

# Moving Genomics Forward:

## Emerging and evolving policy issues

Laura Lyman Rodriguez, Ph.D.

National Human Genome Research Institute

Science Reporters' Workshop

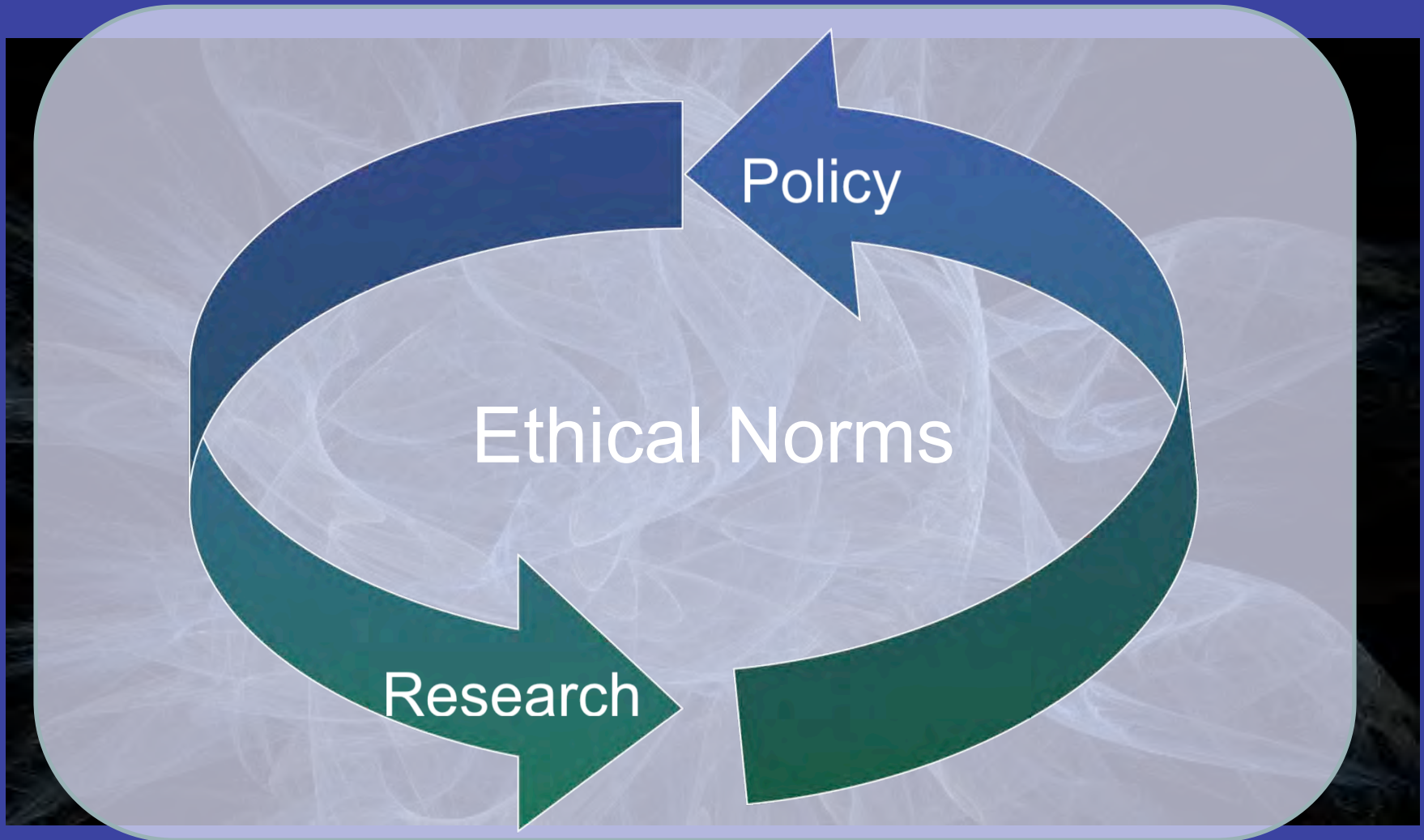
June 13, 2013

NATIONAL  
HUMAN GENOME  
RESEARCH INSTITUTE



# Aim: Iterative Dialog and Process

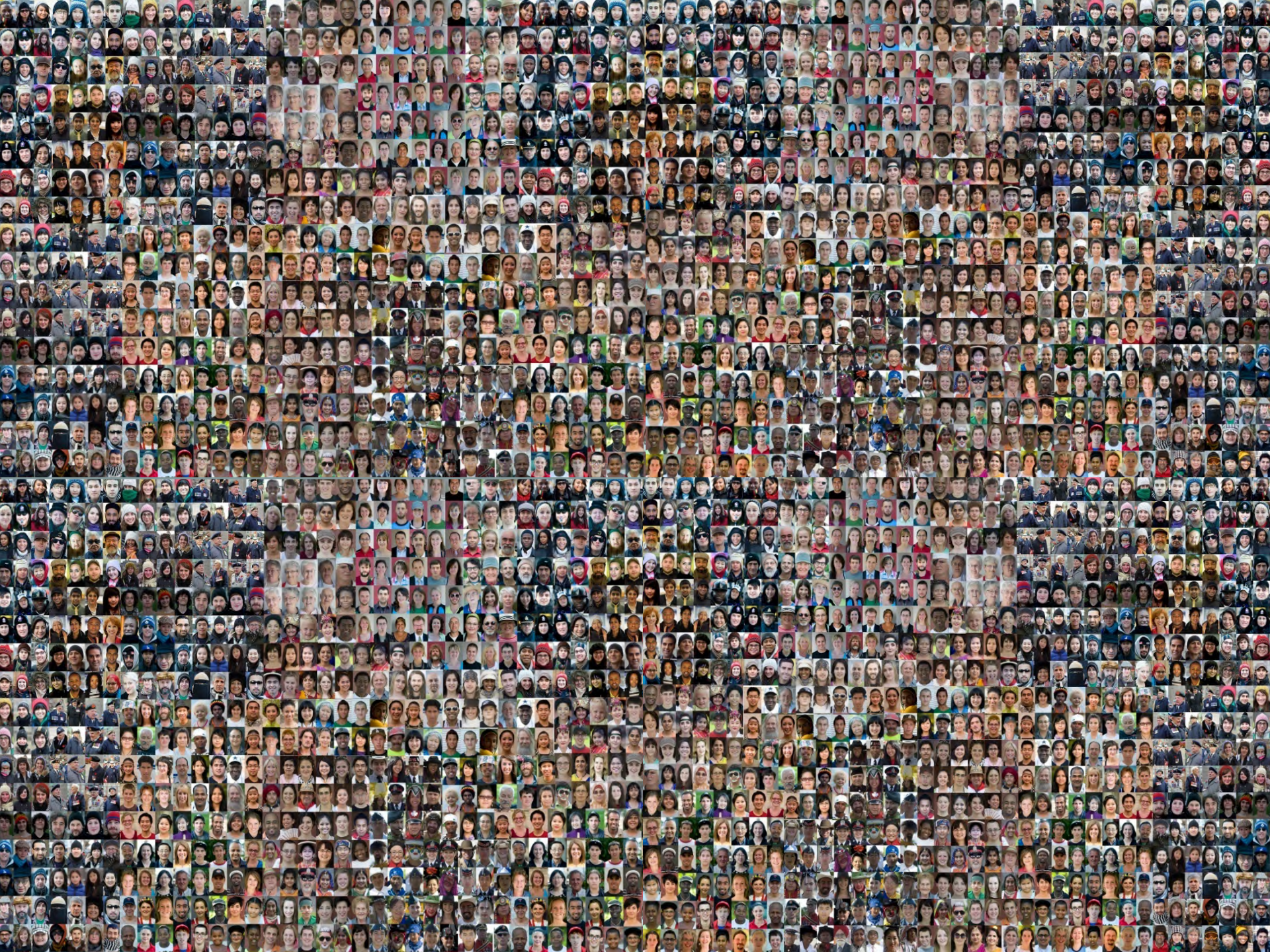
---



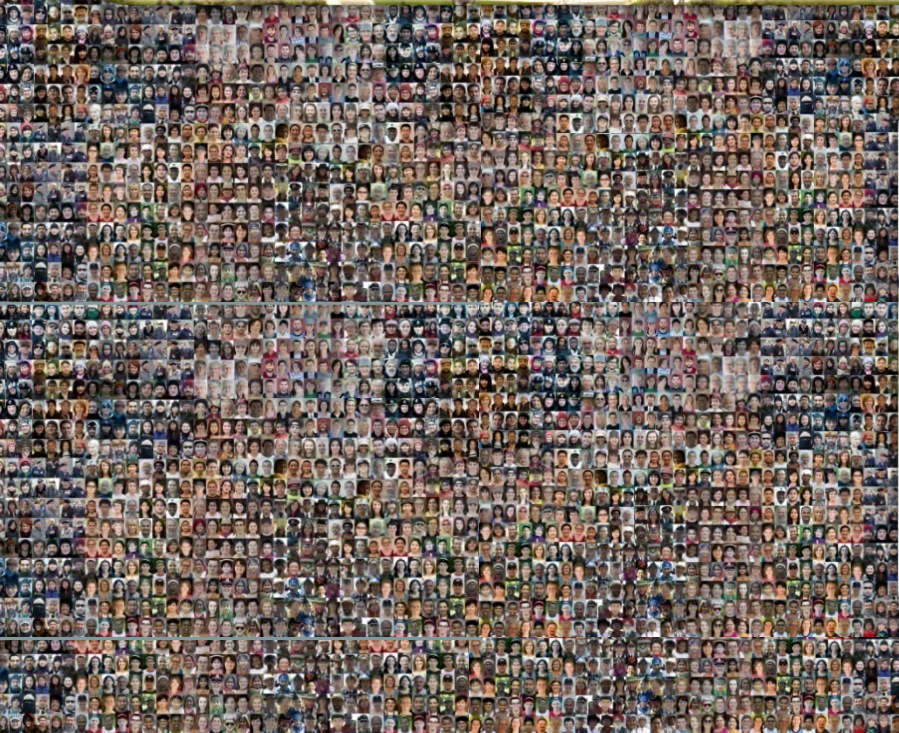
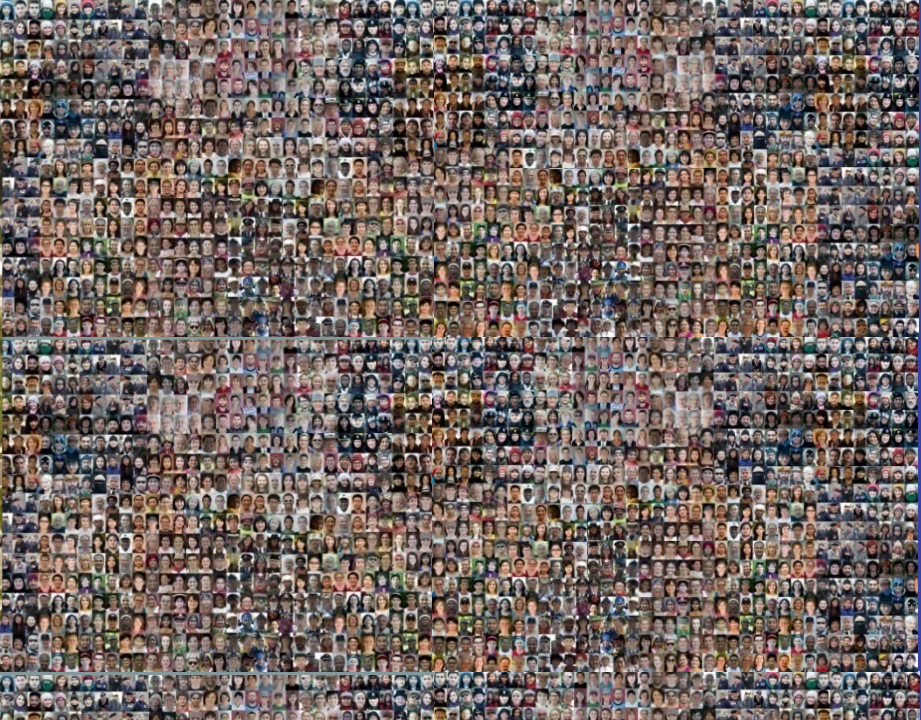






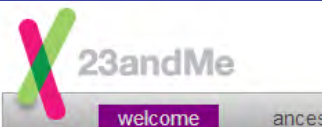








# Direct Access to Technology



## Ready to discover your family story?

Simply start with yourself and we'll do the searching for you.

**14-Day Free Trial**  
GIVE ME ACCESS

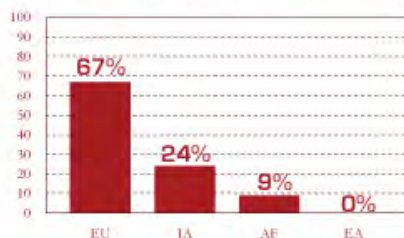


*Kathryn Baker  
Toledo, OH*

### Kathryn discovered her AncestrybyDNA™

*"I was surprised to learn that I'm as much as 24% Indigenous American. It must come from a closer relative than I thought. I can't wait to share this with my family."*

Bar Graph of Results



- 67% European
- 24% Indigenous American
- 9% Sub-Saharan African
- 0% East Asian

[Submit Your Testimonial](#)

### : Additional Ancestry DNA Tests Available :



- **Paternal Lineage (Y-STR) Testing**  
This type of DNA testing determines what migration routes your ancient paternal ancestors took.

[Go to page](#)

- **Maternal Lineage (mtDNA) Testing**  
This type of DNA testing determines what migration routes your ancient maternal ancestors took.

[Go to page](#)



# Participants Driving Research Momentum

patientslikeme™

Username or Email Password Sign in

Remember me I forgot

Live better, together

making healthcare better for everyone through sharing, support, and research

PrivateAccess™, Inc.

Home Products & Services About Us News & Events Contact Us

Privacy and access in perfect balance

and access to critical health information.

Learn about life

treatings

Commons

- Principles
- Synapse
- Blog / Podcasts
- Congress

Partners

Center for Cancer Systems Biology

Sage BIONETWORKS

Sage Bionetworks

- Careers
- Sage Bionetworks EU
- Shanghai Bioforum
- Presentations
- Directors
- News

23andWe Research

A Hint of Bitterness

Does raw broccoli taste bitter to you?

Does black coffee (no cream or sugar) taste bitter to you?

We're interested in advancing genetic research. Get involved and learn about yourself while contributing to science.

visit the store

try a demo



23andMe isn't just about you. Our research arm, 23andWe, gives customers the opportunity to leverage their data by contributing it to studies of genetics. With enough data, we believe 23andWe can produce revolutionary findings that will benefit us all.

Get involved in a new way of doing research.

Our customers are saying...

"I'm more interested in the research-y aspects of it. The fact that you guys have started actually asking questions and relating that to ongoing research I think is interesting. That direction is interesting. It just seems like it's a great repository of information."

~ John G.



# Moving from Anecdote(s) to Common Practice





# Key Policy Issues (selected)

---

Participant Autonomy

Oversight &  
Regulation

Access

Privacy

Healthcare Readiness





# The View from the Public

## Identifying Personal Genomes by Surname Inference

Melissa Gymrek,<sup>1,2,3,4</sup> Amy L. McGuire,<sup>5</sup> David Golan,<sup>6</sup> Eran Halperin,<sup>7,8,9</sup> Yaniv Erlich<sup>1\*</sup>

SCIENCE VOL 339 18 JANUARY 2013

## Police DNA swab of people under arrest

2013

The case pitted the practical benefits of modern technology against the centuries-old right to privacy.



WASHINGTON -- A narrowly divided Supreme Court ruled Monday that police can collect DNA from people arrested but not yet convicted of serious crimes, a tool that more than half the states already use to solve unsolved crimes.



Edmond Tilousi, 56, who can climb the eight miles

By AMY HARMON

Published: April 21, 2010



## The Washington Post

German lab apologizes for pu



# 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**.

Human  
Subjects  
Protections

UPDATE



# 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

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 [Print](#) [Email](#) [Share](#)

## 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."

opened by the National Human Genome Research Institute suggests more discoveries are likely to occur over the next few decades.<sup>1</sup> 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.<sup>8</sup>



# Building Resources

 GENETICS/GENOMICS COMPETENCY CENTER FOR EDUCATION

[Home](#) [My Resources](#) [Help](#) [About the Project](#) [Survey](#)

 [Competencies & Guidelines](#)

## Search Educator Resources:

Genetic Counselor



[Search](#)

Nurse




[Search](#)

Physician Assistant



[Search](#)

 [Submit Your Resources](#)

ite:

Genetics resources to use in your classroom

locate courses-enhance your class content with

genetic/genomic resources for [Nurses](#), [Physician Assistants](#), and [Genetic Counselors](#).

2. [Review](#) the Competency Guidelines and Curriculum Map

Need to match your classroom genomics teaching to competencies? View the guidelines and the curriculum map to find the right resources to download.

3. [Share](#) resources, activities, and assessments to be included on this site

Do you have activities, resources or assessments you would like to share? Submit websites,





# Moving into the Clinic

TIBTEC-1075; No. of Pages 3

ARTICLE IN PRESS

Forum: Science & Society

Cel  
PRESS

Inc

POINT-COUNTERPOINT

POINT-COUNTERPOINT

Patient Autonomy on

On M  
practi  
Clinic  
condi  
and r  
recon  
recon  
consi

Patie  
The A  
seque  
patho  
is like  
couns  
canno  
expec  
nature  
etc.) a  
return  
physi  
gener

The r  
near c  
comm  
predi  
unkn  
patho  
and w  
unkn  
with c

Incid

Some  
geneti  
Acad  
child  
diseas  
be or



tal  
exome

second, they do not  
encos in reporting  
guardians) should  
er they desire the  
lation specifically

Working Group to  
s with information  
find these recom-  
tical and a practi-  
eddefine incidental  
he implication that  
Iden by physicians  
ported in modern

incidental findings'  
ags' in the past, the  
erent meaning: 'we  
er to indicate the  
thogenic or likely  
are not apparently  
which the sequenc-  
contradicts com-  
cidental findings.  
diagnostically rele-  
referred to as 'ge-  
dedicatively test-  
y history of serious  
rds genetic screen-  
been supported in  
in children [4]. In  
s turn the occasion  
for the test, into a

s. First, the recom-  
that the extra test-  
ratories that 'add  
their sequencing  
payers have sup-

some of us (A.L.M., R.C.G.) were members of the ACMG Working Group that wrote the recommendations, this paper does not represent the official views of the Working Group or the ACMG.

of Obstetrics and Gynecology, Feinberg School of Medicine, Northwestern University, Chicago, IL 60611, USA.

\*Corresponding author. swolf@umn.edu

search  
a radio  
pected

variants as listed in the recommendations and include

Corresponding author: Alyse, M. (megan2@stanford.edu).  
Keywords: whole genome sequencing; returning results; incidental findings.

ported genetic testing only in the event of symptomatic  
evidence or an established family risk [5]. It is unclear  
whether payers will cover the cost of predictive testing for  
conditions when neither of these conditions apply. Indeed,

# In summary...we want to ~~seek~~

---







# NATIONAL HUMAN GENOME RESEARCH INSTITUTE

**Thank you!**

[laura.rodriguez@nih.gov](mailto:laura.rodriguez@nih.gov)



*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

---



## PRIVACY and PROGRESS in Whole Genome Sequencing

Presidential Commission  
*for the Study of Bioethical Issues*

October 2012



**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**



## Opinion of the Court

NOTICE: This opinion is subject to formal revision before publication in the preliminary print of the United States Reports. Readers are requested to notify the Reporter of Decisions, Supreme Court of the United States, Washington, D. C. 20543, of any typographical or other formal errors, in order that corrections may be made before the preliminary print goes to press.

**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