Moving Genomics Forward:

Emerging and evolving policy issues

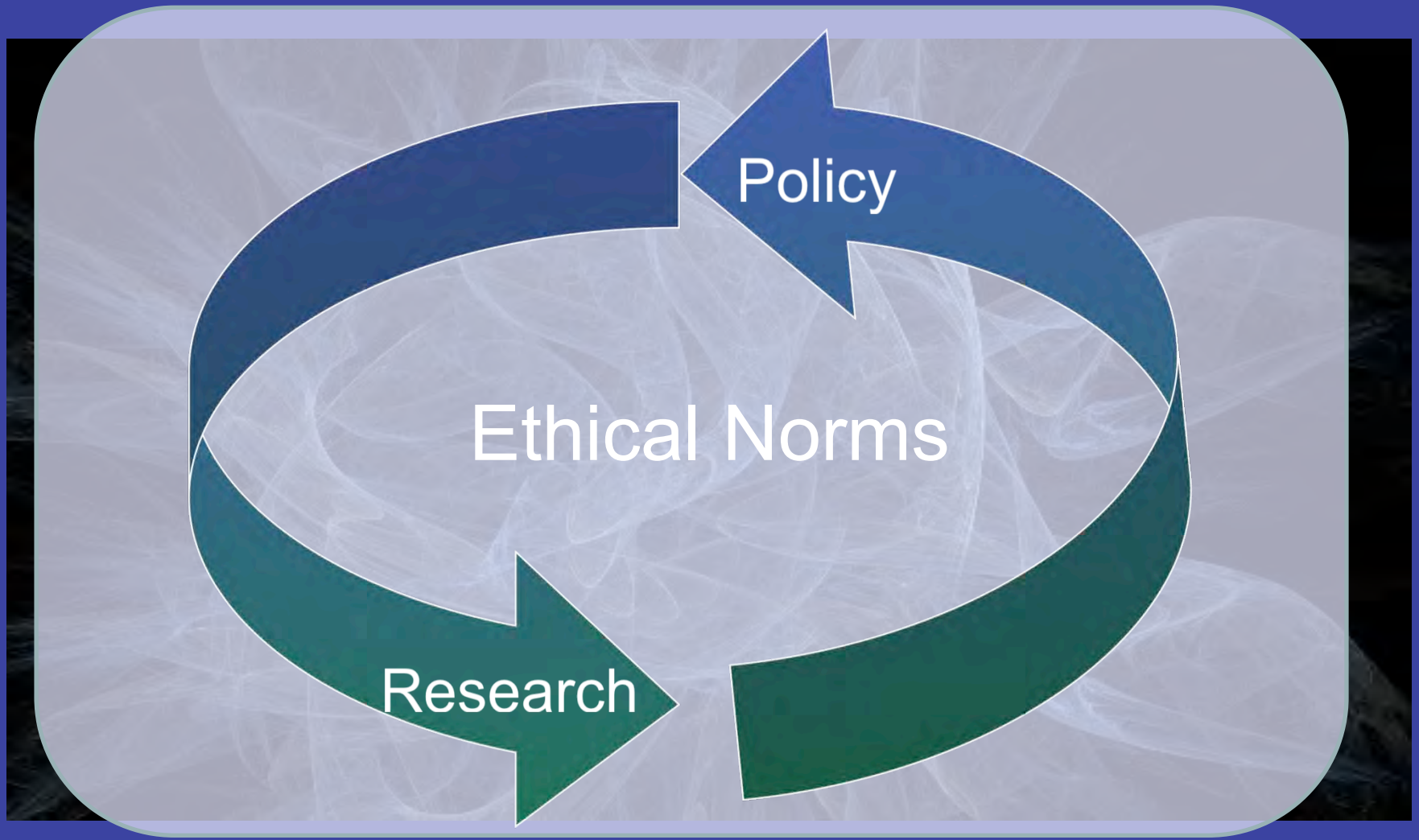
Laura Lyman Rodriguez, Ph.D.

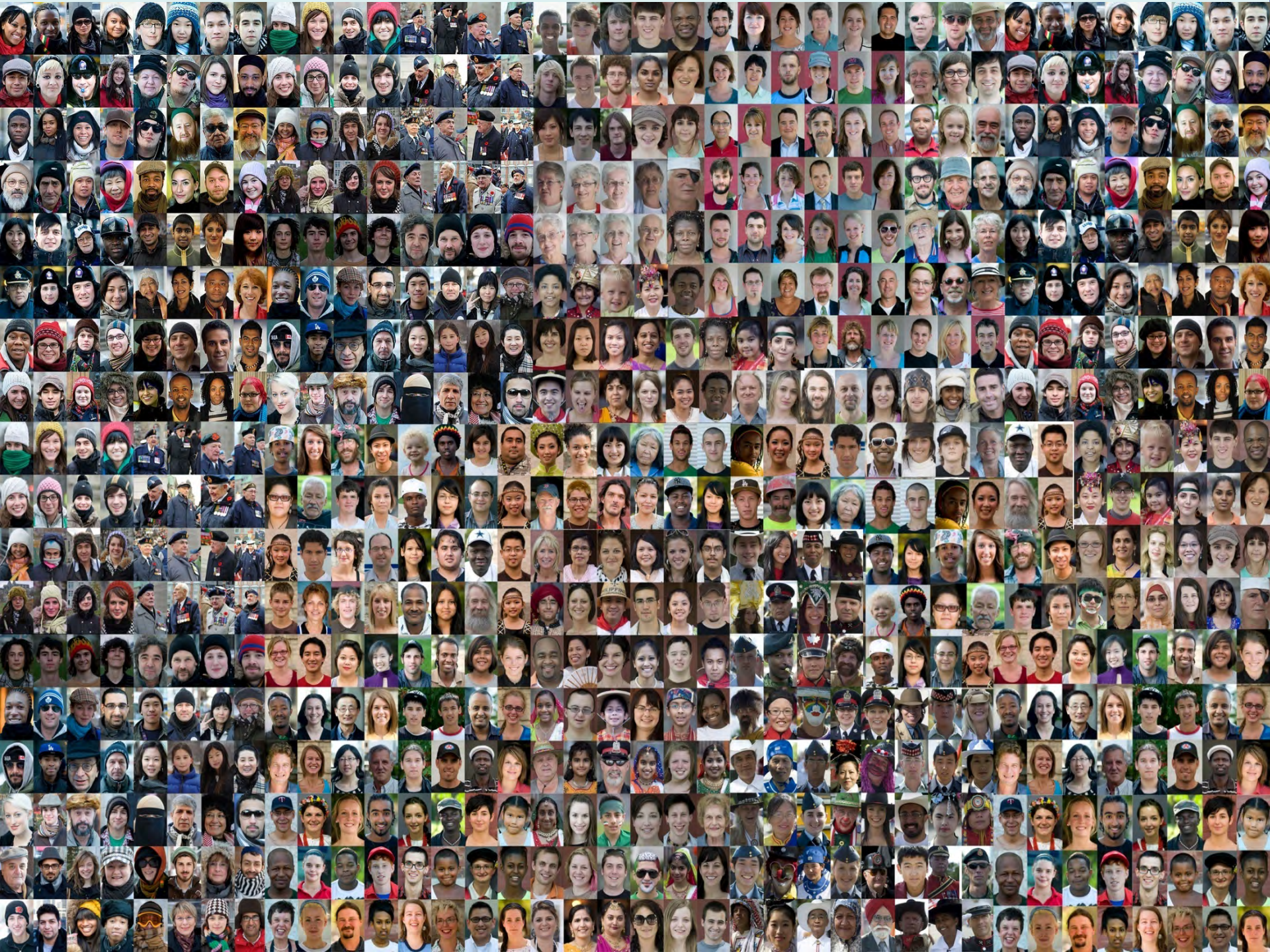
National Human Genome Research Institute

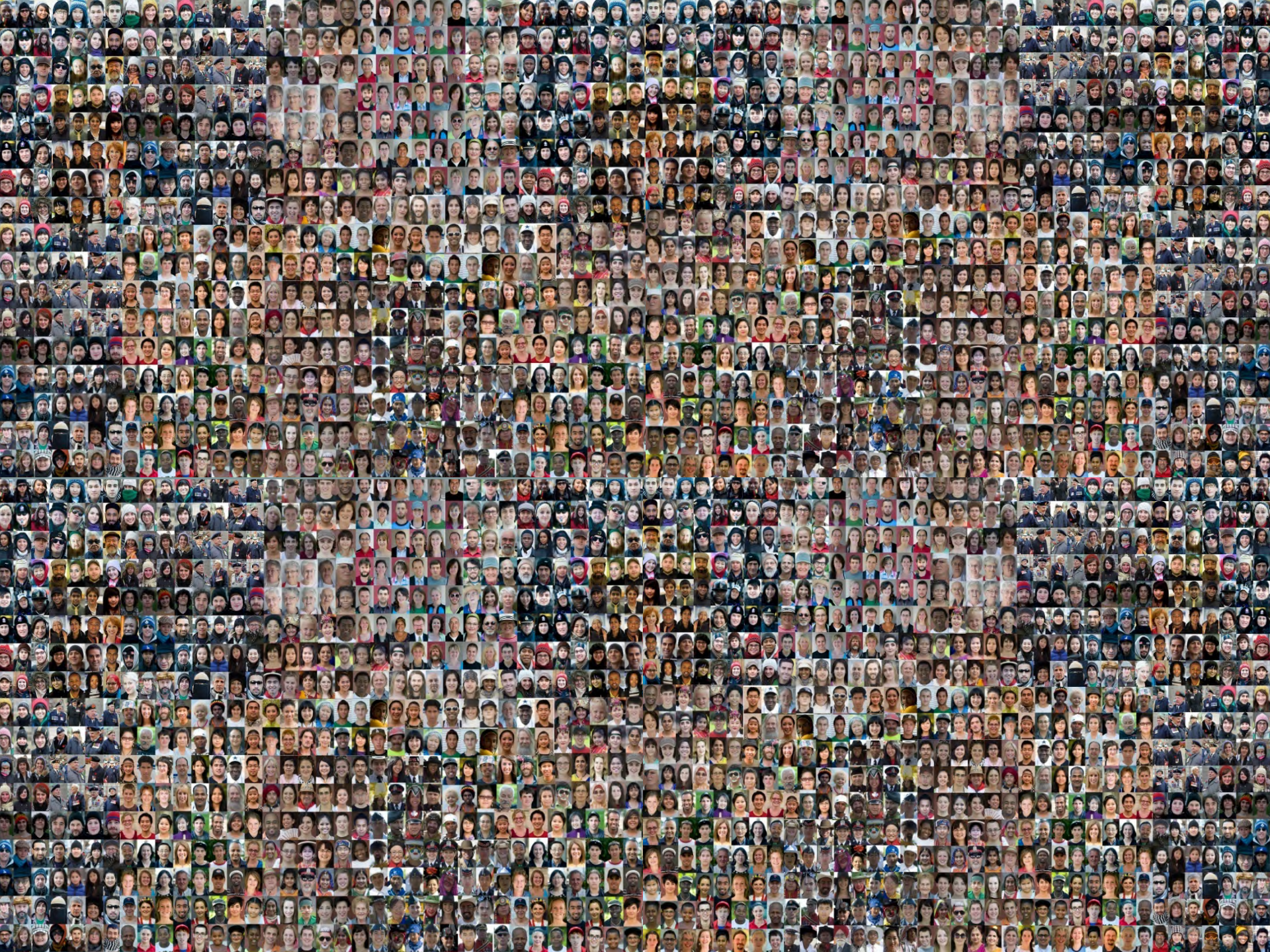
Science Reporters' Workshop

June 13, 2013

Aim: Iterative Dialog and Process

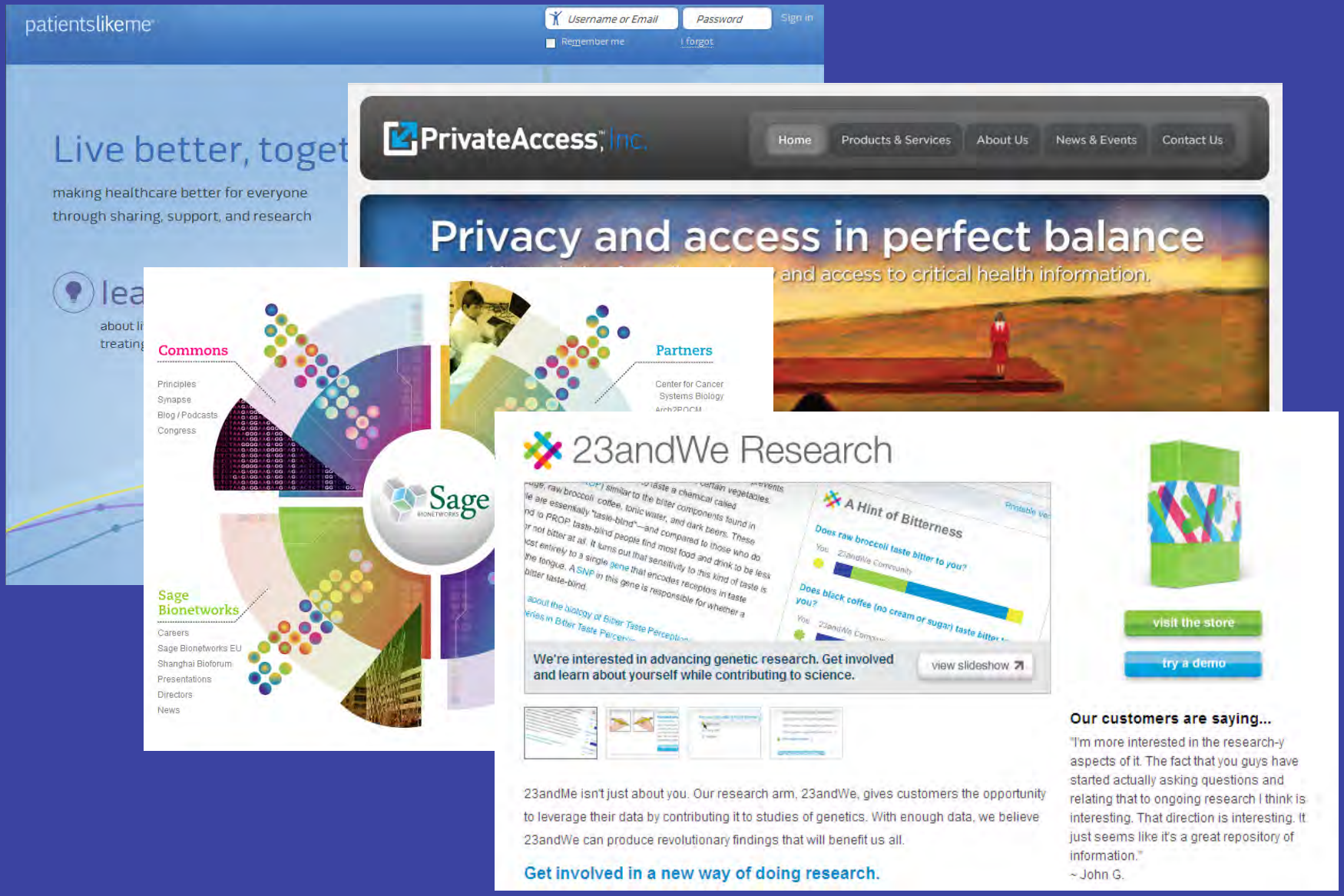






Direct Access to Technology

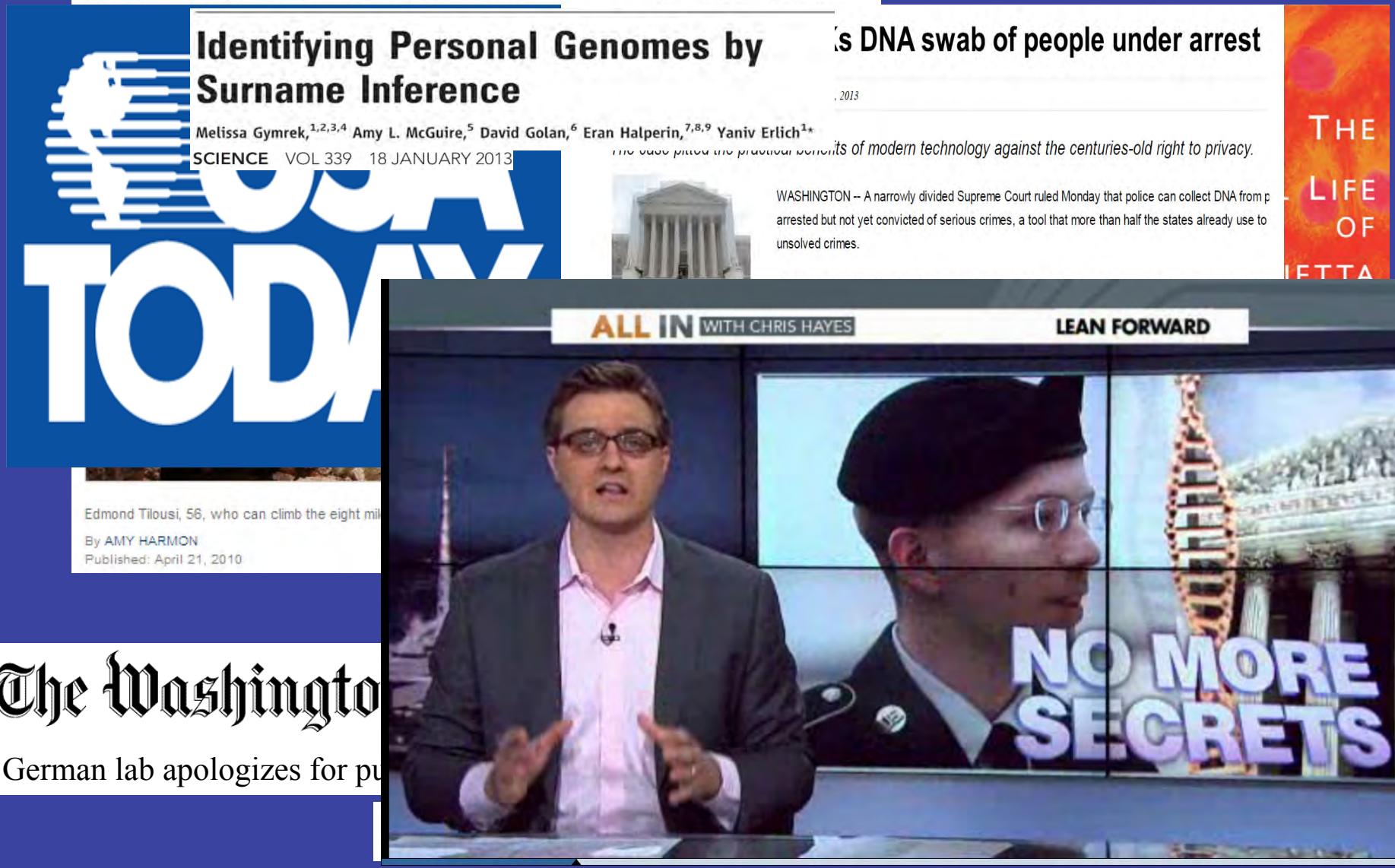
Participants Driving Research Momentum



Moving from Anecdote(s) to Common Practice

Key Policy Issues *(selected)*

The View from the Public



Time to Review and Reimagine

Home > News & Publications:News & Multimedia > International partners describe global alliance to enable secure sharing of genomic and clinical data

International partners describe global alliance to enable secure sharing of genomic and clinical data

By Broad Communications, June 4th, 2013

Over 70 leading health care, research, and disease advocacy organizations that together involve colleagues in over 40 countries have taken the first steps to form an international alliance dedicated to enabling secure sharing of genomic and clinical data. The cost of genome sequencing has fallen one-million fold, and more and more people are choosing to make their genetic and clinical data available for research, clinical, and personal use. However, interpreting these data requires an evidence base for biomedicine that is larger than any one party alone can develop, and that adheres to the highest standards of ethics and privacy. These organizations recognize that the public interest will be best served if we work together to develop and promulgate standards (both technical and regulatory) that make it possible to share and interpret this wealth of information in a manner that is both effective and responsible.

both societal goods.

NEW: Genomic Data Sharing (GDS) Policy

Notice on Development of Data Sharing Policy for Sequence and Related Genomic Data

Notice Number: NOT-HG-10-006

Key Dates
Release Date: October 19, 2009

Issued by
National Human Genome Research Institute (NHGRI), (http://www.genome.gov/)

Purpose

The purpose of this Notice is to inform the research community of plans by the National Institutes of Health (NIH) to:

1. Update data sharing policies for NIH supported research, including extramural and intramural projects, involving sequence and related genomic data obtained with advanced sequencing technology (*e.g.*, medical resequencing data, sequence data from non-human species, including microorganisms, transcriptomic and epigenomic data, as well as data needed for interpretation, including associated clinical, other phenotype and metadata, such as supporting study documents and methodologies);
2. Encourage investigators and IRBs to consider the potential for broad sharing of sequence and related genomic data in developing informed consent processes and documents for such studies involving human sequence data; and,
3. Communicate the agency's intent and current underlying considerations related to developing a policy pertaining to the deposition of these large datasets into centralized databases, such as the GenBank Short Read Archive (SRA) or the Database of Genotypes and Phenotypes (dbGaP), so that they are available as broadly and rapidly as possible to a wide range of scientific investigators.

- Extension of the rationale for data sharing policies
- Accessibility of whole genome sequencing and other genomic technologies advancing rapidly
- Consistent approach with regard to consent
- Complexity of data and project types much greater

Updating the Common Rule

- Propose written consent for all uses of existing research samples (short forms, broad consent OK; only applies prospectively)
- Biospecimens considered inherently identifiable based on genetic data – risk classified as "informational"

Advanced Notice of Proposed Rule Making (ANPRM)

Read more about the July 22, 2011 ANPRM for changes under consideration to the Common Rule.

These changes, the most extensive since the Department of Health, Education, and Welfare published proposed rules for the protection of human subjects involved in research on August 14, 1979, are **available for public comment until September 26, 2011**.

Access: Gene Patents

- Potential implications for research and clinical access
- Decision out this morning!!

Access: Coverage of Genetic Testing

- Choice
- Ability to make the choice

The New York Times

The New York Times

May 14, 2013

My Medical Choice

By ANGELINA JOLIE

LOS ANGELES

MY MOTHER fought cancer for almost a decade and died at 56. She held out long enough to meet the first of her grandchildren and to hold them in her arms. But my other children will never have the chance to know her and experience how loving and gracious she was.

We often speak of "Mommy's mommy," and I find myself trying to explain the illness that took her away from us. They have asked if the same could happen to me. I have always told them not to worry, but the truth is I carry a "faulty" gene, BRCA1, which sharply increases my risk of developing breast cancer and ovarian cancer.

My doctors estimated that I had an 87 percent risk of breast cancer and a 50 percent risk of ovarian cancer, although the risk is different in the case of each woman.

Only a fraction of breast cancers result from an inherited gene mutation. Those with a defect in BRCA1 have a 65 percent risk of getting it, on average.

Once I knew that this was my reality, I decided to be proactive and to minimize the risk as much I could. I made a decision to have a preventive double mastectomy. I started with the breasts, as my risk of breast cancer is higher than my risk of ovarian cancer, and the surgery is more complex.

Readiness: Need for Education

Bloomberg

September 12, 2012

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Fumbled DNA Tests Mean Peril for Breast-Cancer Patients

By Robert Langreth - Sep 10, 2012

Debbie McCarron was prepared to get both of her breasts taken off if a blood test in December 2006 revealed she carried a gene that vastly increases the risk of breast cancer. Having survived the disease five years earlier, she didn't want to risk getting it again.

To her relief, her oncologist told her the test, done by Myriad Genetics Inc. (MYGN), had come back negative, "just like I knew it would," McCarron recalls her doctor saying.

He was wrong. The results, in fact, were positive. McCarron didn't learn this, though, until July 2009, more than two years later, when a genetic counselor reviewed the test following McCarron's surgery to remove a new malignant breast tumor. Since then, her oncologist, Haresh Jhangiani, told Bloomberg he isn't clear about what happened.

"I don't think she was positive. Was she positive?" the doctor said. "I would not tell her it was negative if the test was positive, there must be something more to it."

McCarron, now 50, was devastated when she found out. "The whole world crumbled down on me that day," the Huntington Beach, California, woman said in an interview. "I never would have gotten that second cancer."

oped 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; how-

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]

Building Resources

Moving into the Clinic

In summary…we want to avoid


National Human Genome
Research Institute
Thank you!
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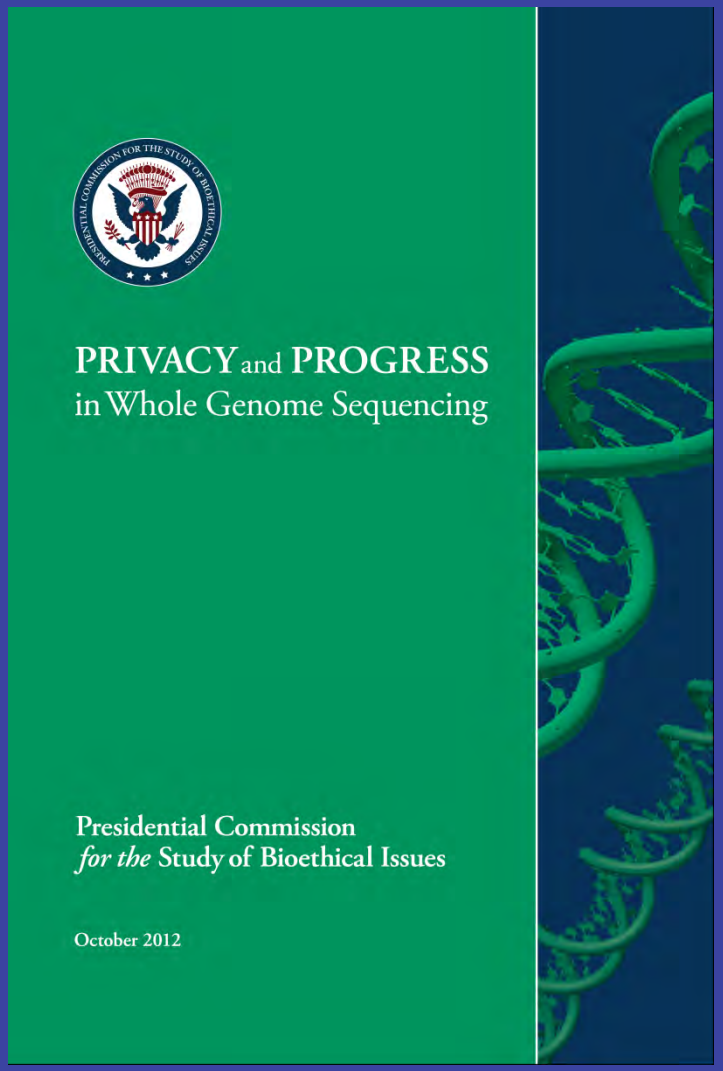
Advancing human health
through genomics research

Putting the Pieces Together

- **Scientific Design**
  - Research aims and objectives
- **Ethics**
  - Guiding principles
- **Policy**
  - Applicable laws and regulations

Presidential Bioethics Commission



Recs 1: Clear and consistent policies

Recs 2: Standards for security & access

Recs 3: Informed consent

Recs 4: Exchange information between research and clinical domains

Rec 5: Access for all citizens

On-going: Incidental Findings

SUPREME COURT OF THE UNITED STATES

No. 12–398

ASSOCIATION FOR MOLECULAR PATHOLOGY, ET AL., PETITIONERS *v.* MYRIAD GENETICS, INC., ET AL.

ON WRIT OF CERTIORARI TO THE UNITED STATES COURT OF APPEALS FOR THE FEDERAL CIRCUIT

[June 13, 2013]

JUSTICE THOMAS delivered the opinion of the Court.

Respondent Myriad Genetics, Inc. (Myriad), discovered the precise location and sequence of two human genes, mutations of which can substantially increase the risks of breast and ovarian cancer. Myriad obtained a number of patents based upon its discovery. This case involves claims from three of them and requires us to resolve whether a naturally occurring segment of deoxyribonucleic acid (DNA) is patent eligible under 35 U. S. C. §101 by virtue of its isolation from the rest of the human genome. We also address the patent eligibility of synthetically created DNA known as complementary DNA (cDNA), which contains the same protein-coding information found in a segment of natural DNA but omits portions within the DNA segment that do not code for proteins. For the reasons that follow, we hold that a naturally occurring DNA segment is a product of nature and not patent eligible merely because it has been isolated, but that cDNA is patent eligible because it is not naturally occurring. We, therefore, affirm in part and reverse in part the decision of