



Remarks from the NHGRI Director

Eric Green, M.D., Ph.D.
Director, NHGRI

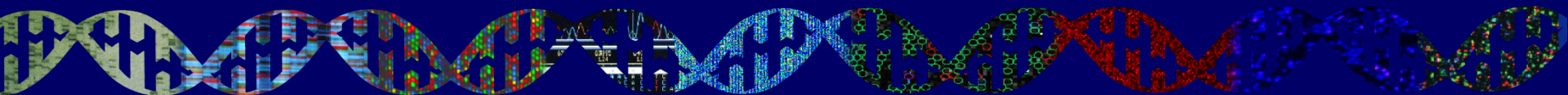


I. NHGRI Education Efforts

II. Undiagnosed Diseases Program

III. NIH & Big Data

IV. 2013 Celebrations

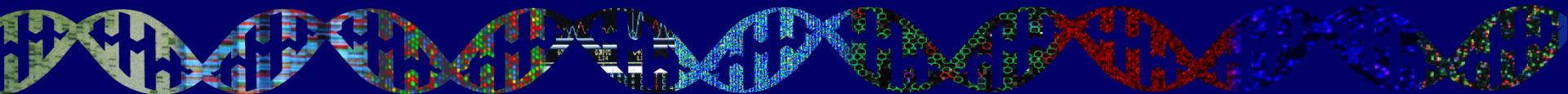


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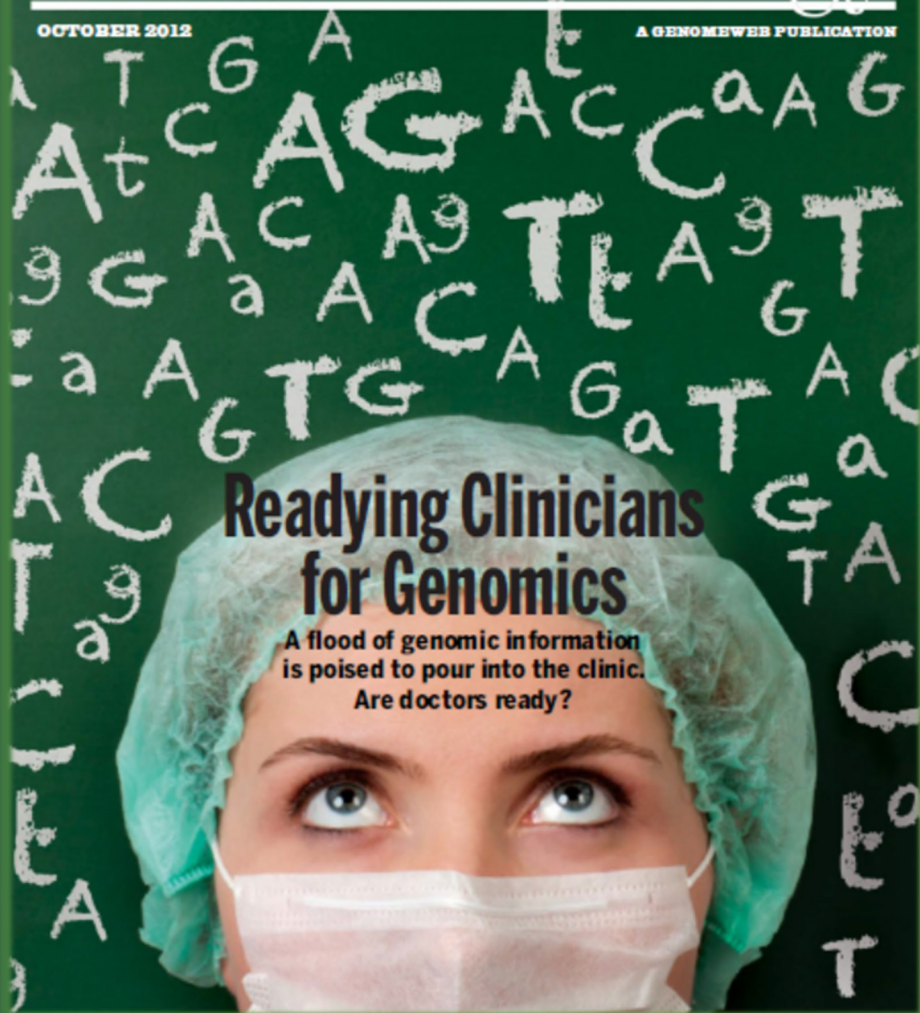
Genome Technology

OCTOBER 2012

A GENOMEWEB PUBLICATION

Readying Clinicians for Genomics

A flood of genomic information
is poised to pour into the clinic.
Are doctors ready?



PERSONALIZED MEDICINE

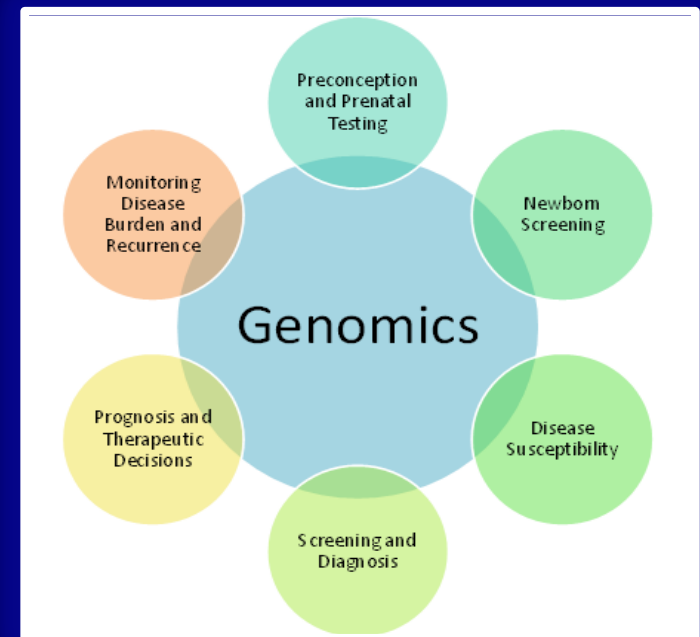
Reality Check: Educating Physicians on Genomic Medicine

Medical schools are increasingly adding genetics and genomics to their curricula, but will it be enough to make personalized medicine a reality for patients?



NHGRI & Healthcare Provider Education

- Important component of NHGRI's mission
- Elements within different NHGRI components
- Long-term goal is to advance understanding of healthcare providers about advances in science, technology, and evidence development
- Assure productive utility of genomic information for clinical care



NHGRI Educational Resources

G2C2: Resource Repository

Talking Glossary

G3C: Case Scenarios

The 2012-2013 Genomics in Medicine Lecture Series

This five-lecture series by top experts in genomics will enhance health-care professionals' understanding of the intersection between genomics and medicine. The series is sponsored by the National Human Genome Research Institute (NHGRI), in collaboration with Suburban Hospital and Johns Hopkins University School of Medicine. Each lecture takes place at Suburban Hospital's lower level auditorium at 8600 Old Georgetown Road in Bethesda, Md. All are welcome to the hour-long lectures, which begin at 8 a.m. on the first Friday of the month. Advanced registration is not required; however, those requesting continuing medical education (CME) credits are asked to sign in.

Lectures are recorded and posted on [GenomeTV](#), NHGRI's YouTube channel at a later date.

Suburban Hospital

For more information about the *Genomics in Medicine* lecture series, please contact Susan Laine at Suburban Hospital, slaine@suburbanhospital.org, or Alice Bailey at NHGRI, baileval@mail.nih.gov.

See Also:
[YouTube Series Playlist](#)
[2011-2012 Genomics in Medicine Lecture Series](#)

Genomic Medicine Lecture Series

NHGRI and Clinical Journals

COMMENTARY

Genomics Education for Health Care Professionals in the 21st Century

W. Gregory Feero, MD, PhD
Eric D. Green, MD, PhD

90% lacked confidence in their clinician's ability to understand and use genomic information.⁷ Past efforts to enhance the genomics literacy of health care professionals have often taken the form of a push of information from the genomics community to other professional groups. The underlying assumption of these efforts has been that spontaneous interest in additional genomics education would follow. The push approach has met with reasonable success in the nursing and physician assistant communities. For example, the nursing profession has internally developed genomics education competencies, which have now been broadly adopted across 50 organizations.⁸ Linking these competencies with program accreditation and

RECENT GENOMIC DISCOVERIES HAVE BROUGHT ABOUT far-reaching advances in understanding the molecular basis of human health and disease. The vision for the future of genomics research developed by the National Human Genome Research Institute suggests more discoveries are likely to occur over the next few decades.¹ These insights have helped reveal remarkable and unexpected complexities of human biology; however, this scientific reality has slowed the immediate trans-

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

Research Funding | Research at NHGRI | Health | Education | Issues in Genetics | **Newsroom** | Careers & Training | About | For

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Recent Journal Articles by NHGRI Researchers and Staff

Archive of Recent Articles from NHGRI 2002 - 2008

New England Journal of Medicine Series on Genomic Medicine

New England Journal of Medicine Series on Genomic Medicine

- Genomic Medicine: An Updated Primer
- GWAS and Assessment of the Risk of Disease
- New Therapeutic Approaches to Mendelian Disorders
- Ancestry and Disease in the Age of Genomic Medicine
- Genomics, Type 2 Diabetes, and Obesity
- Genomics and the Continuum of Cancer Care
- Genomics and Drug Response
- Genomics and the Eye
- Microbial Genomics and Infectious Diseases
- Genomics, Health Care, and Society
- Genomics and the Multifactorial Nature of Human Autoimmune Disease
- Genomics of Cardiovascular Disease
- Genomics and Perinatal Care
- Genomics, Intellectual Disability, and Autism
- An Editorial: Realizing Genomic Medicine

Genomics in PA Practice

Parkinson disease
Nguyen H. Park, MS, PA-C May 28, 2012
This article reviews the diagnosis and treatment of Parkinson disease and defines how genetics and genetic testing can play a role.

Hereditary breast and ovarian cancer
Phyllis Barks, MPH, PA, Constance Goldgar, MS, PA-C March 26, 2012
Patients who test positive for the BRCA1/2 mutations that cause breast and ovarian cancers have options when it comes to surveillance and prevention.

Chromosomal microarray testing
W. Andrew Faucett, MS, CGC, Melissa Savage, MS, CGC
January 30, 2012
CMA testing of children with autism, intellectual impairment, or multiple congenital anomalies detects clinically significant chromosomal deletions and duplications that are not visible by routine chromosome studies.

Preimplantation genetic diagnosis
R. Lynn Holt, MS, CGC September 22, 2011
This article reviews the use of preimplantation genetic diagnosis (PGD), an assisted reproductive technology that has been used to evaluate embryos for genetic conditions prior to implantation.

JAMA

Education Issue

(2011)

NEJM

Genomics Series

(2010-2012)

**Journal of Nursing
Scholarship**

Genomics Issue

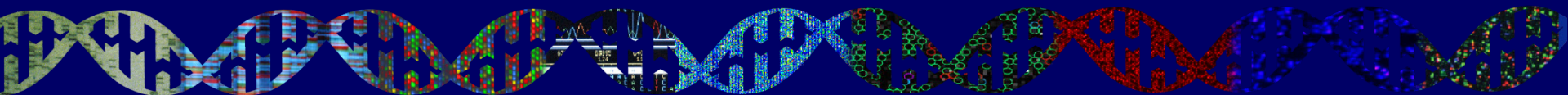
(Pending 2013)

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Undiagnosed Diseases Program (UDP)



- To assist patients with unknown disorders reach an accurate diagnosis
- To discover new diseases that provide insight into human physiology and genetics
- >500 patients to date; definitive diagnoses in >39
- >16 new human genetic disorders identified

UDP: A New Common Fund Program

- **Common Fund Support: ~\$145M over 7 years**
- **Expansion to a national UDP Network**
- **Network of ~5-7 extramural sites**
- **Improved data storage, access, and analysis**
- **Fund basic researchers to elucidate mechanisms of disease**
- **Training and fellowship programs for rare disease diagnostics**

UDP: Initial Funding Announcements



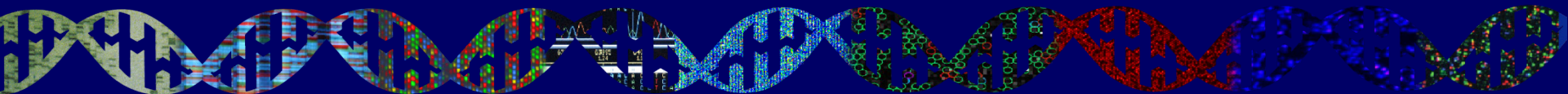
- **RFA-RM-12-020: Coordinating Center for an Undiagnosed Diseases Network (U01)**
Applications received; upcoming review
- **PA-13-076: Gene Function Studies to Investigate Rare and Undiagnosed Diseases**
Receipt date: February 26, 2013

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Breaking News



The Largest Bottleneck in Biomedical Research... ...Pick Your Data-Related Metaphor!



4 September 2008 | www.nature.com/nature | \$10

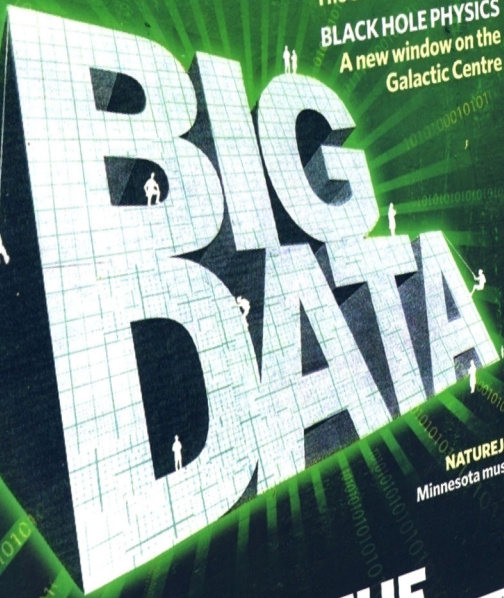
THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

nature

THE BITER BIT
Viral infections for viruses

TROPICAL CYCLONES
The strong get stronger

BLACK HOLE PHYSICS
A new window on the Galactic Centre



BIG DATA

NATUREJOBS
Minnesota musings

SCIENCE IN THE PETABYTE ERA

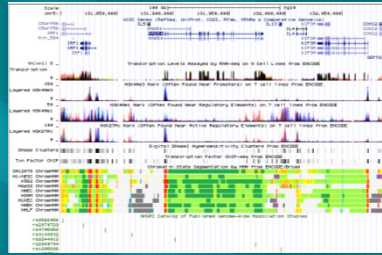


11 February 2011 | \$10

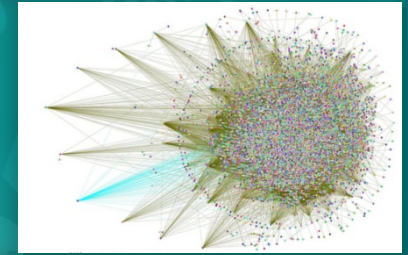
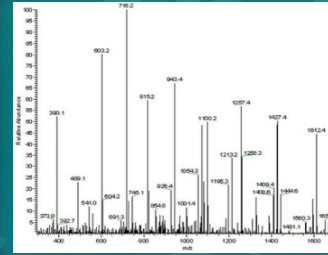
Science



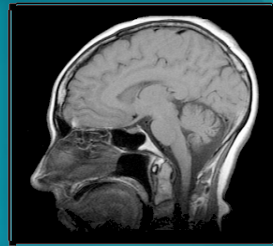
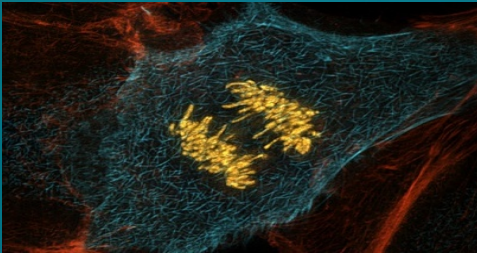
Myriad Data Types



Genomic



Other 'Omic



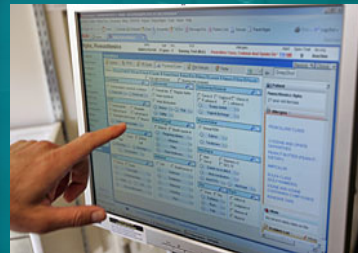
Imaging



Phenotypic



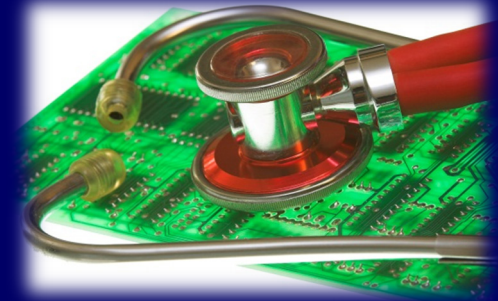
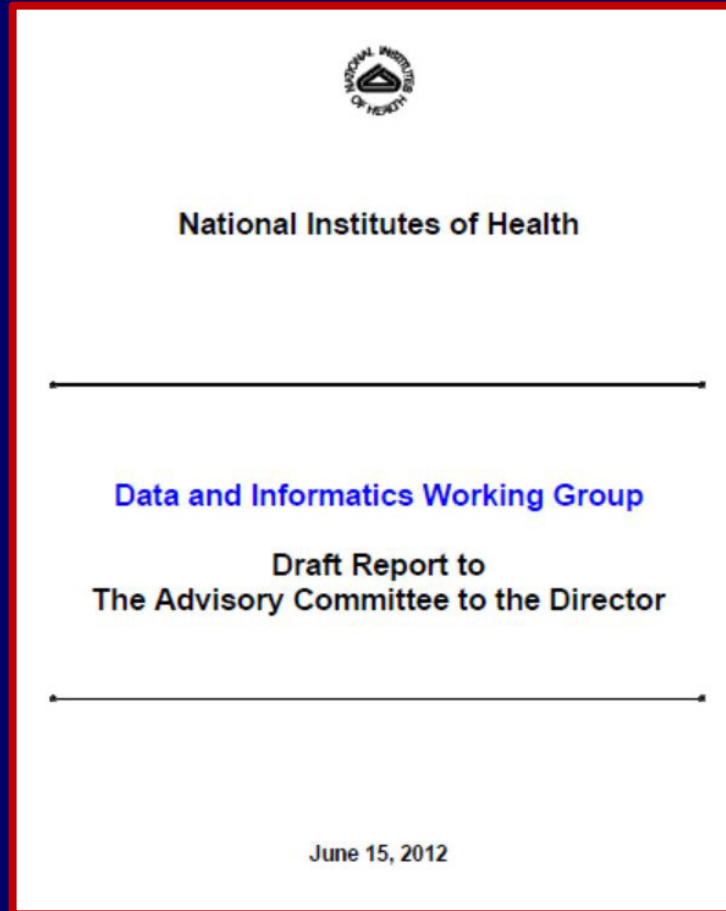
Exposure



Clinical

Data and Informatics Working Group

ADVISORY COMMITTEE TO THE DIRECTOR



acd.od.nih.gov/diwing.htm

NIH is Tackling the 'Big Data' Program

1. New NIH Leadership Position:

Associate Director for Data Science

2. New NIH Scientific Data Council

3. New Trans-NIH Initiative:

Big Data to Knowledge (BD2K)



BD2K: Four Programmatic Areas

I. Facilitating Broad Use of Biomedical Big Data



II. Developing and Disseminating Analysis Methods and Software for Biomedical Big Data



III. Enhancing Training for Biomedical Big Data



IV. Establishing Centers of Excellence for Biomedical Big Data



I. NHGRI Education Efforts

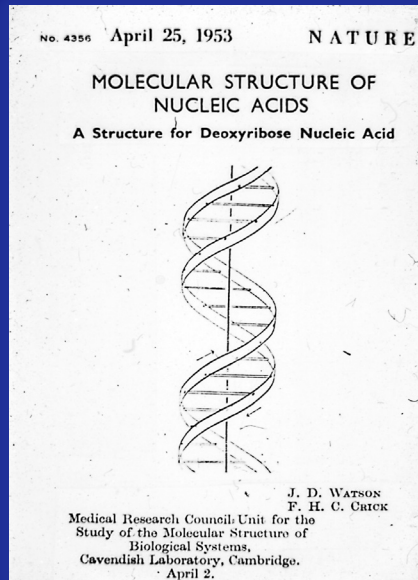
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2013: A Celebratory Year for Genomics



60th Anniversary



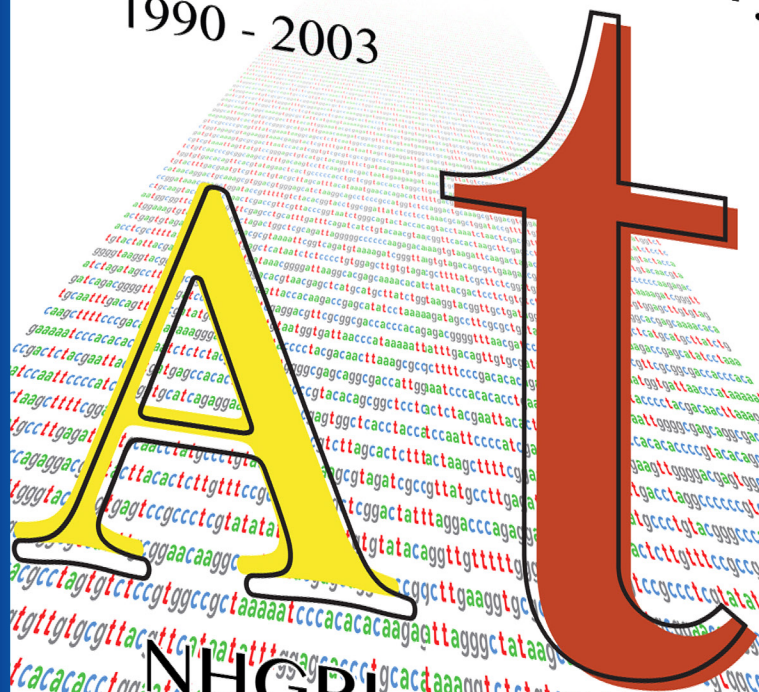
10th Anniversary

10th Anniversary Human Genome Project 1990 - 2003



NHGRI

Human Genome Project 1990 - 2003 10th Anniversary



NHGRI



Smithsonian NHGRI Genome Exhibition

Exhibition Advisory Board

Exhibition Announcement

HGP10

The Genomics Landscape a Decade After the Human Genome Project

Share Print



In April 2003, the International Human Genome Project (HGP), led in the United States by the National Institutes of Health, was completed ahead of schedule and under budget. For the first time, anyone could freely read the fundamental instruction set needed to make a human body. But much more still must be learned about life's operating system in order for it to be fully applied to human health.

"The Human Genome Project has had an incalculable impact on science over the past decade," said Eric D. Green, M.D., Ph.D., director of NHGRI, who was recruited to NIH early on in the 13-year project. "I am especially pleased that our varied events highlight genomics in so many ways—because the ongoing work of human genetics and genomics benefits all of us."

Beginning in February 2013, the National Human Genome Research Institute (NHGRI), the NIH institute that spearheaded the HGP, will

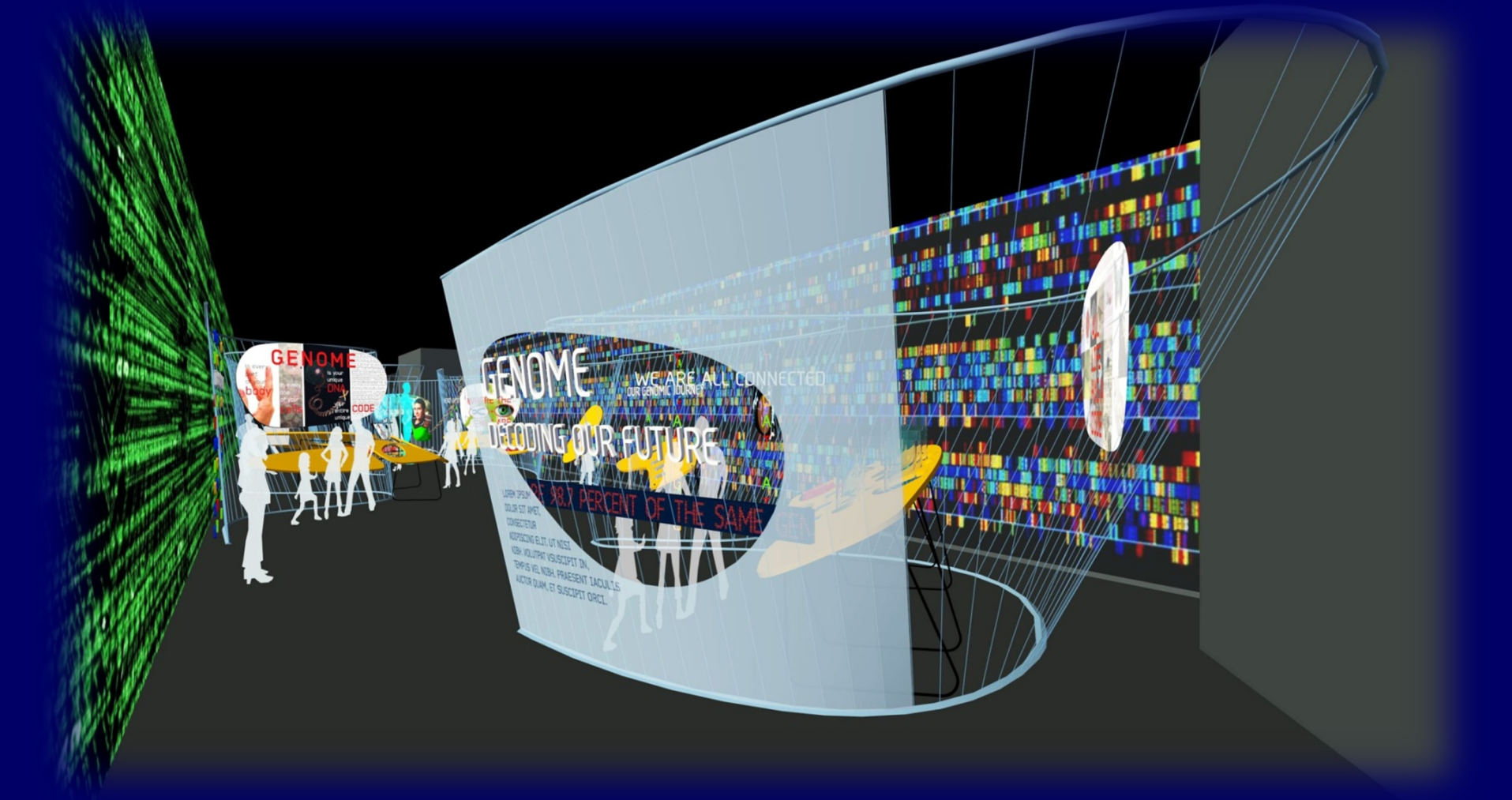
celebrate the 10th anniversary of the completion of the Human Genome Project with a series of stimulating talks, a thought-provoking symposium and a fascinating interactive exhibit to mark the project's 10-year anniversary and to reflect on the HGP's revolutionary influence on biomedicine.

Upcoming Events

- [HGP 10th Anniversary Seminar Series](#)
February - March, 2013
- [HGP 10th Anniversary Symposium Agenda](#)
April 25th, 2013
- [Smithsonian NHGRI Genome Exhibition](#)
NHGRI's official exhibition page for the HGP 10th Anniversary

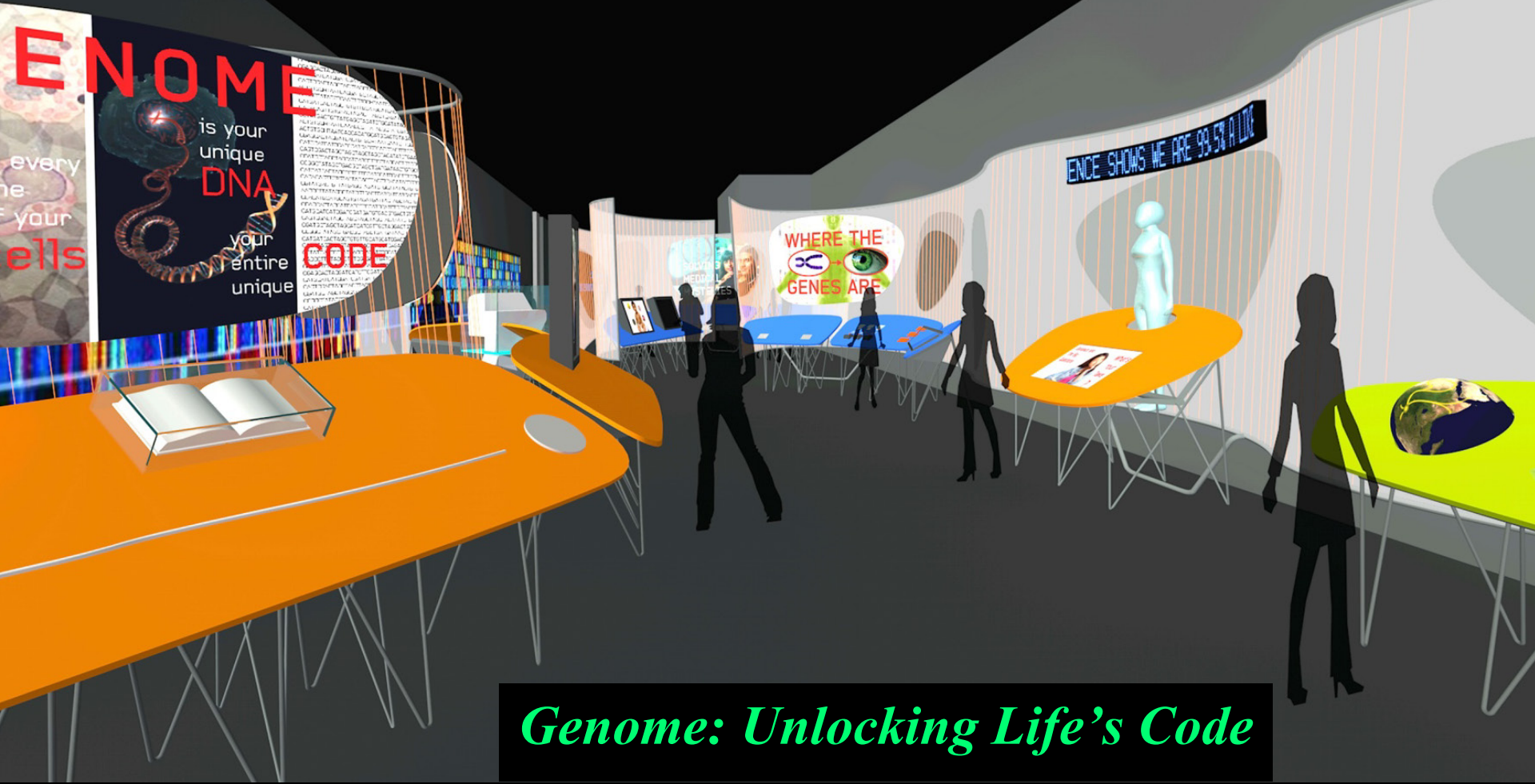
Background Resources

NHGRI-Smithsonian Genome Exhibition



Opening in June 2013

Smithsonian National Museum of Natural History



Genome: Unlocking Life's Code

Hot tickets for 2013 in science and art

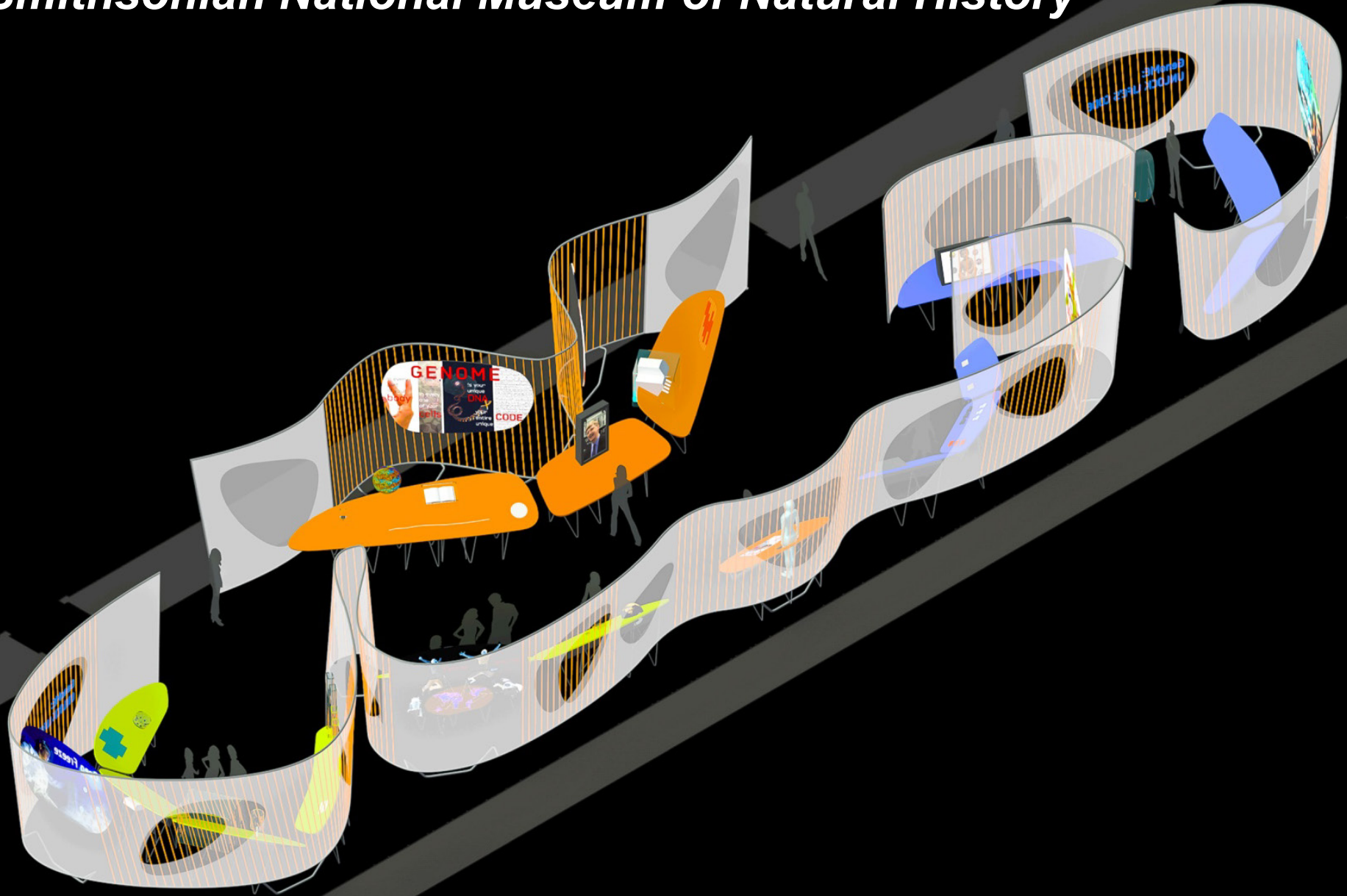
Human Genome exhibition

*Smithsonian National Museum of Natural History,
Washington DC
June 2013 to June 2014*

In a year that sees both the 60th anniversary of Francis Crick and James Watson's elucidation of DNA's structure and the 10th anniversary of the human genome's complete decoding, the Smithsonian Institution is pulling out all the stops. For this exhibition, its natural history museum joins forces with the National Human Genome Research Institute in Bethesda, Maryland, to explore what the genome is, what it tells us and how this information could revolutionize health care and our understanding of our place in the world. After its time on the National Mall, the show will travel around North America.

Opening in June 2013

Smithsonian National Museum of Natural History





NATIONAL HUMAN GENOME RESEARCH INSTITUTE



***Advancing human health
through genomics research***

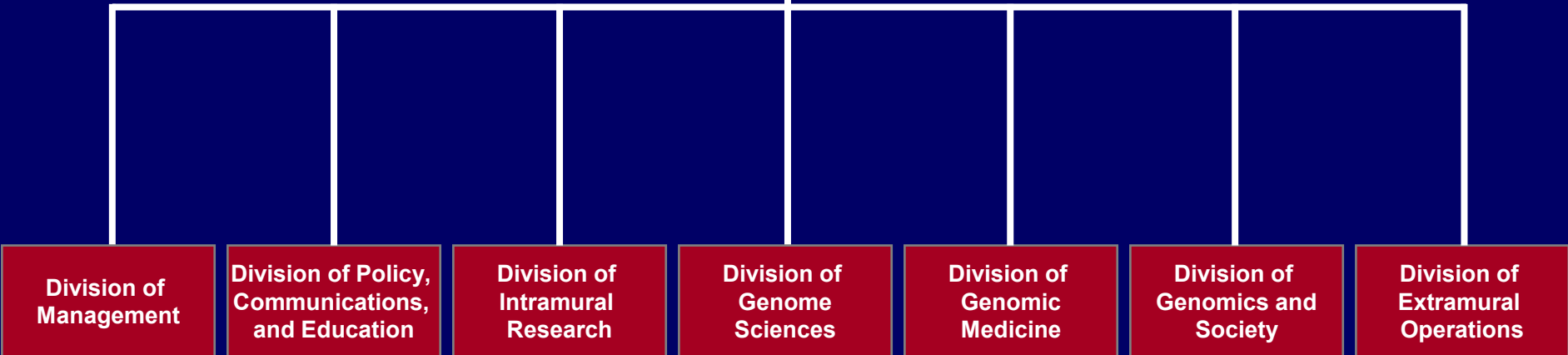
NHGRI Reorganization



***October 1, 2012:
Implementation of New
Organizational Structure for NHGRI***

(see genome.gov/reorg)

New NHGRI Organizational Structure



Extramural Research Program