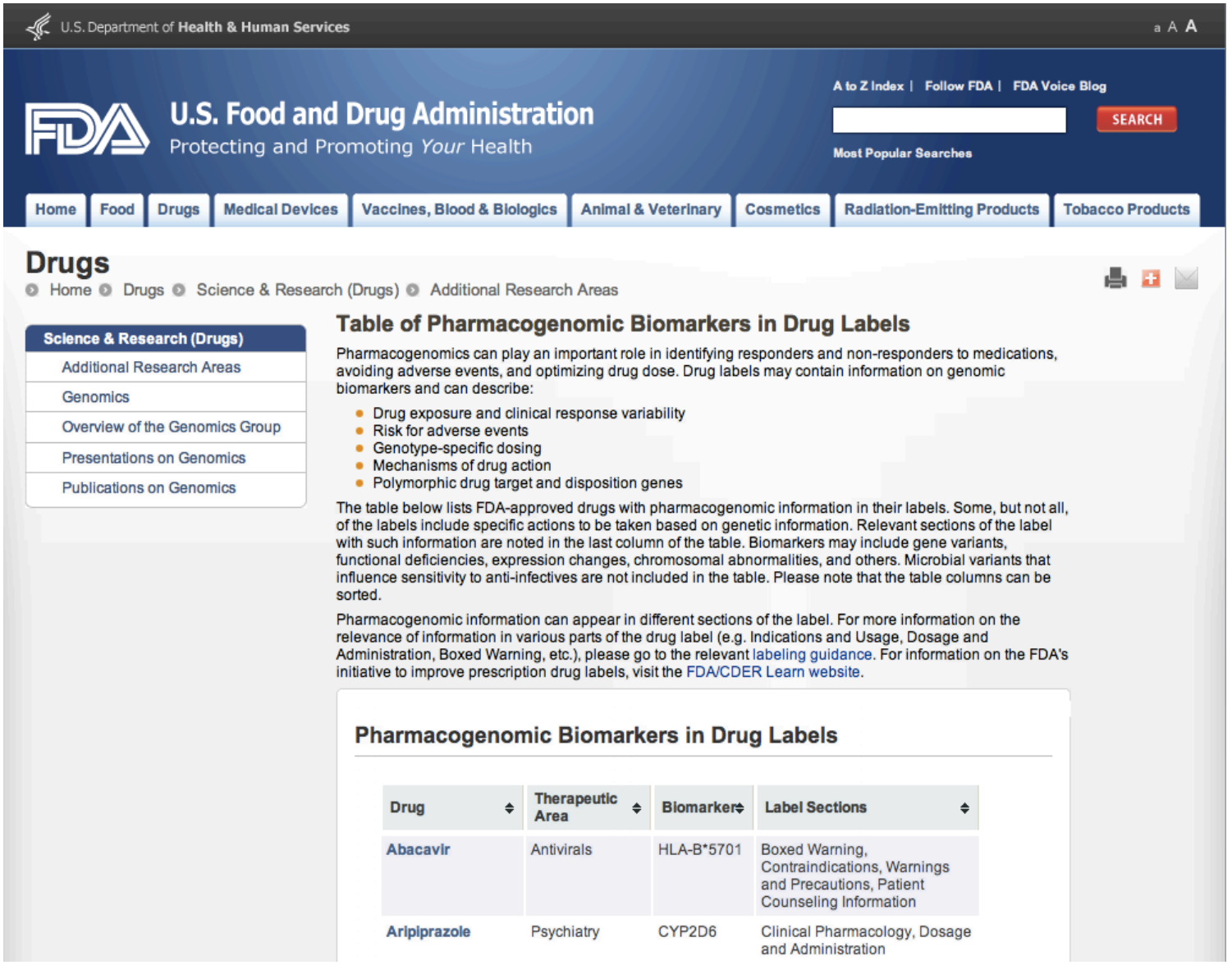
"The completion of the Human Genome Project should not be viewed as an end in itself…. it marks the start of the era of the genome in medicine and health…we urge *all scientists and people around the globe* to join us in turning this vision into reality."

-- Francis Collins --

Ten Years Later:
Major Advances in Genomics and Medicine

- Sequencing – rare and undiagnosed disease
- Family history and risk assessment tools
- Cancer prognosis, diagnosis and risk assessment
- Pharmacogenomics (germ-line, cancer)
- Targeted therapies (cancer, CF, other diseases)

Over 120 FDA Drug Labels Have Genomics …(seldom used)



U.S. Department of Health & Human Services

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Drugs

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Science & Research (Drugs)

- Additional Research Areas
- Genomics
- Overview of the Genomics Group
- Presentations on Genomics
- Publications on Genomics

Table of Pharmacogenomic Biomarkers in Drug Labels

Pharmacogenomics can play an important role in identifying responders and non-responders to medications, avoiding adverse events, and optimizing drug dose. Drug labels may contain information on genomic biomarkers and can describe:

- Drug exposure and clinical response variability
- Risk for adverse events
- Genotype-specific dosing
- Mechanisms of drug action
- Polymorphic drug target and disposition genes

The table below lists FDA-approved drugs with pharmacogenomic information in their labels. Some, but not all, of the labels include specific actions to be taken based on genetic information. Relevant sections of the label with such information are noted in the last column of the table. Biomarkers may include gene variants, functional deficiencies, expression changes, chromosomal abnormalities, and others. Microbial variants that influence sensitivity to anti-infectives are not included in the table. Please note that the table columns can be sorted.

Pharmacogenomic information can appear in different sections of the label. For more information on the relevance of information in various parts of the drug label (e.g. Indications and Usage, Dosage and Administration, Boxed Warning, etc.), please go to the relevant labeling guidance. For information on the FDA's initiative to improve prescription drug labels, visit the FDA/CDER Learn website.

Pharmacogenomic Biomarkers in Drug Labels

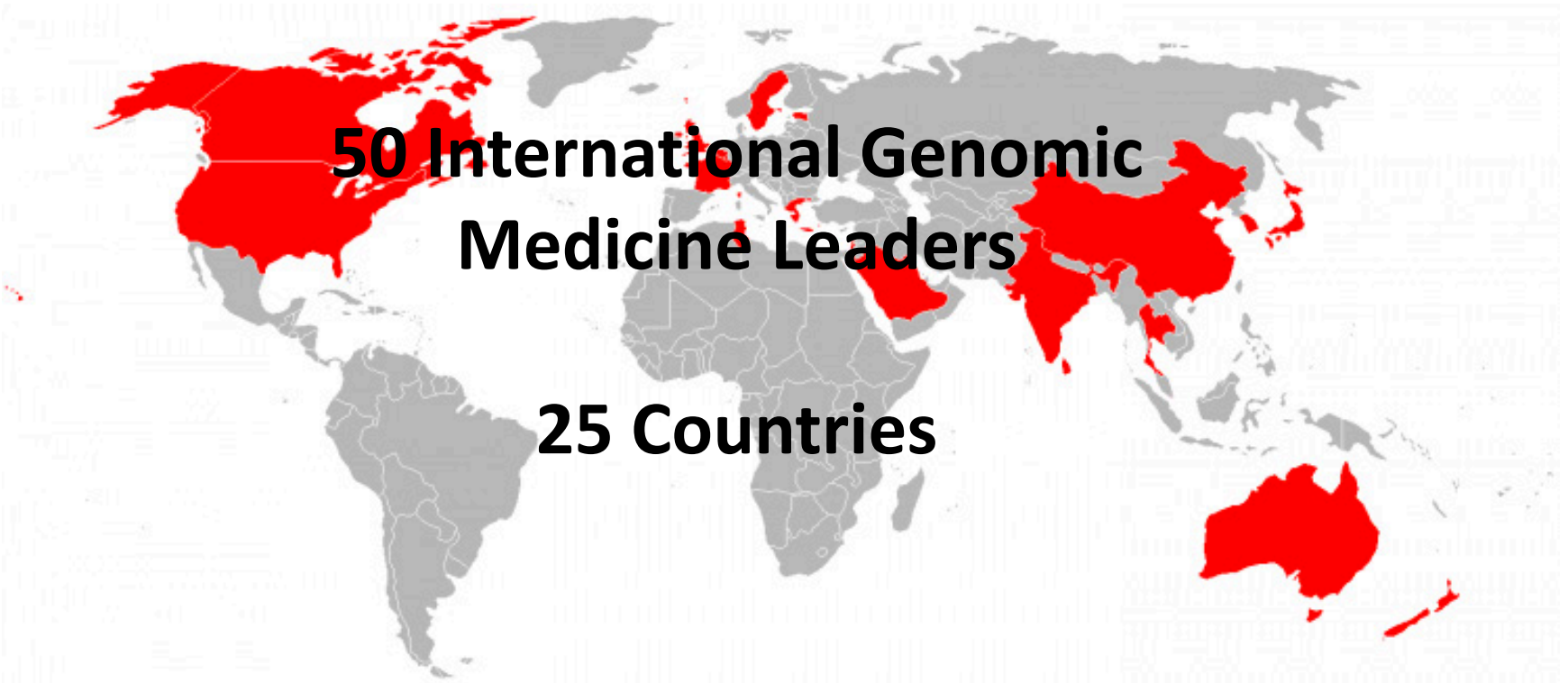
Drug	Therapeutic Area	Biomarker	Label Sections
Abacavir	Antivirals	HLA-B*5701	Boxed Warning, Contraindications, Warnings and Precautions, Patient Counseling Information
Aripiprazole	Psychiatry	CYP2D6	Clinical Pharmacology, Dosage and Administration

Major Questions for Genomic Medicine

- How to develop evidence of benefit/value and what evidence is needed?
- How to engage institutional leadership and physicians
- How to educate patients, physicians, public
- How to achieve full EMR integration of genomic results, custom reporting tools and decision support software
- How to create a viable financial model -- not by **adding costs** but by **reducing costs**

Source: Manolio et al GIM 2013

Global Attendance – We Are Grateful



The International Landscape

	Today (%)			Desired in 3-5 years (%)		
Clinical Genomic Capability	Not at all	Specialized Centers	Widely available	Not at all	Specialized Centers	Widely available
Pharmacogenomics	23	**66**	11	17	29	**56**
Germline sequencing	23	**66**	11	11	**72**	17
Tumor sequencing	17	**72**	11	11	**60**	29
Newborn sequencing	**64**	36	0	11	**72**	17
Maternal fetal sequencing	29	**65**	6	11	**66**	23
Rare disease diagnosis	23	**71**	6	6	**77**	17
Sequencing for identification of infectious agents	17	**72**	11	11	36	**53**
RNA profiling	**50**	**50**	0	11	**66**	23
Metabolomics	**53**	47	0	11	**78**	11
Proteomics	**64**	36	0	29	**60**	11
Systematic family history	17	36	46	6	23	**71**
Genetic counselors	23	**47**	30	6	17	**77**
Electronic medical record	23	**47**	30	6	0	**94**
Clinical decision	33	**33**	33	6	0	**94**

Global Grand Challenges for Genomic Medicine

- Evidence of efficacy or effectiveness
- Lack of Reimbursement
- Evidentiary thresholds
- Bioinformatics and EMR infrastructure
- Access to POC education and CDS
- Expertise and training programs
- Where to invest?

Possible Outcomes

- An international steering group
  - Develop a collective agenda to enable genomic medicine implementation
- Working groups
  - develop and implement key components of such an agenda
- International collaborations or pilot projects
- Others?

genome.gov
National Human Genome Research Institute
National Institutes of Health

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

Global Leaders in Genomic Medicine: Agenda

- General Overview of Genomic Medicine in US
- International Genomic Medicine Applications and Initiatives
- Panel Discussion on International Implementation of Genomic Medicine
- International Genomic Medicine Applications and Initiatives (con't)
- Three NIH initiatives
- Smithsonian exhibit – educational initiatives for the public
- Breakout sessions on 5 topics
  - IT/Bioinformatics
  - Education and Workforce Building
  - Evidence Generation
  - Pharmacogenomics
  - Policy
- Report out – Action oriented
- Next Steps

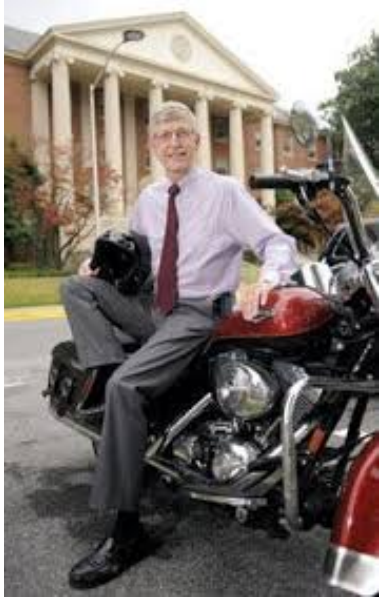
Meeting Objectives

- Identify areas of active translation and implementation
- Prioritize common barriers to implementation in healthcare
- Frame a policy agenda to advance the field
- Highlight nations with unique capabilities
- Discuss opportunities for international collaborations

Introductions

Thank you

- Rita Chambers – Duke University
- Teji Rakhra-Burris – Duke University
- Maggie Bartlett– NHGRI
- Shane Clark –NHGRI
- Alvaro Encinas - NHGRI
- Allison Mandich - NHGRI
- Jackie Odgis – NHGRI
- Tonia Dickerson - IOM
- Patsy Powell– IOM



Francis Collins MD PhD