

# Shaping Public Health Policy in the Age of Genomics

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## A Legislative Briefing Book



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Prepared by PMPH 507 Students  
*Genomics and Public Health Policy*  
Oregon Masters of Public Health Program  
Oregon Health and Sciences University  
Oregon State University  
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## Foreword

Genomic science and associated technologies are facilitating an unprecedented rate of discovery of novel insights into the relationship between human genetic variation and health. Genomics is a new discipline that studies the functions and interactions of all the genetic material in the genome, including interaction with environmental factors.

Advances in genome research in the last 20 years have revolutionized knowledge of the role of inheritance in health and disease. It is now known that DNA and RNA determine not only the cause of single-gene disorders, which affect millions of people worldwide, but also predispositions (“susceptibilities”) which are based on new technologies which allow researchers to examine genetic mutations at the functional genomic unit level and to better understand the significance of environmental factors such as chemical agents, nutrition or personal behavior in relation to the causation of diseases like cardiovascular diseases, allergies, cancer, psychiatric disorders or infectious diseases.

These rapid advances in genomics and accompanying technologies are triggering a shift in the comprehension of health and disease as well as in the understanding of new approaches to prevention and therapy. This emerging knowledge also opens a window of opportunity for other sciences such as law or ethics to develop a normative framework even while the science of genomics is being widely applied.

Many questions arise when genomic knowledge is translated to the timely and responsible practice of medicine and public health. Indeed these questions, and many more, lie at the heart of the chapters of this legislative briefing book, each created by the students and teaching faculty participating in the 2011 spring term online course, ***Genomics and Public Health Policy: Current Issues and Trends:***

- What are the technologies by which medicine of the future will manage an individual’s personal health instead of managing a patient’s disease? [Chapter 1]
- Will our current health care system of balancing risks and benefits to patients fail if we have genomic data (of still imperfect quality) about how patients are likely to respond to different therapies? [Chapter 1]

- Will genomics lead to a single payer system because of challenges in maintaining privacy and difficulties of insurance companies in determining which individuals to insure at what rates? [Chapter 2]
- Will genomic knowledge result in unbelievable advances in health care but widen the health disparities among groups who have persistently experienced historical trauma, social disadvantage or discrimination, and systematically worse health or greater health risks than more advantaged social groups? [Chapter 2]
- Who gets access to the new genomic technologies? [Chapter 2]
- Are new emerging genomic reproductive technologies paving the way to a “new eugenics?” [Chapter 3]
- Should universal carrier screening become a routine part of medical care? [Chapter 3]
- What psychological toll might there be to discovering you are at risk for certain diseases like Alzheimer’s? Do you even want this information? [Chapter 4]
- What do genes tell us about ourselves, and about our potential children? How much is clear, how much is uncertain, and how do we know the difference? [Chapter 4]
- Genomic knowledge is generated by research paid for by tax dollars, but does that mean that companies should be allowed to provide it directly to consumers, with no medical personnel acting as a filter? [Chapter 4]
- Should tissue samples taken from AIDS patients be made available in a public database, so that other federally financed researchers can use them for additional studies? Should there be policies in place to regulate this kind of research activity? [Chapter 4]
- Many experts are advocating that all newborns have a complete genome analysis done so that preventive measures and preemptive medicine can begin early in life. How will this astonishing achievement play out in our lives? Will our privacy be protected? Will we be pressured, by insurance companies or by our employer, to get our genome sequenced? And will the government or the medical establishment come between you and your genome? [Chapter 5]

- How will research currently underway on the Human Microbiome Project (an international effort to catalogue the 1 quadrillion microbial cells that live in and on our bodies) alter our conceptions of health and disease and ultimately determine the kinds of policies that will be needed to regulate the use of products (e.g., probiotics and drugs) generated by this research? [Chapter 5]
- How will the emerging science of epigenomics allow us to create policies designed to specifically address the decreased life span, non-alcohol-induced fatty liver disease, increased cardiovascular disease, increased incidence of stroke and type 2 diabetes currently estimated to affect over 65 million adult Americans suffering from morbid obesity? [Chapter 6]
- How can policy makers, the public, and technical experts engage with one another as partners to create responsible and effective upstream genome policy for the common good? [Chapter 7]

Although public health genomics is still in its infancy, rapidly advancing science and patient and public expectations require a strategic approach to the assessment, development and implementation of genomic policies. The development of such policies must begin now, given the pace of genomic science, particularly in view of the need to educate and train a whole cohort of practitioners—and policy makers-- in the principles of genetics and molecular science.

The science of genomics is a fantastic gift to humankind if used wisely. But the greatest gifts can be the most socially and ethically abused; the best protection remains awareness, vigilance, and well-informed, evidence-based policy.

## **The Course**

More than two years in development, the course, *Genomics and Public Health Policy: Issues and Trends* was targeted to OMPH students, but also open to selected post-baccalaureate and graduate students in the health sciences at Oregon Health and Science University, Portland State University, and Oregon State University, and members of the public health workforce.

The course was designed to help students understand how systems work instead of how the isolated genes make things happen on their own, to seek to understand genetic factors that contribute to individual and group variation in disease risk, and to translate that knowledge into actions and practice reflect in the core functions of public health.

The course featured asynchronous critically reflective Weblog discussions of assigned primary research papers in genomic science, ethics and policy, and genomics forums with genomic researchers, bioethicists, and policy makers.

\*[Course overview, syllabus, and evaluation criteria](#)

## Teaching Faculty

**Gregory Fowler, PhD.**  
**Course Instructor**



Gregory Fowler is a Senior Research Associate in the Center for Health Policy Studies, School of Community Health at Portland State University and Associate Clinical Professor, Department of Public Health and Preventive Medicine, Oregon Health and Science University. In 1998 he co-founded [Geneforum](#), a non-profit organization committed to promoting dialogue at the intersection of genetics, ethics, and public values. His interest areas include science education, the societal and ethical implications of science and technology, and deliberative democracy. Past recognitions include an appointment as a Fellow in the Science and Engineering Diplomacy Program of the American Association for the Advancement of Science, and the recipient of an Ethics and Values in Science and Technology Individual Award of the National Science Foundation and the National Endowment for the Humanities to study the social and ethical implications of genetic engineering. He is currently a standing member of the Oregon Legislature's Advisory Committee on Genetic Privacy and Research mandated to create opportunities for public education and input on issues of genetic privacy and research, and an elected Fellow of the World Academy of Art and Sciences.

**Michael Flower, PhD.**  
**Course Co-Instructor**



Michael Flower is a developmental and molecular biologist at Portland State University whose interests turned from the laboratory study of embryos and genes to the ways in which these scientific objects of inquiry have come to be the objects of political battle, economic interest, legal wrangling, and moral dispute. He spent a year studying political and moral philosophy at The Hastings Center, followed by three years at the Salk Institute and four years in the Science, Technology and Public Affairs program at UC San Diego where he joined Clifford Grobstein in carrying out one of the first studies of the public policy consequences of the then-emerging techniques of in vitro fertilization and genetic engineering.

**Amy Boucher**  
**Course Teaching Assistant**



Amy Boucher is currently finishing her junior year at Portland State University in the School of Community Health. Drawing on her background in core biology, physiology and public health classes, she became intrigued with the topic of genetics and public health policy while providing home health care to children with Fragile X Syndrome. Adept at website design, she led the effort to setup and develop the virtual classroom component of the spring term genomics course. She has held several positions in the health arena. In the summer of 2010 she interned with Dr. Tom Becker, Chair of the Department of Public Health and Preventive Medicine at OHSU, and the Northwest Portland Area Indian Health Board. Before moving to Oregon, she worked with an organization for people with special needs in Kalamazoo, Michigan. At Residential Opportunities, Inc. she held the post of Assistant Director of Home Health Care for Children, in which capacity she organized staff schedules and trainings, and worked directly with clientele. After graduation, she plans to apply to the Oregon Masters of Public Health Program and eventually work in the areas of health-related research and policy development.

## **Course Research Resource Faculty**

### **Chapter 1**

R. Stephen Lloyd, PhD  
Oregon Health and Science University  
[http://www.ohsu.edu/xd/research/centers-institutes/croet/faculty/profiles.cfm?facultyID=214&WT\\_rank=4](http://www.ohsu.edu/xd/research/centers-institutes/croet/faculty/profiles.cfm?facultyID=214&WT_rank=4)

Eran Klein, MD, PhD  
Oregon Health and Science University  
<http://www.ohsu.edu/xd/research/centers-institutes/neurology/alzheimers/faculty-staff/klein.cfm>

Kirk Allison, PhD, MS  
University of Minnesota  
<http://www.med.umn.edu/phrh/biographies/allison/home.html>

## Chapter 2

Kirk Allison, PhD, MS  
University of Minnesota  
<http://www.med.umn.edu/phrh/biographies/allison/home.html>

John R. Stone, MD, PhD  
Creighton University  
<http://chpe.creighton.edu/people/profiles/stone.htm>

Senator Laurie Monnes-Anderson  
Oregon State Legislature  
<http://www.leg.state.or.us/monnesanderson/>

Carlos Crespo, Dr. P.H, MS  
Portland State University  
<http://www.pdx.edu/sch/faculty-and-staff>

## Chapter 3

William Lambert, PhD  
Oregon Health and Science University  
<http://www.ohsu.edu/xd/education/schools/school-of-medicine/departments/clinical-departments/public-health/people/lambert.cfm>

Melanie Gillingham, PhD  
Oregon Health and Science University  
<http://www.ohsu.edu/xd/education/schools/school-of-medicine/about/spotlight/gillingham123009.cfm>

Cheryl Hermerath, MBA, DLM, RM  
<http://genes-r-us.uthscsa.edu/resources/consumer/StatePages/Oregon.htm>

Gabriel Lazarin, MS  
Counsyl  
<https://www.counsyl.com/about/counsyl/#gabrielLazarin>

## Chapter 4

Patricia Kramer, PhD

Oregon Health and Science University

<http://www.ohsu.edu/xd/research/centers-institutes/neurology/alzheimers/faculty-staff/kramer.cfm>

Ken Rice, PhD

University of Washington

[http://sph.washington.edu/faculty/fac\\_bio.asp?url\\_ID=Rice\\_Kenneth](http://sph.washington.edu/faculty/fac_bio.asp?url_ID=Rice_Kenneth)

Eric Juengst, PhD

University of North Carolina

<http://philosophy.unc.edu/people/resource-faculty/eric-juengst>

Eran Klein, M.D, PhD

Oregon Health and Science University

<http://www.ohsu.edu/xd/research/centers-institutes/neurology/alzheimers/faculty-staff/klein.cfm>

Steve May

Boston, Massachusetts

<http://www.geneticequity.org/?l=about>

## Chapter 5

Joe Gray, PhD

Oregon Health and Science University

<http://portal.ohsu.edu/blogs/researchnews/2010/12/07/top-cancer-and-genomics-researcher-joe-gray-phd-to-join-knight-cancer-institute/>

R. Stephen Lloyd, PhD

Oregon Health and Science University

[http://www.ohsu.edu/xd/research/centers-institutes/croet/faculty/profiles.cfm?facultyID=214&WT\\_rank=4](http://www.ohsu.edu/xd/research/centers-institutes/croet/faculty/profiles.cfm?facultyID=214&WT_rank=4)

Amy Harmon

New York Times

[http://topics.nytimes.com/top/reference/timestopics/people/h/amy\\_harmon/index.html](http://topics.nytimes.com/top/reference/timestopics/people/h/amy_harmon/index.html)

Eric Juengst, PhD

University of North Carolina

<http://philosophy.unc.edu/people/resource-faculty/eric-juengst>

## Chapter 6

Kent Thornburg, PhD

Oregon Health and Science University

[http://www.ogi.edu/people/dsp\\_person.cfm?person\\_id=85AA90EB-EDCE-6CE9-430F15169659FBF2](http://www.ogi.edu/people/dsp_person.cfm?person_id=85AA90EB-EDCE-6CE9-430F15169659FBF2)

Mark Rothstein, J.D.

University of Louisville

[http://www.law.louisville.edu/faculty/mark\\_rothstein](http://www.law.louisville.edu/faculty/mark_rothstein)

Eran Klein, MD, PhD

Oregon Health and Sciences University

<http://www.ohsu.edu/xd/research/centers-institutes/neurology/alzheimers/faculty-staff/klein.cfm>

Representative Mitch Greenlick, PhD

Oregon Legislature

<http://www.leg.state.or.us/greenlick/>

## Chapter 7

Kirk Allison, PhD, MS

University of Minnesota

<http://www.med.umn.edu/phrh/biographies/allison/home.html>

Michael Garland, DScRel

Oregon Health and Science University

<http://www.ohsu.edu/xd/education/schools/school-of-medicine/departments/clinical-departments/public-health/people/garland.cfm>

## Course Participants

### Alysia Cox Cleys



Alysia Cox Cleys is an OHSU Master's of Public Health student in the Epidemiology and Biostatistics track. For her undergraduate degree she went to University of Oregon and studied Psychology and Spanish. The plan was to take a year off and return to school for a graduate degree in psychology. However, her first job was research in clinical trials in oncology. She was hooked. Six years later, she is still in pediatric research working on the regulatory side of trials. She is intrigued by policy and how

it affects the way in which research is conducted. This class, especially the Newborn Screening and Genetic portion sounded interesting, as she just had her first child in January.

### **Emily Carter**



Emily Carter is an MD/MPH student at OHSU Master's in Public Health Program in the Epidemiology and Biostatistics track. She will be starting a Pediatrics Residency in July 2012. Emily received her BA in Human Biology with an emphasis in International Women's Health Policy from Stanford University in 2005. Prior to attending medical school she worked for a non-profit called the Health Technology Center, which was a think-tank focused on forecasting technology innovation in the healthcare sector. Through that experience Emily became interested in personalized medicine and genomics research. For a short time during medical school she considered becoming a medical geneticist, but realized that regardless of what she does, genetics will be a part of her medical career. She feels that this class was an essential component of her education in both medicine and public health. The most important thing she learned was the role of policy and policy makers in pushing forward innovation, particularly in genomics, and in medicine in general. As a physician-scientist this will be an important factor in Emily's career and will help her to more appropriately educate and work with policy makers.

### **Jenna Gribbin**



Jenna Gribbin is an MPH student at OHSU in the Epidemiology and Biostatistics track. She completed her undergraduate degree at the University of Oregon in 2008 with a major in biology and a minor in chemistry. Since 2008 she has worked for Floragenex Inc., a small biotech company using genomics tools to solve problems in plant sciences. Working here, she realized that genomics is the area in which she want to focus, but she wants to move into the human arena, which is what prompted her to start my Master of Public Health. She found this course, Genomics and Public Health Policy, to be in exact alignment with her personal interest, as well as indispensable for anyone going into the health field. The most important thing she learned in this class is the importance of scientists being able to communicate their research to the public and to policy makers, so that everyone can stay informed and effective policy can be developed!

### **Tyler Johnson**



Tyler Johnson just finished his first year as an MPH student in the International Health track at Oregon State University. He has two bachelors degrees, one in Philosophy and the other in History. He also has a certificate in Biomedical Ethics. His interest in this class comes from his background. At one point he was a chemistry major, but was converted to the social science after a little over a year. He chose the International Health track because he wants to work with native, displaced, and other vulnerable populations. On a more personal note, Tyler tends to push for the bioethical principles of autonomy and respect for persons. This comes from his patient background. He is 29 and has had over 40 surgeries, most of them neurosurgeries. Until he enrolled in the MPH program, he volunteered at Harborview Medical Center in Seattle, WA as a patient/family liaison to their ICU's. The knowledge that he gained in this class will help further his understanding of the genetic differences between populations throughout the globe. This understanding will help him take the integration of cultural competency to the next level when studying and helping other cultures.

### **Lynn Ketch**



As a way to be a well informed community member engaged in the needs of her community Lynn Ketch applied and was accepted to OHSU's Public Health Certificate Program. Her concern is for the vulnerable members of the community who are marginalized by their low income, low social status or ethnicity. They often have no voice in the public forum. She took the genomics class for personal interest. She wanted to learn how it might be used to either advance justice for the poor or further undermine their status. Her last genetics class was 30 years ago and she is amazed by the scientific progress. She is dismayed that our society seems no more prepared for the ethical issues than we were 30 years ago. After this experience Lynn intends to stay current and participate in the public forum watching for potential impacts regarding the vulnerable.

### **Nicole Lucero**



Nicole Lucero completed her Bachelor of Science degree in Health Sciences: Community Health Promotion and earned a minor in biology from Northern Arizona University. Currently, she is working towards a Master of Public Health in Environment, Safety & Health with an emphasis in ethics and policy at Oregon State University. Her interest in this course stems from an ethics

course she took and from her general interest in environmental and occupational health -- specifically the effects that chemicals and various environmental factors have on the human body. From this course she has learned the importance of providing various stakeholders (general public, policymakers, healthcare providers, etc) with the tools and information to make sound decisions regarding genomics. These decisions range from simply deciding to partake in a DNA test to forging legislation for such a vast subject with numerous issues that need to be addressed. Although she is not sure how she will use this knowledge going forward, she has no doubt that many aspects of genomics will apply to whatever she decides to dedicate her future work, tasks, and career to.

### **Corrie Morse**



Corrie Morse is a student in the nurse midwife program at OHSU. She received a BA in communication sciences from Case Western Reserve University and a master's degree in clinical social work from Smith College. As a social worker, she has worked with people living with HIV, mental illness and addiction disorders. Her interest in genomics stems from research published in the last several years about the use of genomes in HIV vaccine research. As a provider of both mental and physical healthcare, P4 medicine is an exciting direction in which to be headed. Corrie looks forward to following genomic advances and being able to discuss the applications with her patients.

### **Denise Musacchio**



Denise Musacchio has her Bachelor's degree in Anthropology from Portland State University. She has future plans to obtain her Master in Public Health. She has been working as a medical assistant for the past 22 years. Because of her experience she has seen the benefits of genetic testing. She originally applied for this course to get an idea of what a graduate level courses were like. The idea of studying genomics had never occurred to her until she was introduced to this course. She found the topic intriguing but until she had a chance to explore the subject matter she had no idea of the many ways genomics could apply to healthcare. Having served 23 years as a Navy Hospital Corpsman, the health of military members, fellow retirees and their families has always been important to her. Denise hopes to use her future experience to continue tending to those needs.

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# CHAPTER 1

## GENOMICS AND P4 MEDICINE

*Corrie Morse and Lynn Ketch*

### Executive Summary

Genomics is the study of the genetic material of an organism. The clinical application of genomics is [P4 Medicine](#): where advances in the field will empower us to **participate** in our health management which will in turn give our physicians the ability to **personalize** our health care by using individual genomic information to **predict** our health outcomes and **prevent** disease. The promise of P4 Medicine is health management rather than disease management. Genomics and P4 Medicine have the potential to revitalize healthcare, a refreshing notion for a field currently bogged down in healthcare reform debate.



### Introduction

Cancer is a frightening diagnosis, the treatment for which often has more severe side effects than the disease itself. Chemotherapy has saved thousands of lives, but takes a toll on the body, physically and emotionally, as well as affecting a patient's family and caregivers. Genomic research is now discovering methods to target chemotherapy, making it more effective in destroying cancer cells and decreasing debilitating side effects. Research has also made it possible to test for genes known to cause cancer, providing the option for interventions such as [prophylactic mastectomy](#). As genomic research continues so will the medical

breakthroughs that save lives by improving treatments and providing more options for patients and their providers. Physicians and scientists in Oregon are already contributing to this work through [multiple institutions](#), with capacity for more research being [added](#).

Work in the field of genomics provides three lasting impressions. How will individual privacy and confidentiality be protected in the Genomic Age? In addition, genomics can raise social issues in regards to expense; what implications will there be for the poor and vulnerable of our communities and developing countries of the world? Finally, due to the unresolved conflict in our country over reproductive privacy and the status of the unborn it raises the question, are we prepared individually and as a society for the complexity of choices genomics introduces in “P4 Medicine?” The following is a background of the science of genomics with the ethical and social implications of genomics in P4 Medicine.

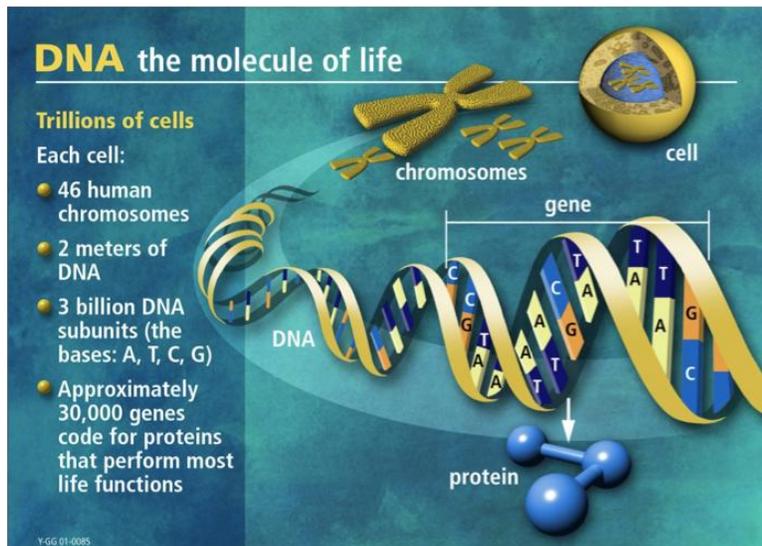
## **Background**

### *What is genomics?*

Unlike genetics (the study of heredity and the functions and effects of single genes), genomics explores not only the actions of single genes, but also the interactions of multiple genes with each other and with the environment. Genomics started with gene sequencing in the 1970’s and has grown in scope over the past forty years. Currently, there are complete genomes sequenced for a number of different bacterium, viruses, plants, animals, and humans, with more being completed all the time. In fact, [researchers at Oregon Health & Science University](#) recently played a key role in analysis for the completed sequence of the rhesus macaque monkey genome, the most common species of monkey used in biomedical research.

## The Human Genome

Most people are familiar with the term genome due to the widely reported on [Human Genome Project](#). In April 2003, the Human Genome Project was declared complete after 13



years and 3 billion dollars. The main goals of the project were to: determine the order of all the bases in our genome's DNA; make maps that show the locations of genes for major sections of all our chromosomes; and produce what are called "[linkage maps](#)" through

which inherited traits (such as those for genetic disease) can be tracked over generations.

Identical DNA (deoxyribonucleic acid) is found in every cell in the body in the form of a double helix coiled into 23 pairs of chromosomes. DNA sequence is comprised of three billion base pairs (each base is one of four nucleotides: Adenine (A), Thymine (T), Cytosine (C), and Guanine (G)). Approximately 25,000 "sections" of DNA (scientists do not know the exact number and estimates range from 20,000 to 30,000) function as protein coding units called genes. The average gene consists of 3,000 bases, but sizes vary greatly, with the largest known human gene (dystrophin) consisting of 2.4 million bases. The functions are currently known for over half of the discovered genes in the human genome. DNA directs cell activity and, in some senses, makes us who we are. The vast majority of our DNA is the same from one person to

another (around 99.9%), but the tiny portion that is different and the environments that we experience throughout our lives are what made each of us unique.

### *Time and Money*

The completed human genome resulted in not a clear blueprint of human growth and development, but a “[complexity explosion](#)” in which scientists are busily trying to find meaning and will continue to do so for decades to come. The Human Genome Project was finished ahead of schedule because of technology that increased sequencing speed. There is a current drive for even faster and cheaper sequencing and an active [X prize](#) challenging scientists to successfully sequence 100 human genomes in 10 days at a cost of no more than \$10,000 per genome. Accomplishment of this goal will give researchers access to more data and, ultimately, make personalized medicine attainable for the public.



### *Health Implications*

Genomics has the potential to deliver major medical advances and change the face of healthcare. Research in the field is helping scientists discover why some people get sick from certain infections, environmental factors, and behaviors, while others do not. Genomics plays a part in nine of the [ten leading causes of death](#) in the United States. Better understanding of the interactions between genes and the environment will help us find better ways to improve health and prevent diseases. Genomics can tell us more than ever before about who we are, where we came from and what diseases to which we are susceptible, but it can also give rise to concerns about discrimination and confidentiality as well as creating difficult medical choices.

## *Ethical Implications*

Three ethical questions arise when discussing genomics:

Who do my genes say I am?

How are my freedom to choose and privacy affected?

What happens to the socially vulnerable?

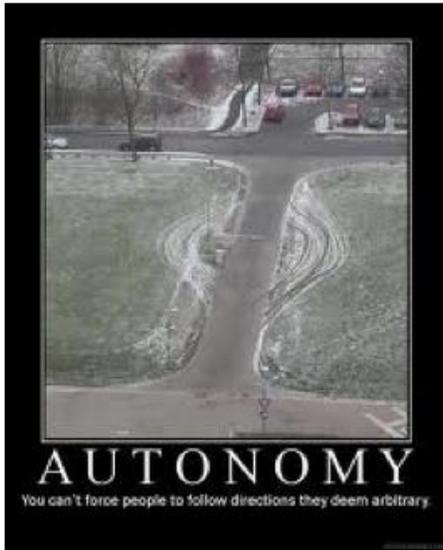
These ethical issues bring into question how we define and value humanness, autonomy, privacy and confidentiality as a society.



In regard to our humanness, the term [genetic determinism](#) has been misused as an oversimplification that we are our genes. In reality genomic and [epigenetic](#) material (covered further in Chapter 6) [interact with the environment](#) to yield expressions that make us individually unique. While genomics helps define uniqueness, it should not dictate value or be used as a social [eugenics](#) tool. Genomics is in danger of being added to the arsenal of those who refuse to abandon racial, class, or disability stratification and devalue the humanness of others.

Just as we value the freedom to choose our careers, lifestyles, and whether or not to have medical treatments, we value our autonomy regarding our genomic data and the choices it offers. Public concern can reflect the loss of personal choice due to [social pressures](#) regarding genetically determined diseases. For example, when the gene for [Huntington's disease](#) is identified before an individual has symptoms and he or she then chooses to have children they will face medical, economic and social pressures that they may not otherwise have to address without this genetic information.

Genomic information must be considered carefully through the lens of privacy and



confidentiality values. Fear of having personal genomic information used against individuals and families is the greatest concern expressed by the public in a [1995 Harris Poll](#). The Genetic Information Nondiscrimination Act ([GINA](#)) has protections against discrimination by health insurance companies and employers with regard to genetic information. However, science and technology have outpaced this policy and there are [no protections](#) for

the epigenetic data currently being gathered.

It is at the convergence of humanness, autonomy and privacy that the ethical debate over a woman's right to choose and fetal life meet, and genomics is integral to this topic (its predictive capabilities and implications for [pre-natal diagnosis](#) are discussed in Chapter 3).

Regardless of one's view on when life begins and rights inferred, it is controversial to [use genetic information in the decision](#) whether or not to terminate a pregnancy. While it is an acceptable medical practice to screen for Down syndrome, those in the Down syndrome community voice their concern that selecting against an afflicted fetus will further erode society's view of the disabled. The discussion



is more problematic when determining what can a fetus be screen for and be selected against, sex? Head size? Treatable diseases or conditions? Social consensus has not been achieved regarding choice and the unborn, genomics seems to make consensus more unlikely.

### *Social Implications*

Social change is slow, genomic science is not. The application of genomics through P4 medicine has been slow compared to the rate new data is being generated and now is the time for broad-based community education and informed dialogue. P4 medicine is a paradigm shift based on the predictive and preventive tools genomics science may render. The goal is personalized medicine in which healthcare consumers can participate. Informed patients will seek out physicians who are literate in genomics. Home testing kits are already available and a primary care provider (PCP) will find they are out of date if they cannot address concerns raised by these [direct-to-consumer](#) (DTC) tests (more detail in chapter 5). As the frontline of medical care, our PCPs can help set public expectations and support medical autonomy.

### **Policy Recommendations**

In the hands of personal health care providers and protected by confidentiality, genomic information has the potential to result in health decisions that are more direct and effective. A [2002 Harris Poll](#) indicated 90% of the respondents believe their doctor should have access to genetic testing results and 69% think doctors helping them prevent disease should do so. As genomic science advances, public health policy and genomic literacy will need to keep pace to support the demand for screening, interpreting, and patient decision making. This goal can be achieved through:

- Continuing education requirements for medical licensing that incorporates genomics for healthcare providers to be up to date on best medical practices, and
- Education for:
  - [the public](#)
  - [healthcare professionals](#)
  - [policy makers](#)

This education will be essential to building capacity for advancing technology and the associated ethical questions. This will take a combined effort of the public, the press, medical training institutions, health professionals and their medical associations, state and county health departments supported by public policy that encourages community collaboration.

To protect the public from those who may use genomic information to discriminate or stigmatize, action must be taken to ensure that :

- current antidiscrimination and privacy policies are [updated](#),
- [healthcare reform](#) addresses this important measure; a single payer system may eliminate the possibility for discrimination in health insurance,
- employers are prevented from discriminating based on genomics.

In the spirit of P4 medicine, individuals will be empowered to participate in their healthcare. The medical home model (in both [adult](#) and [pediatric](#) settings), is a system of care in which medical as well as psychosocial needs can be addressed in one setting. The medical home model should include [genetic counselors](#) as

#### For More Information

Dr. Leroy Hood. Foundation for the Future. P4 Medicine and Genomics  
<http://www.futurefoundation.org/>

Amy Harmon's DNA Age Series..  
 New York Times.  
<http://topics.nytimes.com/topics/news/national/series/dnaage/index.html>

appropriate, and widespread use of the model stands to advance healthcare and P4 Medicine in this country.

# CHAPTER 2

## SOCIAL EPIDEMIOLOGY AND HEALTH CARE DISPARITIES

*Tyler Johnson*

### **Executive Summary**

Oregon is growing more diverse, and that brings problems for medical professionals, who have to navigate an array of cultures that can make health care delivery difficult. Oregon's Senate Bill 97 was introduced to the state legislature in January of 2009, and fizzled two years later. SB 97 specified regulatory boards to develop standards and list of opportunities for continuing education in cultural competence and to develop and implement such education for licensed health care providers. SB 97 brought no new mandates, but was designed to help develop the knowledge and understanding of different cultures aimed at delivering health care that is both medically effective and [culturally sensitive](#). On May 9, 2011, shortly before the defeat of SB97 in the Oregon House, student authors of this briefing book provided verbal testimony to members of the Oregon House Health Care Committee. The **text** reads as follows:

## SB 97: Testimony - House Health Care Committee Hearing May 9, 2011

Students: Emily Carter, Alysia Cox Cleys, Jenna Gribbin, Tyler Johnson, Lynn Ketch, Nicole Lucero, Corrie Morse, Denise Musacchio

Good Afternoon Committee Members. My name is Nicole Lucero and I'm here today, on behalf of my classmates currently enrolled in a new Master of Public Health course at OHSU, **Genomics and Public Health Policy**, to provide testimony on Senate Bill 97.

During one of our sessions, Senator Monnes-Anderson spoke to us about this bill as part of our work on **Social Epidemiology and Health Care Disparities**.

Currently there are huge disparities in the access and quality of health care among different ethnic and cultural groups. In light of growing diversity in Oregon, enforcing the requirement for health care professionals to gain cultural competencies is vitally important in **all** areas of health care, but specifically we would like to address the necessity for cultural sensitivity when using genomics and P4 medicine in clinical practice.

**Genomics** is the study of the entire human genome, including not only the actions of single genes, but also the interactions of multiple genes with each other and with the environment.

P4 medicine refers to the idea that biotechnology can influence medicine and create a system that is predictive, preventive, personalized, and participatory, managing health instead of treating disease.

Without culturally appropriate patient-clinician communication, the use of genomics in clinical practice has the potential to amplify current disparities in health care.

There is an increasing demand for health care personnel who can explain the risks and benefits of these aspects of health care to patients, but in order to convey genomic information effectively to patients and families, **healthcare providers need to understand the cultural values of each of their patients and anticipate how these values will influence the way in which patients, families, and communities will interpret and act on this information.**

Research has shown that "poor and minority patients desire to have as much information regarding their conditions and genetic predispositions as anyone else...(yet) they received **less information, less positive or reinforcing speech, and less talk** overall."

Professional humility that is less authoritarian and respects the patient's agenda and perspective will bridge the cultural poverty and ethnic gap, providing more effective communication about complex medical decision-making.

Encouraging healthcare providers to know about cultural issues that create healthcare disparities ensures that every race, gender and socioeconomic background will be able to benefit from the future of personalized medicine.

To make the best use of genomic data, it will be essential for clinicians to be able to educate their patients about the beneficial applications of genomics, as well as its limitations within the context of cultural concerns.

Together, such measures may help to reduce disparities in health. Social progression as a result of the translation of genomic discoveries to the practice of personalized medicine for EVERYONE will be what we **want it to be**, and NOW is the time to make our case. With the passage of Oregon's SB 97, Oregon will have the opportunity to be at the leading edge of cultural competency in the practice of healthcare in the United States.

On behalf of my student colleagues, and our instructors, THANK YOU for the opportunity to share these perspectives with you today.

## Background

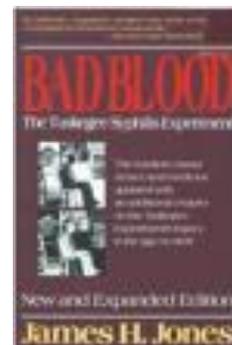
### *Introduction*

The population of the world is very diverse. It is because of this growing diversity that there is an increasing need for health care providers to be culturally competent. The fields of

genetic science and social epidemiology are making us take another look into just how different we really are. Since we are all approximately 99% similar at the DNA level, new discoveries in genomics are fueling many of the debates. This is especially true when it comes to health and healthcare.

Differences exist in many aspects of our society. These differences are usually between population groups based on gender, age, ethnicity, socioeconomic status, geographical location, sexual orientation, and disability. Disparities arise out of inequalities between different groups. When these inequalities are related to the differences in the incidence, prevalence, mortality, burden of disease, or other adverse health conditions, they are labeled health care disparities. Today, health disparities tend to be more prominent in populations that have been marginalized and/or are persistently faced with discrimination and historical trauma. This has led these health disparities to be viewed as unjust. Furthermore, if the cause of these disparities is avoidable or unfair, the situation is considered to be unequal or have inequity. These inequities can occur in many different forms across many aspects of daily life. Some of these inequities include:

- Lack of access to healthy food choices
- Lack of knowledge about health impacts of daily routine items (i.e. food, behavior, etc.)
- Exposure to unsafe, unhealthy, and stressful conditions at work and/or home.
- Inadequate access to basic health care and other essential services
- Lack of adequate health insurance or no insurance at all
- Cyclical cycle of unhealthy (i.e. poor health leads to poverty which leads to even poorer health)
- Social stigmatization
- Issues of trust ([Tuskegee Study](#))



## Science

According to genomic studies, all people are very similar on the genetic level—about 99 percent. Only a small percentage of our genome differs from person to person, yet we can visibly see a huge (phenotypic) difference due, largely, to the interaction of our 3 billion DNA base pairs with the environment (“[genomics](#)”). Disparities arise out of [haplotype](#) associations within different populations.



An example of this would be the treatment, or cost of treatment using a “new technology,” of a disease that targets a certain ethnic group (i.e. BiDil). [BiDil](#) is a heart-disease drug that is marketed to African-Americans because they seem to have a better response to the drug. Scientists are starting to wonder if this strong response to the drug comes from a genetic predisposition among African Americans and, if so, could have the potential “to spark a new era of racism” if there is no cultural sensitivity in explaining its effect.

Another example is found in “[Next Generation Disparities in Human Genomics.](#)” This paper states that genome-wide association studies (GWAS) are almost exclusively focusing on people of European descent. The ratio of studies on Europeans relative to other ethnicities is 12:1 and in each individual study the average sample size was at least 2:1. This experimental

design creates a two-fold problem. First, it reduces the confidence level, i.e., a level of accuracy of a given study correlating a certain gene with a certain disease. Secondly, it does not address issues relating to [allele](#) frequency. The problem with this research error of [Type 1](#) is its potential effect on the widening of the health disparities gap, but with particular vigilance, GWAS studies can be helpful. One question remains, does the inevitable march of science lead to the propagation of health care disparities?

### *Ethics*

Whenever discussing issues around health and healthcare, it is always important to bring certain ethical principles, like cultural competency, to the forefront of the debate. This is to insure the equitability of the burden of disease, as well as, the equitable distribution of the benefits relating to treatments. The same is true when discussing health disparities (e.g. SB 97). This stems from the belief that many societal factors create, or further, these disparities. Some of these factors include: poverty, lack of education, inequitable access to healthcare, and inadequate resources to make the recommended adjustments. For example, impoverished communities may not have easy and local access to healthcare. Having to commute to a clinic to receive care may not be an option if one has to work in order to survive. Leaving work for care may result in unpaid leave, which leads to a double financial burden by having both the cost of care and the loss of wages. In a speech during the 2007 Presidential Forum, then Senator Obama remarked that--

*“Close to 80 percent of those without health insurance, work. They are, in some cases, actually folks who are working in the health care system but don’t have health care benefits. And they’re cleaning bedpans and, you know, cleaning floors in hospitals and unfortunately don’t have access to the same system, which they’re a critical part of. And those groups are disproportionately minority.”*

Note: According to the [CDC](#), about 46.3 million Americans are currently living without health coverage.

Reaching out to these communities is important since informed equitable access to new emerging technologies, like genomics, is a major contributor to reversing health care disparities. Race and genes create a unique pattern and is the biological map of characteristics that define genetic ancestry. It is important to define these populations to help healthcare providers tailor preventative treatments that will prove to be most successful for a particular group (e.g. BiDil).

As the technology proves itself useful, it will be available to health providers. It is then up to the providers to decide when to employ this new technology. But is this the way it should be? Shouldn't others be included in the decision, especially those whom the technology will have the greatest effect (i.e. the public)?

Dr. Rotimi, the current director of the Center for Research on Genomics and Global Health at the National Institutes of Health ([NIH](#)) discusses theoretical principles that should guide physicians and scientists. Many of these principles discuss the need for the 'gaps' in knowledge to be filled, as well as, treating people with a sense of equity and respect. For Rotimi, understanding human genetic variation through genomics is important in global efforts to reduce health inequalities, and these efforts must "ensure all human populations are engaged in the use of genomics to solve societal problems from hunger to health." The same sentiment is echoed in [The Belmont Report](#). It is also worth noting that these themes are also present in community dialogues hosted through the [Center for Public Health and Community Genomics](#) at the University of Michigan but expanded to include issues of trust (and distrust), education, and

access (to new technologies). Why it is important to include members of the “community,” including a representative sample of ethnic and socio-economic groups, in this dialogue is because they are stakeholders in the debate. Furthermore, if disparities do arise, they will be the first to feel its effects.

Since the ramifications of genomic data affect everyone on the planet, the stakeholders are just that, the entire human population, but separated into discrete groups.

The first group of stakeholders is the **individual**. Because of the “long arms” of genomic data, the concept of individual does not conform to the age standard used in biomedical ethics. For example, one may argue that a 16 year old is not legally qualified to make a surgical decision. However, when it comes to genomics technologies, no such barrier exists. Indeed, any genetic test ordered by an adult member of the 16-year old’s family would have relevance to him/her, as well. The same logic could be applied to, say, a four year old. The average 16 year old American is unable to understand genomics technologies, much less four year olds. Therefore, when it comes to issues of “informed” consent, the permissible age gap for making decisions about the personal use of genomic technologies (e.g., genetic testing) needs to be addressed by policy makers.

**Society** also should be viewed as a stakeholder when talking about genomic data. This is especially true given the constantly evolving landscape of social epidemiology and healthcare disparities in the Age of Genomics. After all, the burden of many of the diseases with a genomic component is felt by the societal system through healthcare, health insurance, and other avenues.

**Future generations** are another less talked about stakeholder in this debate. Although their voices may not be heard literally, we *must* keep a watchful eye out for unforeseeable reactions to genomic interventions that will impact the coming generations.

**The Scientist** doing this ground breaking genomic work is another stakeholder. Without the tireless pursuit of this scientific technology, we would not be having this debate. As a result, we *have* to continue to include science in *every* aspect of this coming debate about the place of genomic data in the world of health care disparities.

**Healthcare delivery systems** are yet another stakeholder in this debate. These systems, and the medical professionals who work within them, are going to be on the “front line” of the battle in implementing this new technology.

**Insurance companies** must also be considered a serious stakeholder in the implementation of this genomics information. After all, in the current healthcare system, they are the final arbiter in deciding reimbursement issues and setting the guidelines for the “Haves” and “Have Nots” based on coverage allowed by one’s particular policy, never mind all those individuals without health care protection of any kind.

One final –and critically-important--stakeholder in this debate is the **policy maker**. Policy makers have to make sense of all of this new genomics information and figure out a way to try and implement its use free of any unintended consequences. They also must remain vigilant and for these unintended consequences and be capable of making the appropriate changes as quickly as possible when the need arises.

## **Current Policy**

The rise in the modern usage of genetic testing has led to the furthering of some privacy concerns about privacy, confidentiality, and [informed consent](#). One part of concern to an individual's privacy relates to discrimination, whether it be societal or economic. Concerns about privacy are reflected by the passage and adoption of the Genetic Information Nondiscrimination Act ([GINA](#)) in 2009. Although this relatively new law creates a sense of comfort, it still leaves much work to be done as new scientific findings emerge. A perfect example of this is [Direct-To-Consumer Marketing](#) (DTC) of genetic testing and the issues of privacy which it raises, issues not expressly addressed in GINA.

Another example is the recent study done on the Havasupai Native American tribe. In this case, the Havasupai were discriminated against. Throughout the study there was no attention paid to the concept of respect for persons. Since the [Havasupai](#) are Native people, they have a slightly different healthcare system. Whereas, the main healthcare system in the United States is individual/patient centered, the Native system is community oriented while upholding the concept of respect for persons. Yet the Havasupai have an added measure of social disparities that stem from being a part of a minority *and* a native population. This begs the question if the protections afforded by GINA, and furthered by the tribal system will this be enough to keep this established genetic connection between tribal relation and diabetes risk from creating further social and healthcare related stigmatization and disparities?

## **Policy Recommendations**

When discussing policy around implications stemming from genomics, it is important to not overlook the concerns of "The Public." The need to include the values of the public is

because the public will be the main “consumers” of application of genomic technologies. These fears stem from issues like employer or insurer discrimination based on genetic data, accuracy of [genetic test results](#) proper education of medical professional (and public) on how to interpret [test result](#) and how to best use this information once it has been gathered. In September of 2007, the Secretary of Health and Human Services (HHS), [Michael Leavitt](#) suggested, that personalized medicine needs to be defined in a way that people can understand. He stated that we should harness what we have learned into

1. Developing evidence-based standards for measuring and improving quality of care.
2. Building systems of health information technology to make patient records available when and where they are needed
3. Use those same systems to gather medial evidence from the day-to-day practice of medicine so that clinicians can make the best medical decisions for their patients
4. Use out personal genetic information to tailor treatments more effectively for each patient.

Since then, the Center for Disease Control (CDC) and the HHS have jointly identified 3 crucial areas to focus on. They are:

1. The need to question who has access to basic data from researchers
2. The need for clear policies concerning appropriate oversight of genetic tests. This includes a better understanding of the validity of individual tests.
3. A need to continue to address privacy and security issues in health information technology

Overall, these are the same issues that still need to be addressed in policy concerning genetic testing and its implication for the general population.

## **Conclusion**

As science and medicine moves closer to understanding the genetic map of minority groups as individuals, and as part of the whole population, the more we realize just how similar

we are on the genetic and socially-cultural level: WE ALL BENEFIT FROM GOOD HEALTH CARE. Unless the advancements of policy keep the advancements of science along the genomics front in check, the equity gap in health care will continue to widen. This widening will be seen in the “Haves and Have Not” issue, as depicted in the 1987 film “[GATTACA](#)”. In order to avoid the widening of the health disparity gap, multi-sectoral policies and educational campaigns need to be created in order to insure the optimal usage of, say, new genomic technologies by an informed public. Although science and its products are “value-neutral”, societal interpretations of scientific data, as well as the social and ethical ramifications that accompany it, are not. This comes from the process of integrating a scientific concept into an already value laden system (society). If we continue the trend of having the newest technology only affordable to the rich, the health disparity gap will remain. By including the whole spectrum of society, however, society can be poised to reap the highest benefit from this technology. But the question is how to do this? The answer to access is, as always, “cost” (drugs, everything that humans need to prosper), involves a functioning political and economic infrastructure with “the common good” at its center. How likely is that to happen? In the words of (then) Sen. Obama speaking about his proposed overhaul of the U.S. healthcare system in the context of how to reduce health care disparities:

*“But the most important thing that we can do is to make sure that every single person has coverage, every single person has access to basic primary care and preventive care. If we’re doing that, we’re going to see those disparities close significantly.”*

At the same time, it is vital that policies are in place to ensure proper usage of genomic discovery and the information coming from it. Genomics, like other branches of science before

it, has the ability to eliminate or widen the health disparity gap. But we have to do *something*—and soon, for the words of Martin Luther King ring just as true today as they were 45 years ago—

*“Of all the forms of inequality, injustice in health is the most shocking and the most inhuman.”*

--The Rev. Martin Luther King, Chicago, March 25, 1966

Despite the beneficial advancements in genetic and genomic discovery, the public still maintains a fear of discrimination. The question that remains is, what policies need to be put in place in order for the public to feel confident in utilizing these technologies?

As reported in a 2007 US public opinion poll on the uses of genetic information and genetic discrimination--

*Americans clearly support the use of genetic testing to further their own health and the health of their families. The public also supports the use of genetic testing in medical research. However, this enthusiasm about genetic testing is tempered by widespread public concern and distrust about the discrimination that could result if insurers and employers access and use genetic test results. Three in four Americans support laws to ban such discrimination. Without such laws, much of the promise of the Human Genome Project to identify the causes of disease and promote public health is likely to remain unfulfilled.*

# CHAPTER 3

## GENETIC EPIDEMIOLOGY, PRE-NATAL AND NEWBORN SCREENING

*Alysia Cox Cleys*

### Executive Summary

In the United States, the [newborn screening test is required](#). However, that is where the commonality stops. The requirement for the test is the only thing each state has in common, as



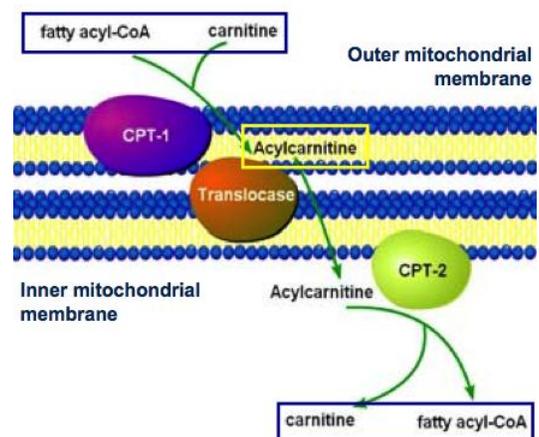
which genetic disease are tested to [how long the samples are stored](#) and who is allowed to use it varies from state to state. The law is unclear if the samples will be used at some point in the future for research or other clinical diagnoses. These newborn screening tests have [prevented death](#) for preventable diseases. In the state of Oregon, it has about [34 disorders](#) they screen on the newborn screening test.

Unfortunately, at times, a specific test is not done and the genetic disorder results in the unthinkable, [death](#).

### Background

#### Introduction

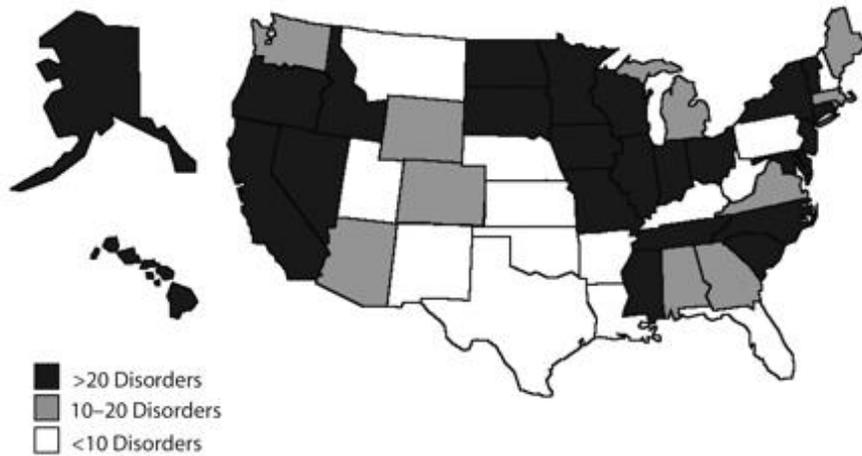
[Fatty acids](#) are the building blocks of the fat in our bodies and provide an important source of energy for our bodies. The fatty acids that humans eat are broken down and eventually bind to membranes of



muscle fibers where they are either stored for future use or used for energy. The breakdown of fatty acids is the release of energy. This process takes place in 3 major steps. Firstly, the fatty acid is transported across the outer mitochondrial membrane by [carnitine palmitoyltransferase-1](#) (CPT-1). CPT-1 then takes it across the inner mitochondrial membrane where it ends up in the matrix. Once inside the matrix, the fatty acid has been converted to a fatty acyl-carnitine and this reacts with coenzyme A to release the fatty acid and acetyl-CoA, allowing the body to use when needed. It is thought that CPT-1-A allows the fatty acid levels to rise and it accumulates in the skeletal muscle, thus decreases the ability of muscles to oxidize fatty acids. This can cause a fatal outcome, death. This enzyme was first found in Native Alaskan populations through newborn screening tests. Once found, healthcare providers and parents can avoid situations where the baby is fasting, causing the fatty acid levels to pull energy stores from the tissue. This is exactly what happened to a [native Alaskan boy](#) who was transferred to a local Portland hospital for a bilateral Achilles tendon release. The boy was fasting for the procedure and eventually died due to the body's inability to use the long-chain fatty acids for energy. The boy was born several months prior to the newborn screening standard in Alaska.

Since the 1960's, public health programs continue to screen newborns for genetic abnormalities and diseases that can lead to severe side effects, possibly even death. Dr Robert Guthrie created blood tests to find [phenylketonuria](#) (PKU), a genetic disorder that can lead to mental retardation if early detection and a specific dietary treatment are not given. Babies with PKU do not produce an enzyme needed to break down the amino acid, phenylalanine. This amino acid helps kids grow and develop into an adult. If the enzyme that breaks phenylalanine is absent, phenylalanine builds up and cause damage to tissues in the brain. Once the deficiency is

detected early (via newborn screening test), this potentially detrimental disorder can be controlled if there is education on how to avoid the triggers. Thus, PKU is completely



preventable if treated and “screening for phenylketonuria has been a particularly favorable case for cost-benefit analysis because the untreated disease

imposes considerable societal costs against which to offset the costs of diagnosis and treatment” (Pollitt, 2001). This test alone, birthed the newborn screening testing era in the 1960’s. Since then, “pulmonary, genetic, hematologic, endocrine disorders and infectious diseases have been identified by laboratory testing and added to the panel” (Orlova, 2009). Many of these are metabolic disorders (inherited errors of metabolism) that disrupt the body’s use of nutrients to keep tissues healthy and produce energy to survive. Other disorders that screening can detect are hormonal problems, blood and other inherited disorders. These disorders can hold back an infant and meeting those developmental, physical and mental milestones. Since parents can unknowingly pass a gene or genes for a certain disorder to their child, a simple blood test can determine if the baby will have certain conditions that could eventually cause lethal outcomes later. While these genetic disorders are considered rare, most of the babies’ tests are fine. However, early detection and immediate treatment can make the difference between lifelong injury and healthy growth. The federal government allows the state

to determine which disorders will be included in the panel. The states can vary from [three to sixty-three conditions](#) (Orlova, 2009). The map below shows by varying shades of color to demonstrate the [difference between each state](#) and the number of tests it requires. As you can see from the map, it varies from state to state with no consistency. With each new baby born, within 72 hours, a heel prick and splotches of blood are placed on the card and sent off to the state for analysis (Newborn Dried Bloodspot Screening Business Process Analysis, 2008). While the federal government has supported newborn screenings, the results of the newborn screening tests remain the state's property, without much involvement on behalf of the federal government in terms of storage or which tests will be included in the pane. In addition, each state is able to determine which conditions will be tested, if the screening is mandatory and if informed consent process is part of the informational step prior to samples being taken and if a second heel stick is necessary, usually done by the pediatrician at the two week wellness visit. The newborn screening is a mandated program and adding new disorders to the panel requires a review of costs versus potentials savings from early detection.

Personally, I was not aware of the initial newborn screening test. Having my first baby in January, I was informed about the newborn screening heel stick at the two-week wellness visit, but not about the test that was done with 72 hours of birth. The information shared regarding the test at 2 weeks was vague. I knew the test would look at genetic diseases and they would let me know if something came up. There was no mention of what kind of tests was being done. In addition, the persons who were informing me of the procedures was not well educated in why the test was being done, where the samples were being stored and how fast would I hear

back if the results were positive. For all the phlebotomist knew, “the tests were stored in a box in the basement”.

### *Science*

While some genetic disorders are still unknown, CPT-1 deficiency is known and treatable. It is a rare metabolic disorder that has been noted in the native Alaskan population. The body prevents the exchange of fatty acids into energy, especially during periods of fasting. A variety of symptoms may be present in patients with this deficiency, and in severe forms it can be mistaken for sudden unexplained infant death (SIDS), due to the inability to maintain glucose levels over night therefore expiring in their sleep or as Reye syndrome where liver failure leads to the toxic agents normally filtered by the liver impairing the function of brain cells. In infants and young children, periods of fasting and illnesses such as fever and infection often trigger the debilitating effects. Symptoms can be prevented by avoiding fasting and closely monitoring blood sugar levels during illness or other periods of increased risk to insure the child is maintaining proper energy levels.

Genetic epidemiologic studies can provide information on the extent to which the ancestral risk of occurrence is attributable to genes that are being passed down generation to generation. Such as the CPT-1 deficiency gene is common in Alaska natives, researchers can reduce morbidity and mortality through prevention and treatment if discovered during the newborn screening test. As we move from the research results and are able to translate genomics to human disease, the need to involve counselors and therapists for drug, gene and genetic purposes. In contrast, public health prevention is most effective when applied to common diseases at the population level. The prevention of phenylketonuria and CPT1-A

through newborn screening are two examples of the successful integration of genetics and public health (Pollitt, 2001). Potential environmental factors should be included in large-scale gene searches, or it may not provide as clear of a picture (Neil, 2003). The public health impact of a disease depends on the population prevalence and the social impact the disorder will have. If research and interventions only study genes that are associated with rare disorders it may not have as big of an impact if we try to reduce common diseases that are more prevalent (Neil, 2003). Research designed to decrease exposure or increase protection from these environmental exposures would have more impact on population (Neil, 2003) than rare disorders and disease. In conclusion, identification of genes through the powerful technology of genomics is likely to have a greater impact on Mendelian diseases (diseases that are passed down from parents) and some complex diseases than on others.

### *Ethics*

Recently, the Washington Post wrote an article about newborn screening and the future of the blood samples smeared on the Guthrie cards (Stein, 2009). At the time, the state of Minnesota allowed medical researchers to use the blood for research, unbeknownst to the non-consented of the legally authorized representatives, in this case, the [parents of the newborns](#). Obviously, the blood was collected for newborn screening tests, however consent was not given nor an option when I came to future research. Working in the research world, [having an endless supply of specimens is extremely exciting](#), but they were not obtained ethically. Each year 4 billion newborns go through this heel prick ritual (Stein, 2009) and have been since the 1960's, which make it a standard of care by default. At the time of the Washington Post article, seven states kept the blood stains cards indefinitely (Citizens' Council

on Health Care, 2009). In 2002, the Center for Disease Control and Prevention (CDC) proposed to create a databank of the leftover blood samples for use of “important public health surveillance and potential epidemiologic research, including population-based data on prevalence of genetic variants, markers of environmental exposure and infectious disease.” The opportunity to opt-in or opt-out of this bio-repository was not mentioned. Unfortunately, several parents in Texas were enraged about the collection and storage of newborn samples which were subsequently [destroyed](#) due to lack of consent.

As science advancement in the world of genetics and genomics continues to rapidly advance, soon we will be able to eliminate disorders before they even start showing signs. Will this be done with the use of the newborn blood samples? When technology allows for us to select certain genes at preconception, our society may be headed towards a world with [designer babies](#). If we can remove disorders and diseases, what will stop us from wanting to select for IQ level or eye color (Big Think, 2011)? In the [video interview by Amy Harmon](#) of the New York Times, a couple was profiled for using [pre-implantation genetic diagnosis](#) (PGD). PGD is a costly procedure which can start around \$20,000. This procedure tests several eggs for the genetic disease (which the client is able to choose); in this case it was colon cancer. The one without the gene was selected and implanted in the wife’s uterus. (Harmon, 2011). Genetic epidemiologists study the role of genetics and how it determines health and disease in populations and how these genes interact with the environment. They will have an increasingly important role in addressing the gaps in the translation of genomic information, especially in the conduct of population-based research aimed at improving health and preventing disease. The past few years has seen the promotion of an epidemiological approach to the human

genome, so-called, "[Human Genome Epidemiology](#)," an evolving field of inquiry that uses systematic application of epidemiological methods to assess the impact of human genetic variation on the occurrence of disease. The important role human genomic epidemiology will continue to play in the continuum from gene discovery to the development and applications of genomic information for diagnosing, predicting, treating, and preventing disease will be crucial to the evidence-based integration of human genomics into the practice of medicine and public health in the 21st century. Translating genomic research into benefits for human health requires large numbers of population-based biological samples.

### *Policy*

According to the Center for Disease Control, epidemiology is defined as "the study of the distribution and determinants of health-related states in specified populations, and the application of this study to control health problems." [Genetic Epidemiology](#) is the study of genetic factors and how they determine health and disease in families and in populations and also how the genes interact with environmental factors. With

With the information we have provided and with support of the American Academy of Pediatrics (AAP), the federal government's Health Resources and Services Administration, the following suggestions have been made in terms of a policy:

- Parents (or legal guardians) should be informed about screening procedures and have the right to refuse future research, as well as the right to keep the results private and confidential.
- Parents (or legal guardians) should be well-informed about the benefits and risks associated with newborn screening
- Prohibit insurance companies (or other entities) from discrimination of babies whose test is abnormal or positive.

Education is by far the most important. The information shared should be given in advance, thus allowing the parents to review the information instead of trying to comprehend just after the birth of a child. This will give parents the time to refuse (or opt-out) of the procedures if the newborn screening is not mandatory. Which if the parents are fully informed; there should be no reason to opt-out of the newborn screening since the benefits outweigh the costs significantly. By giving the information regarding the newborn screening at the time of the test, is unjust as it is taking advantage of the situation and emotional overload the new parents may be experiencing at the time of birth and refusal may be the easy answer. Being well informed about the risks and benefits of screening is an integral part of the research process as it allows the guardians to make an informed decision about participation and the level of usage of the specimens for other research tests. Knowing where the samples will be stored and how they will be used in the future, will give confidence to the guardians that their baby's samples will be protected and any results will remain confidential from insurance companies which may use this confidential information as evidence for predisposed conditions and limiting coverage. These policies will help determine what the [state will do with the samples after the screen](#). Lastly, at a federal level, newborn screening should test for the same disorders, as consistency is key. This will avoid inconsistencies in healthcare if transferred from one state to the next.

## Conclusion

In the future, the post-birth screen, may not be required, as the science of genomics advances and the thought of [eugenics](#) approaches normality, soon we will be able to eliminate disorders before they even start showing signs, thus making the newborn screen a moot point. With companies, such as [Counsyl](#), the preconception review process allows potential parents to screen their genes for potential genetic diseases. This will inform the parents on the risks they may pass down to their offspring. The decision to conceive or use PGD is then left to the couple.

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# CHAPTER 4

## GENETIC TESTING AND PRIVACY

*Denise Musacchio and Nicole Lucero*

### **Executive Summary**

It is clear that the potential impact of genomics is enormous. People can find out their genetic family history or learn if they have a predisposition to a particular disease or disorder.



Pharmacological therapies can be developed to establish personalized treatment for cancer patients or those suffering from chronic disease.

Now, crimes that have been deemed “cold” can be solved through the use of familial DNA searches.

The potential is truly endless. There are however, privacy issues that have caught public attention. The pursuit of privacy is not a new issue. Public angst about privacy is increasing due to rapid advances in the study of the human genome and how it is linked to computer technology. Genomics is very important to the future of medicine because its key goal is to increase the efficacy of treatment and eventually prevent disease. Genomic research has a lot to offer, but details that are published are small parts of a person’s identity. Information seekers can put together those bits and pieces and by process of elimination discover things that a person had not intended to make known. People worry more and more about anonymity when they donate their DNA to participate in research studies for one simple reason, DNA is self-identifying. Today ordinary citizens are submitting their DNA to private companies with the hopes of unlocking information about the future of their health.

Currently, well-known drug stores chains are selling over-the-counter-test kits that can be used by the public to verify paternity. These kits can be used in ways that cross the line when it comes to the invasion of privacy.

## **Background**

When a person's entire genome is at your disposal everything about their genetic identity can be exposed. In January 2011, the fifth edition of the [Cogent Genomics, Attitudes & Trends](#) study reported that of 1,000 American's surveyed, 71%, "Are concerned about how their genetic information would be stored and who would have access to that information." Issues that the public is concerned about accessing their genetic information include insurance companies, employers and the government. In addition, 37% stated that they would likely not participate in genetic tests in the future. Fears and uncertainty about the power of biotechnology, in general, is trumping the enthusiasm and hopes for what genomics/genetics can accomplish for us personally. Even more shocking, 77% reported that they did not know if there were any protections currently in place to prevent discrimination (referencing to the [Genetic Information Nondiscrimination Act](#) [GINA]) and 81% of physicians are not familiar with GINA (Business Wire, 2011). The lack of familiarity is disconcerting especially when industries are trying to tout the endless benefits of [genomics in practice](#). The fact is, those surveyed are generally concerned with privacy issues. This concern for privacy combined with a lack of education about the benefits of genomic research is resulting in a public that is becoming more and more apprehensive about turning over their genetic material.

### *Current Applications*

There are many benefits to genomic testing. DNA enables us to diagnose hereditary disorders such as Neurofibromatosis or Hemochromatosis (a disorder that results in excess iron storage in the body). Currently, the amount of research data that is being collected for publication is enabling us to link certain genes with devastating diseases such as cancer and



Alzheimer's. The use of genetic material is going beyond diagnostics. Genomic knowledge is evolving to the point of being able to predict disease with hopes of being proactive with medical intervention. Pro-activity can be a vague issue especially when faced with the uncertainty how to interpret and use genomic information. A perfect example is when a woman has to make choices after learning she has inherited the same genetic mutations

from a family member that suffered through breast cancer.

### *Breast Cancer*

Breast cancer can be one of the most devastating diseases. If found and treated in a timely manner it does not have to be an automatic death sentence. Healthy human genes have protective mechanisms in place. Breast cancer susceptibility gene 1 and 2 (BRCA1, BRCA2) are classified as tumor suppressors. When there are mutations in these genes a person has increased susceptibility to breast cancer. Asymptomatic women with strong family histories of breast cancer are choosing to undergo genetic testing to determine if they have BRCA1 or BRCA2 mutations. As a consequence these women are now facing serious decisions about what

course of action to take with the results of these tests. On the one hand, she may be asymptomatic meaning she has neither malignant lumps nor findings on a mammogram. Typically if she is over forty, this type of patient needs only routine self-exams and some form of yearly medical screening. On the other hand, she may have several primary family members such as a parent or sibling who are being treated for or have succumbed to breast cancer at a fairly young age. The emotional aspect created by how a woman perceives her risk factors will determine what she does with the information.

### *Ethical Issues*

There are more and more cases of women making the choice to become a “[previvor](#)” by having [prophylactic mastectomies](#). A previvor is someone who undergoes treatment prior to symptoms or diagnosis. Although the decision to be a previvor is very personal, the complexity of this issue and its many implications make it difficult to avoid having it become a family matter. Family members may end up divided when their ethical beliefs conflict with the choices that are being made. Diagnosed breast cancer survivors may experience guilt because they may have passed on “bad genes” to the next generation. Out of pocket cost can be a huge factor for a person deciding on prophylactic mastectomy. Insurance companies will not likely cover the expense of a procedure that is not clinically indicated. There is a risk of people making their own conclusions about health based on results of private Direct to Consumer ([DTC](#)) testing and without the benefit of a knowledgeable health care provider. Decisions should always be based on education and never the result of fear or misinformation. Lastly, there is a strong ethical issue in honoring the Health Insurance Portability and Accountability Act ([HIPPA](#)) when nondisclosure could mean a delay in diagnosis and treatment for a patient’s family member.

### *Direct to Consumer Testing*

It is well known that the art of sales require good marketing strategies. Direct to Consumer (DTC) DNA testing companies provide a service to the public that can be accessed on the Internet. The primary goal of DTC testing is to provide the consumer with a personalized list of genetic mutations associated with a variety of anomalies or disease processes. By ordering a kit and sending in a vial of saliva, companies such as "[23 and Me](#)" offer the consumer a chance for active participation in their own health care. They also offer a discounted rate if you sign up for a monthly membership. [Navigenics](#) offers the option of [pharmacogenomic](#) screening that helps the consumer understand how their body may react to certain medications. Their website appears to be committed to having health care providers involved and even has a "Physician log in."

### *Ethical Issues*

Many people understand that DTC testing is not to be used as a primary tool for diagnosing. By in large, testing for genetic mutations includes light hearted and entertaining information such a predisposition for male pattern baldness. Amusement with this information may depend on whom you ask, however. There is a risk however that some may make life-altering decisions based on these genetic predictions. Imagine your personal result stating you have a predisposition to a genetic mental disorder such as [Schizophrenia](#), what would you do with that information? Although Schizophrenia is not thoroughly understood, many patients seem to acquire it in



adulthood. It is conceivable that discriminatory practice could occur through insurance companies, employers, schools, coworkers etc. as a result of a privacy breach. Worse of all, a person who is convinced of deleterious diagnoses like Alzheimer's may start to make end of life plans that could go as far as contemplating suicide. DTC companies need to have some regulation to ensure proper and responsible advertising and education.

### *OTC Paternity Test Kits*

Popular convenience stores such as Walgreen's are now selling over the counter paternity test kits. Affordable to many, companies like [Identigene](#) charge for the initial kit then add an additional charge for lab analysis. These definitely have value for sorting out private family issues; a simple cheek swab is all you need. If paternity needs to be proven for legal purposes, the consumer must contact the company and meet with a professional where papers are submitted to the state and samples are obtained in a controlled environment.

### *Ethical Issues*

There are a few [covert ways that DNA has been collected](#) and some fall in to shady areas with regard to invasion of privacy. According to Amy Harmon of The New York Times, there are now cases of ordinary citizens who stalk others waiting for discarded material. They use these simple DNA test kits to test for familial connection. Others wait for the death of a relative and without prior consent collect DNA samples to test for paternity or to learn about any deleterious genetic mutations that could be inherited. Since this technology is now very affordable and it is so easy to use it enables the ordinary layperson to become an amateur forensic scientist. Section One of [The Massachusetts proposed Genetic Bill of Rights](#) states "The Commonwealth recognizes that genetic information is a unique product of an individuals body,

the unauthorized use of which interferes with both privacy rights and property interests of that individual". Before establishing punishment for violation of privacy, we must first establish that [DNA is the private property of an individual](#).

### *GWAS Studies*

It is one thing to use DNA to make personal decisions about your health. Genome Wide Associated Studies ([GWAS](#)) use large amounts of genetic data from volunteer research participants. The data is available to the scientific community for the purpose of studying common genetic variants that are associated with many different diseases. In order to keep a person's identity private certain pieces of information are protected from public view. Such information would most likely include name, date of birth, social security number and address. Other bits of information may be needed to add relevance to the study such as sex, age, height, weight and ethnicity. A lot of [genomic research data that is published on the Internet](#) is viewable by the public. Although aggregate studies may be presented with just bits and pieces, a person with research familiarity and a little computer savvy can potentially figure out a person's identity and connect the private information that comes along with it.

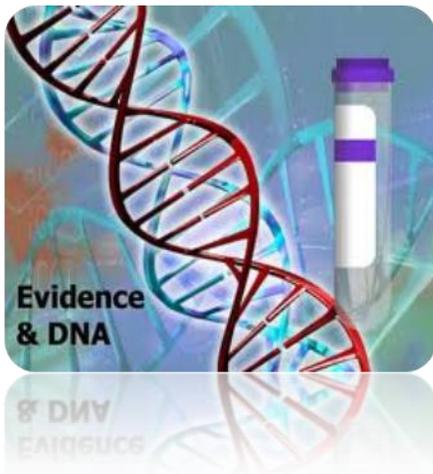
### *Ethical Issues*

Although GWAS studies are extremely valuable to the future of genomic science that information may be hindered by the threat of keeping research participants' identity private. Even small subsets of data can be used to predict a phenotype or basically figure out who you are. Policy makers may start considering restricting data searches to only those scientifically qualified to do research. Some researchers believe that anonymity cannot be attained and that research participants should be thoroughly informed of the risk. If they are not willing to take

that risk, they should be eliminated from the study. Choosing only research participants who are willing to completely forgo anonymity may limit the amount of data available for future studies.

### *Forensics*

One of the primary objectives of forensics is to use DNA for identification. Crime labs do



DNA testing to help solve crimes or over-turn convictions and they acquire the materials needed to test for DNA rather easily; a simple swab from a discarded coffee cup or cigarette. The FBI maintains a database called the

[Combined DNA Index System](#) (CODIS). There are instances

where a suspect has no information entered in CODIS but a family member has. So while an investigator may not pin a

match on the DNA obtained from a discarded coffee cup, they can use partial matches to find

close relatives to the potential suspect. [Familial DNA](#) has successfully been used in two

California cases. However, who is at risk for a privacy violation? An even riskier question, do

and should criminals have privacy rights like the general public? Articles that discuss DNA data

banks state that, "The societal value of DNA data banks outweighs the privacy interests of

convicted offenders." Courts usually recognize that once a person has been convicted of a

crime, their privacy rights quickly diminish. In addition, the courts generally feel that since

collection of DNA is non-invasive it is not a huge invasion of privacy. The courts fail to consider

aspects of genetic privacy outside of DNA identification and the retention of the sample.

## *Ethical Issues*

Familial DNA is not the only piece required for an investigation. Other information such as housing, employment, and banking records of the matched family members can be obtained to assist investigators. Basically, by the end of an investigation, an investigator will know everything, from DNA to financials about the potential perpetrators family. The field of forensic science for the purpose of crime solving cares little about anonymity. The ethical conflict is that despite the nature of their crime, even a convicted felon deserves the right to a level of privacy regarding their medical issues. Although the FBI maintains very strict standards with regard to what is entered into CODIS there is a remote possibility that innocent people could inadvertently end up having their DNA stored in police databases.

It is difficult to say no to a process that helps prosecute criminals who have committed heinous crimes even if some states have already chosen to not use familial DNA. Even though certain methods of collecting genetic information are considered legal, many law enforcement entities find privacy to be an obstacle while trying to fight for the common good and enforcement of the law. A New York state district attorney stated in an article, "The failure to use familial searching is insanity. It's disgraceful. If I've got something of scientific value that I can't share because of *imaginary* privacy concerns, it's crazy. That's how we solve crimes." In contrast, opponents of familial DNA say the use of this process results in civil liberties not being protected and that the criminal justice system is quick to sacrifice privacy to promote the well-being of society. They view the expansion of this type of technology as interference from government. Both proponents and opponents are passionate about their causes thus making it difficult to find common ground. However, expanding DNA technology is more likely to result

in a more tangible result, getting criminals off the streets, while not expanding is more abstract when trying to understand the benefits. Both aspects need to be discussed and developed in a cohesive manner and not viewed separately when developing policy around this particular issue.

### **Policy Recommendation**

The field of genomics is making so much progress that research should not be hindered by the threat of privacy invasion. At the same time forensic science should not be hindered by the threat of a lawsuit by someone who deserves to be in jail. DNA is a valuable tool for forensic science when it comes to crime solving. Careful guidelines must be in place in order to create a balance for crime-solvers to be able to do their jobs without fear of technicalities. Citizens have a right to investigate their own genetic material to review their family history or discover any disease predispositions without the fear of their privacy being violated.

It has been well established that the general population is concerned with privacy. What are the limits? How will these limits and boundaries be developed and by whom? Clear guidelines and policy need to be established and enforced so that consumers can have assurance when participating in any type of genetic testing. In order for any of this to occur, stakeholders from all arenas need to be involved. The stakeholders that need to be considered are the general public, policy makers, health care providers, health educators and the criminal justice system, to name a few.

Here are a few guidelines to consider while developing policy around genetic testing and privacy:

- **Assessment:** There needs to be an assessment of what laws and policies currently exist and can be used as a foundation to either create a more comprehensive privacy law in relation to genomics and specifically the various types of genetic testing. A question that also needs to be asked, can current policy be amended to address these issues?
- **Strategy:** A strategy needs to be developed to address what has been assessed. This would require communication between all stakeholders. Also, this is the point where stakeholders need to be involved to develop the who-what-where-when and why of how policy will be established and/or amended.
- **Integration:** How will the policy be implemented throughout the country and through each genetic test?
- **Financial Arrangement:** What budget is required if any? Will a training budget need to be established? This is also important when creating a cost-benefit analysis.
- **Identify Technical Tools/Competencies/Capabilities Required:** What tools can be used for auditing (if needed)? What will this require of health care professionals? Policymakers? Other government officials? Educators?
- **Evaluation:** How will those involved be able to evaluate how the new or amended policy is working? How will desired improvements be addressed and completed?

To conclude, we must ask again, do the benefits outweigh the risks? This question resonates in all topics and discussions surrounding genomics. If genomics ultimately creates a more personalized experience and treatment from a physician and provides an individual with a significantly better health outcome, then all of the potential risks were indeed worth it. As expressed by various testimonies and scenarios, the most prevalent reservation with DNA testing of any sort is privacy and a lack of commitment to addressing these reservations in order to develop policy could hinder any strong advancement of genetic testing.

# CHAPTER 5

## GENOME SEQUENCING

### AND PERSONALIZED MEDICINE

*Jenna Gribbin*

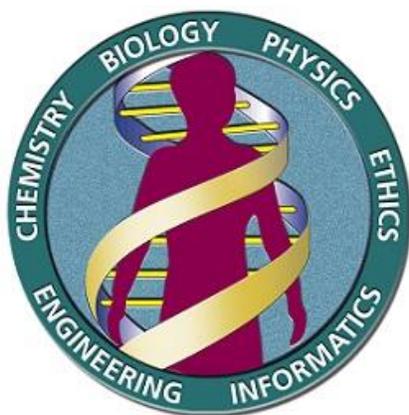
#### Executive Summary

With the rapid development in DNA sequencing feasibility, and its simultaneous reduction in cost, truly [personalized medicine](#) is likely to become a reality for many people.

Genomic data is being generated [faster](#) than physicians are able to apply it to their practices while the ethical debates surrounding this information are continuing at pace. As it becomes easy for the average consumer to access parts of their own genetic code without ever leaving their home, society must start to come up with answers regarding *what should be done* with all this information. It needs to be protected from those who could use it against a patient, such as an insurer or employer, but made available to those doing research, all the while finding a way to create personalized applications in medicine and ultimately fulfills the promise of personalized medicine.

#### Background

The [human genome project](#) sequenced the first human genome, cost two billion dollars,



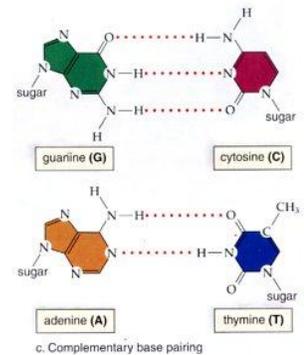
and took a large dedicated staff 13 years to complete the effort. Now the cost of gathering this sequence data has dropped by many orders of magnitude. In the near future, the cost of sequencing an entire human genome will be

under \$1000, making it accessible to the average person and making “personalized” medicine realizable. As we begin to learn more about the human genome, we are finding that it is not simply the nucleotide [base pairs](#) that hold the key to disease, but it is

a complex system of genes, their packaging, and even symbiotic bacteria that influence these processes. The implications of next generation sequencing technology in the healthcare fields are

tremendous. It opens the possibility of personalized medicine based

on a patient’s unique genetic code. In a short [video](#), George Church explains how he is trying to recruit 100,000 people to sequence their genome and aid in the progress of personalized medicine. Yet these advances add another controversial layer of new ethical questions regarding the equitable availability of these services and the privacy of the results.

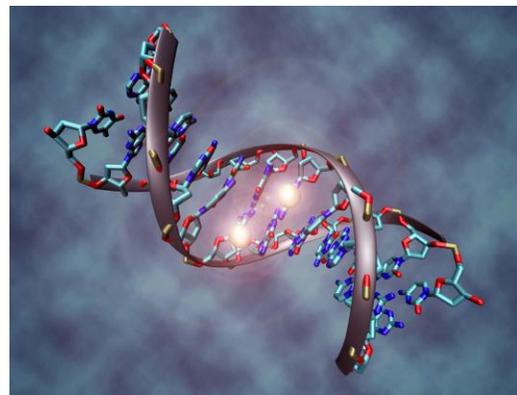


As [sequencing platforms](#) improve, and human genetic data become readily available, we are learning that inheritance of common genetic disease may be more complex than we once thought. Single nucleotide polymorphisms, or [SNPs](#) (pronounced “snips”) are some of the most easily generated sequence data. These single base pair differences can be used to determine differences between humans. SNPs are found within [alleles](#), which are alternate forms of a gene. For instance everyone has “gene” for eye color, but some people have an allele for blue eyes, and some people have an allele for brown eyes. It was originally thought that common genetic diseases would be due to the most common allele variants, but as it turns out this is not the case. While common risk factors can be identified, they do not usually explain a high percentage of the variation of a disease among humans. We are discovering that rare mutations are just as important as common ones. This emphasizes the importance of

*personalized* medicine, as it has become clearer that a “one-size-fits-all” model will not work for treatment of genetic diseases. The advent and subsequent price reductions of more genetic techniques, such as [exome sequencing](#), where only the coding region (genes) of the genome are sequenced, have aided in the search for these rare variations that lead to predictable disease outcomes. As our ability to sequence and [analyze](#) greater portions of an individual’s genome, these variants will become increasingly easy to identify.

### *Current Status of Genome Sequencing and Personalized Medicine*

Cancer is one of the predominant diseases affecting our society (there were 7.6 million cancer deaths worldwide in 2008!), and the fact that it is caused by genetic mutations displays the importance of research in this area. The [Cancer Genome Atlas Project](#) (CGAP) is attempting to “catalogue and discover major cancer-causing genome alterations in large cohorts of human tumors through integrated multi-dimensional analysis”. A resource such as this will ultimately help physicians understand and treat cancer on a molecular level. Cancerous cells are those that have mutations of some form in their genetic code. Understanding these mutations as well as how they are formed and how they interact with markers in our genomes eventually allow for personalized care for cancer patients. Cancer is [not the only disease](#) that will be aided by advances in genome sequencing, but it is the one in which the most research of this type is being done and [applied clinically](#). Joe Gray discusses this topic in his [introductory lecture](#) as the head of the Knight Cancer Institute at Oregon Health and Sciences University.



Cancer is caused by alterations in our DNA, but this is not only limited to base pair changes, as discussed above, it can be due to “epigenetic” changes as well. Epigenetics refers to the way in which DNA is “packaged” (see Chapter 6 for more details). Mammalian genes are packaged in certain ways that determine their expression patterns, and this creates another layer to the gene/disease mystery. The way in which these genes are packaged affects if and when they are expressed. Epigenetics is defined as the study of “heritable changes in a phenotype arising in the absence of alterations in the DNA sequence.” With current technology it is feasible to map the entire epigenome. This knowledge will help us understand the development, cell differentiation, and disease much more in depth than we are currently able to. CGAP is not only taking a close look at genetic sequence, but also recognizes the critical nature of the epigenome in explaining cancers.

In addition to exploring the role of epigenetics, other projects are playing a corollary role in this type of cancer research as well. For instance, the bacteria that co-inhabit our bodies add another layer of complexity to understanding the human genome. This is currently being investigated in the [Human Microbiome project](#). These potentially beneficial microorganisms, which outnumber our own cells ten to one play a variety of roles in human health and disease: from

direct epigenetic modification of our genes, to physical “protection” of our membranes from harmful bacteria. Research suggests that these bacteria play an important role in influencing which genes are expressed in the intestinal wall, and even have a role in the development of a



normal heart. For years the diversity of this vast ecosystem was not known, because we were limited to studies regarding those bacteria that we could culture in a lab. With current sequencing technology, [metagenomic](#) research allows the analysis of the DNA from microbial communities to be sequenced and individual species identified, bypassing the need for cell culture, and allowing us to examine the vast diversity of these symbionts and should eventually allow us to examine their interactions with human diseases. This is just one example of the evolving complexity encountered by researchers on their quest to understanding the human genome and applying it to personalized medicine.

For the prevention of cancer, some people are turning to “direct to consumer” DNA



testing ([23andMe](#)). With this type of testing, any consumer can send in a saliva sample and have their most common SNPs identified for them. This poses a large risk to both the consumer and his or her health care providers. First, as discussed above, many of the common diseases are actually

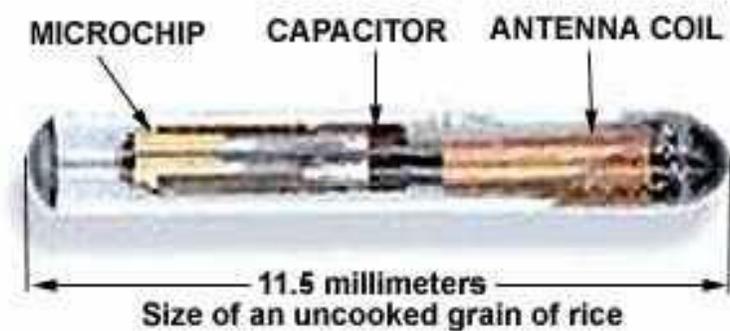
due to rare gene variants instead of common ones, which these tests may not have the sensitivity to identify. Secondly, they are not interpreted in-person or by the patient’s physician.

This creates the potential risk for misunderstanding and misguided complacency or fear regarding a genetic predisposition. Thirdly, a customer may want their own physician to [interpret these tests](#), which may not be possible due to the physician’s specific training or due to ethical or legal concerns regarding interpreting a test he or she did not order.

*What does the future hold?*

Eventually, just as it is current practice to have a patient's blood type or specific allergies on file, it will be commonplace to have not only a patient's own genome, but also that of their

## COMPONENTS OF THE BIOCHIP



microbiome and their epigenome accessible to healthcare professionals (perhaps even packaged nicely in a "biochip"). This will hopefully bring us to a place where the idea of truly personalized medicine can become a reality. While this will likely bring

improvement to clinical treatment practices, it will also bring its own set of thorny ethical and policy concerns. Many of these concerns are already present even while this field is in its relative infancy. Their importance will only increase with the rapid increase in knowledge and implementation of this type of medicine.

### *Ethical and societal Implications*

First, with the volume of data being generated, a logical question is how will it be stored and who will have access to it? This question is already

being negotiated, and with sequence data becoming increasingly abundant, these concerns will continue to grow. There is potential for discrimination, based on undesirable genes, from insurance companies or



employers. Therefore, privacy is extremely important. Not only is this a question after the data

is generated, but also when it comes to [biobanking](#) for the potential future use of a specimen (e.g. blood or tissue sample) for subsequent research purposes. Additionally, we will not only need to be able to protect this data, but preferably share it with other research laboratories. This requires [IRBs](#) and the development of new data-sharing standards to deal with this question as well as patients/participants who are informed and willing to share personal data. The Genetic Information Nondiscrimination Act, or [GINA](#), begins to address some of these privacy concerns, but it is currently incomplete to cover all areas of genomics, particularly epigenomics, in its current state. For instance, GINA protects people from *predictions* that their genes make about their health, but if and when they develop that particular health outcome, GINA no longer protects them.

Secondly, we will face the continuing question of equity in health care (as described in Chapter 2). Presumably the cost of this type of care will be high, and therefore only some people will be able to afford it. Right now there are many people in our country (and around the world) who can afford little to no health care, and the increasing costs associated with this level of personalized care has the potential to widen this gap even further. Will insurance companies be expected to reimburse for this type of care, or will it be the government's responsibility to pay for it in the first place? Do we have a social responsibility to care about the health of all people in our country? What about in the world? These are not new questions and they are not unique to P4 medicine, but they will be exaggerated in light of this new shift in the current US medical care.

Finally, we need to consider that the general public is not likely to understand the science behind new discoveries in genomics, nor the ethical and social implications of its

application to clinical care. This creates a problem in trying to convey the importance of the research, i.e. why we should fund it, and the relevance it may or may not have to personal health. Personalized medicine implies that patients themselves will be an integral part of the medical care they receive, but this will be hard to achieve given the lack of knowledge and understanding of this subject by most people. This problem is something that will need to be seriously considered throughout the policy-making process regarding genome sequencing and personalized medicine.

### **Policy Considerations**

Health and Human Services Secretary Michael Leavitt suggests four policy goals regarding personalized medicine:

1. Find relationships between genetics and disease that can be put into practice
2. Prevent insurers or employers from using genetic data to discriminate against individuals with pre-dispositions to disease
3. Ensure genetic testing is accurate and useful
4. Create standards to ensure data sharing.

It is necessary to also include in this list:

5. The reduction of health care disparities
6. Education of the public

These six goals (4 proximate and 2 longer term) should be used to guide policy decisions and development and implementation of personalized medicine

*1. Find real-time biological connections between genetics and disease that can be put into practice.*

This has already been started with things such as the Cancer Genome Atlas Project and the Human Microbiome Project, but there is a great amount of further research in all of these

fields to get to a spot where findings can be turned into viable treatments. This highlights the importance of continued funding and social support for this type of research. By placing a social value on research in this field and showing that with funding dollars, America will be able to stay at the forefront of this type of medical discovery, and we will be able to decrease the time from discovery to implementation of these advances in a clinical setting. When research-funding legislation is presented, the implications of this type of clinical research should be considered, and it should be remembered that results could affect large portions of the population. The federal government is attempting to [maintain support](#) for this type of research, and it is imperative that it is done at the state level as well.

*2. Prevent insurers and employers from using genetic data to discriminate against individuals with predisposition to disease.*

Just as some insurers and employers currently may want to discriminate based on pre-existing conditions, they will also likely want to do so based on genetic pre-dispositions as this type of data becomes increasingly abundant. This is an area where policy is needed to define the regulations around who has access to genetic data and how it can be used. GINA is a good start to this work, but more legislation will be needed to ensure all types of genetic data (epigenome and microbiome data etc.) are equally protected. There is currently no legislation to protect these “[other](#)” types of genetic data from being used to discriminate against someone. Legislation will need to be drafted to either modify GINA at the state and national level, or create a new policy to protect this type of information. It took thirteen years to get GINA passed, and it’s imperative this is not repeated with other genetic privacy bills.

*3. Ensure genetic testing is accurate and useful.*

This means that testing and interpretation by health care professionals needs to be available. This creates the need for trained healthcare professionals, such as genetic counselors, to be widely employed and a common part of a medical practice. It also creates the need to limit the amount of “diagnoses” and treatment advice private companies are allowed to provide to patients based on incomplete genetic data. Additionally, consumers need to have appropriate warnings regarding direct-to-consumer testing, and they must be made aware of the limitations of this type of test. Even when companies are trying to provide non-biased genomic analysis, their work will still be framed by the consumer’s interests, not that of a health care professional. This issue is certainly on the radar of the [federal government](#), but it needs to be addressed in an aggressive manor at all levels of government. There is no current legislation in this area, but the [Genetics and Public Policy Center](#) does keep up to date on these issues, and potential policy considerations.

#### *4. Create standards to enable data sharing.*

While privacy is a concern for those having genetic testing done, there is also a huge wealth of medically useful information contained within this data. Not only do we need to ensure privacy and non-discrimination based on this data, it is critically important to find a way to share all generated data among researchers and enable further progress in this form of research. Money and intellectual resources should be devoted to establishing standards for data sharing that make it accessible to the greatest number of researchers. This could be done by developing reliable methods by which to share confidential data, or by using research protocols that involve informed consent of patients who are willing to “share” this information and these methods should be developed with the help of local IRBs.

### *5. Reduce health Care disparities.*

There are great disparities within our country surrounding the access to and quality of health care (as discussed in chapter 2 of this briefing book). With this new, and relatively costly field, there is the potential for these disparities to grow substantially as the cost makes them inaccessible to all but the wealthiest of consumers. It is our social and political responsibility to make sure that this knowledge doesn't widen the gap between the "haves" and the "have-nots" within the context of health care. It is imperative that legislation continues to be enacted that provides health care to all citizens. As the cost of health care continues to rise, especially in light of these expensive developments, more money and effort is going to need to be devoted to solving the problem of creating access to reasonable health care for all people. This is particularly relevant to cancer research, as not only are people with fewer resources going to have less access to cancer treatment developments but, in general, these disenfranchised people are more likely to be exposed to carcinogens in the first place.

### *6. Education of the public*

To get to the root of this problem, science literacy and education in [cell biology](#) and molecular biology should be improved in primary and secondary schools nationwide. The general public needs to be sufficiently familiar with science to be able to have a conversation with their doctor regarding personal health. Discussions are being had to address [science education standards](#) and making a case for including basic biology relevant to personalized medicine is necessary. Additionally scientific and clinical research needs to be reported more regularly to the public, instead of being confined to scientific journals. This will provided the public with more up to date information on topics such as personalized medicine. Amy

Harmon, a reporter for the New York Times gave a speech discussing this topic that can be seen [here](#). While this is a tall order and there are many barriers to both of these suggestions, they need to be implemented for our society to be able to fully utilize personalized medicine.

#### *Potential Stakeholders*

Everyone has potential to gain from the sequence data being generated and the research being done within the field of P4 medicine. This form of medicine is on track to be the way of the future. All members of society will be faced with answering questions regarding how to use and store this information as well as who has to pay for it, and how it can be realistically applied to health practices.

It is likely that insurance companies will be hesitant to cover these expensive procedures. While conversely they will want the knowledge of genetic data to determine who is risky to insure. These specific stakeholders will need to be included while considering the implementation of policies and legislation regarding personalized medicine.

The scientific community also has a lot to gain by performing well in this area. Medical and biologic research can become a greater part of our economy, as we look to sustainable job markets. It is also important that Oregon, and the United States, stay on the forefront of this type of research and that we maintain the ability to provide our citizens with this highest standard of care.

## Conclusions

It is clear that medicine is heading in the direction of personalized treatment. With the availability of sequence data at relatively low costs researchers are advancing in leaps and bounds with regard to the genetic basis of human disease. It is only a matter of time before these findings can be implemented to personalized treatment. Policy makers need to pave the way for this type of research to continue and enable access to these technological advances for the general public.

In the short term, resources need to be provided for this type of sequence-based research to continue, while concurrently putting methods into place to alert consumers to the potential of receiving incomplete or misleading information from direct to consumer tests. Privacy protection measures need to be implemented while enabling data sharing between those doing the research. Finally, as a long-term goal, health care disparities need to be diminished, so this level of health care can be accessible to all of our citizens.

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# CHAPTER 6

## EPIGENOMICS AND NUTRITION

*Emily Carter*

### Executive Summary

The saying goes: “you are what you eat” ....but what if it were actually: “you are what your grandparents ate?” Until recently we thought that we only inherited the DNA that was passed down from generation to generation, but the study of epigenetics has taught us that it is much more [complex](#). Environmental exposures, such as what we eat, can change the way in which our DNA is expressed, and those changes can also be inherited. Epigenetics is the process by which environmental exposures modify the expression of certain genes leading to an alteration in the [phenotypic](#) characteristics of the individual. These changes are important to the application of [P4 medicine](#) because they are modifiable and therefore potentially targets for new means of prevention.

P4 medicine will provide a new, more effective means of addressing current health care issues. One of the most serious issues facing our current healthsystem is [obesity](#). Over [60% of Oregonians](#) are [obese or overweight](#), and that number will continue to rise if significant changes are not made. Having an understanding of the epigenetic consequences of this obesity epidemic is essential to comprehend the gravity of this threat to the public’s health. Epigenetic changes are occurring to the DNA being passed down to future generations because of overeating. It is unclear what the exact effect will be to future generations, but currently the [SB 931: Oregon Obesity Prevention Task Force](#) estimates that obesity costs Oregon more than

\$781 million a year, and this number will continue to rise as even more individuals become obese.

## **Background**

### *Introduction*

As discussed in previous chapters, genetic susceptibility plays a significant role in the development of disease, but what about the environment?

Prevention of most chronic diseases is focused on lifestyle modification and changes in environmental exposure. Recent discoveries have shown that nature and nurture are not truly separate, but instead affect one another directly.

Environmental changes can affect the expression of certain genes changing the physical characteristics of an individual.

Epigenetics is the process by which this modification is made to the genome (inherited DNA) without changing the DNA

sequence. Epigenetics is the study of changes to specific genes or a sequence of genes, while epigenomics is a higher level analysis of epigenetic changes across the genome. Epigenetic modifications affect whether, when and how genes are expressed. Understanding epigenetics is important for public health policy makers because the alteration of the epigenome can be inherited and therefore affect disease risk of future generations.

### *Science*

Each cell in the body contains the same genetic material, but the specific characteristics (phenotype) that are expressed by these cells differ. Tissue-specific genes are activated in each

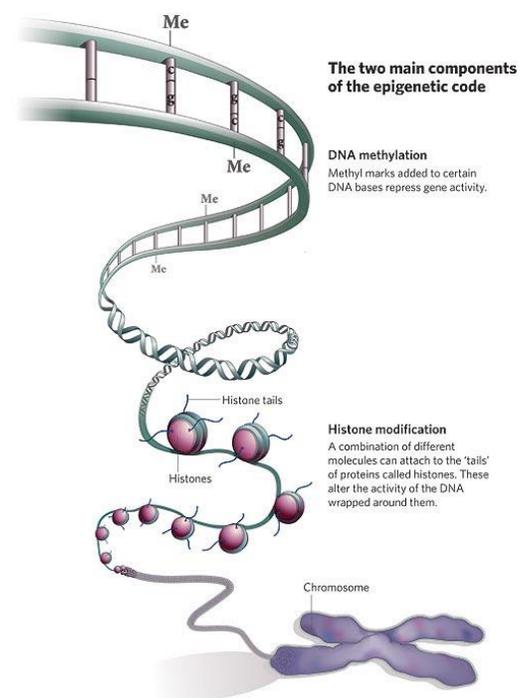


cell, while unnecessary genes are silenced through [epigenetic modification](#). Epigenetic modifications are inheritable modifications to the expression of genetic material contained within the cell's DNA. There are three mechanisms by which a gene can be activated or silenced:

- Methylation of the carbon ring of cytosine base in the gene
- Modification (acetylation, methylation or phosphorylation) of the amino terminal end of the histone tails in the nucleosomes (around which the DNA is coiled)
- Specific binding of small nuclear RNA

These genetic modifications are sufficiently stable to be transmitted to the next generation of cells through both mitosis (cell division) and meiosis (sexual reproduction). Therefore, these modifications can be passed down to the next generation.

The potentially most important aspect of epigenomic function for the purpose of public health is that that it provides an explanation of how human genome is modified in response to environmental changes. This blurs the line between nature and nurture. Many recent discoveries have uncovered the mechanisms by which modifications are passed down through the germ line. This can influence the health of future generations. The epigenome is particularly sensitive to changes in the environment during the development of the organism. The developing organism uses epigenetic function as a way to appropriately mould expression of genes to respond to its anticipated environment. Specifically, we know that factors such as diet, lifestyle and



environmental exposures can cause epigenetic changes leading to modification in physical development of the individual.

Since we know the epigenome is most vulnerable to changes during development, the



affects of environmental conditions on the fetus in-utero have been the focus of much of the research on

epigenetics. There is strong evidence that [predispositions to certain adult onset diseases](#) such as Type 2 diabetes and obesity are caused by epigenetic changes during fetal

development. This new disease model paradigm is referred to as “early origins” or “fetal basis” for disease. One specific model of this theory is genetic predisposition to obesity. This has been named the “thrifty” phenotype in which an individual develops in an environment of nutritional scarcity and therefore the metabolism of the individual is epigenetically encoded for a “thrifty” phenotype. This allows the individual to maximize energy production from limited food supply. When these individuals then have an abundance of resources and are epigenetically programmed to conserve energy there is a mismatch between their environment and their “thrifty” phenotype, which leads to a high risk of developing obesity, type two diabetes and other disease sequelae. In addition, scientists have shown that these epigenetic changes can be passed down to subsequent generations and affect development of chronic disease in their progeny.

### *Ethics*

Unlike the DNA of the genome, the epigenome is environmentally modifiable. This provides opportunity for improvement of the genome, such as novel therapies, but also

potential vulnerabilities which can be passed down to future generations. Epigenetics has shown significant ways in which environmental exposures (diet, lifestyle and toxins) can lead to alterations in phenotypic expression. Epigenetics is important from a public health policy perspective because the epigenome is responsive to environmental influences. Therefore we have an opportunity and responsibility to protect the human genome from environmental insults. Current preventive health initiatives focus on prevention of chronic disease through lifestyle changes. Epigenomic research would suggest that this is even more important than initially understood because poor health behaviors may actually modify the expression of certain genes, and eventually be passed down to future generations.

Although clinical epigenetic testing and therapy is still a thing of the future, it is important for policy makers to be thinking of these issues now, because ethics and law often lag far behind novel therapies. Science has shown that environmental insults to the developing fetus *in-utero* can significantly change the physical outcome of the child, and possibly for generations after that as well. One of the current crises in healthcare is the obesity epidemic, particularly among children. Based on our current knowledge of epigenomics, it is clear that poor diet of the current generation of children will adversely affect humans for generations to come. Therefore it is even more of society's responsibility to address this issue due to multigenerational dimension of obesity and adverse health effects. [Dr. Mark Rothstein](#), a prominent professor at Duke University in bioethics, genetics and public health law, describes four main ethical issues surrounding epigenomics that society will need to address in the near future:

- environmental justice
- intergenerational effects and equity
- privacy and confidentiality
- equitable access to healthcare

**Environmental Justice** is important in epigenetics because damaging environmental exposures are more likely to occur among low income individuals. Socioeconomic affects status nutrition, work exposures and health behaviors. Obesity has been known to be associated with low income, poor nutrition, and psychosocial stressors. Based on the understanding of epigenetics it is likely that the mechanism of association between these exposures and obesity are more complex than, “if you eat junk food you will get fat”. As discussed above environmental exposures have likely changed the epigenome of our population. Therefore epigenetics has the potential to help us understand how these stressors affect the incidence of obesity and other co-morbidities, and potentially help address how these disparities affect the epigenetic coding of individuals genome.

**Intergenerational effects and equity** is the obligation of each generation to serve as a custodian or steward of the planet and its inhabitants for future generations. In the case of epigenomics this means preventing damage to the human genome. The obesity epidemic in the United States is not only damaging the health of these children, but it is causing harm to the epigenome and therefore many generations to come. Because of the obesity epidemic and all of the sequelae related to obesity, this generation of children is anticipated to be the first generation in the US to have a [shorter life-span](#) than their parents. What will happen with their offspring? How will the lack of intervention now affect the health of future generations?

**Privacy and confidentiality** is important in epigenomics as it is with all sensitive health information. The concern with epigenomics in particular is that it is not protected under the current genetic privacy laws. Although there are not currently any epigenetic screening tests, these are not far down the pipeline. As health records become electronic and therefore permanent, it becomes extremely important to protect that information, especially when it could potentially impact the privacy of whole families, as well as individuals.

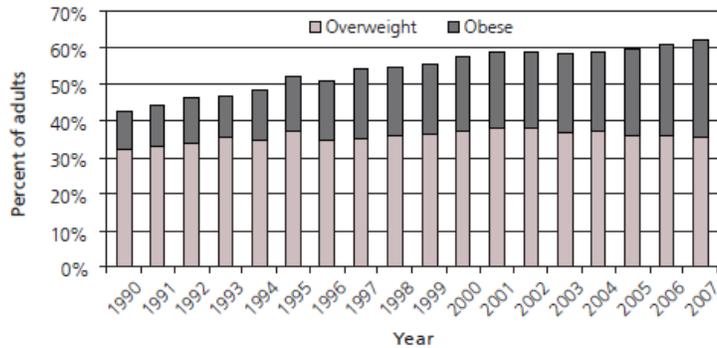
The final ethical consideration for epigenomics is **equitable access to healthcare** (as described in chapter 2). As mentioned in environmental justice, individuals with lower socioeconomic status are more likely to be exposed to damaging environmental conditions, and are less likely to have private health insurance. With the advent of requirements for healthcare coverage it is likely that an even greater number of low income individuals will be reliant on public health insurance providers such as the [Oregon Health Plan \(OHP\)](#). Public insurers, including OHP, have been slow to adopt coverage for sophisticated and expensive new tests. This will likely be no different for epigenetic testing. Therefore those affected groups are most likely to have deleterious epigenetic modifications are the ones most likely to have difficulty finding coverage for these tests. As the mechanism by which we cover our population changes over the next few years we should be considering the impacts of new technology such as epigenetic testing and our obligation to provide coverage for those novel tests and therapies.

## Policy Recommendations

### Background

Obesity is one of the greatest threats to today's healthcare system. Over 60% of Oregonians are [overweight or obese](#), with serious sequelae of premature death, non-alcohol induced fatty liver disease, cardiovascular

Percentage who were overweight or obese among Oregon adults, by year, 1990-2007



Source: Oregon Behavioral Risk Factor Surveillance System

disease, stroke, and Type 2 diabetes. In addition, about a quarter of Oregon 8th graders are overweight. The increase in obesity among children has led to an exponential increase in the number of children developing traditionally “adult onset” disease such as Type 2 diabetes which has now been diagnosed in children as young as 5 or 6. Since we now know that nutrition can influence epigenetic events, altering gene expression, and potentially modifying disease risk for the individuals and their future children. Currently there are no tests or therapies that can be used to identify and potentially treat those with a higher risk of developing obesity. Until this happens, public health will need to continue to focus attention on prevention of obesity through lifestyle modification.

In Oregon we are currently investing \$10 million per year in obesity prevention. The focus of this money should be on prevention of obesity in women of childbearing age and children. By focusing our very limited resources on these two populations we have the potential to limit the spread of this epidemic to future generations through epigenetic modification passed on to offspring. The most important area of focus on prevention for these

two populations is nutrition – particularly since diet has been shown in multiple studies to have such an important affect on healthy fetal development, not just during pregnancy, but prior to conception as well. In addition, we should invest in research on epigenetic markers and/or causes of obesity. Eventually this research could potentially lead to an epigenetic screening test for obesity.

### *Current Status*

In 2007, the Oregon Legislature developed the SB931 Obesity Prevention Task Force to develop specific policy recommendations for the state. The role of this task for was to decide how to allocate monies to evidence-based obesity prevention and education programs. They have focused on communities as a whole, as well on three specific areas: schools, worksites and health systems. Schools should be receiving a majority of this funding as children are the most likely to respond to these interventions and education. Women of child-bearing age are not explicitly targeted in the current recommendations. This should be a more explicit objective as their health is most likely to affect the health of future generations.

### *Stakeholders*

The first, and possibly most important stakeholder is **children**. It is almost unfathomable what affect the exponential rise in the number of children with obesity will do to the health of that generation. Children have the most to gain from interventions and are also the most likely to adapt to these prevention measures.



**Society** in general has a lot to lose if this epidemic is not reversed. It will cost money, more than \$10 million per year, to make significant change, but reducing the prevalence of obesity could significantly improve the health of our communities.

We may be damaging the health of **future generations**. Based on what we currently know about epigenetics, we are successfully damaging the epigenome that we are passing down to future generations. By preventing exposure to poor nutrition we could improve the health of future generations as well as our own.

Investment in the study of epigenetics provides **the scientific community** with a new frontier for scientific innovation. Even more so than genetics, epigenetics provides a potential area for development of interventional therapy. Therefore, genomic science should focus efforts in this area, particularly with regards to obesity, which is becoming so prevalent.

#### *Costs*

It is estimated that medical costs related to obesity currently exceed \$781 million annually in Oregon. These costs will continue to balloon unless we make significant changes to our population's health. Based on what we know about epigenetics, the costs are likely to continue to rise as unhealthy, obese women, go on to have children with epigenetic modifications that put them at a higher risk of developing obesity as well.

## Conclusion

Epigenetics provides a window into the interface between the environment and genetics. Scientific advancement has not yet provided a mechanism to alter these interactions, but this knowledge has given us further evidence of the importance of prevention of chronic disease. The theory of fetal origin of disease will help us more appropriately target prevention. In the case of obesity, prevention focused on children and women of childbearing age will help reduce disease burden in future generations as well as providing some decrease in co-morbidities for these individuals. Continuing investment in epigenetics research could potentially provide a new mechanism of screening and prevention in the world of P4 medicine.

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<http://www.youtube.com/watch?v=d9SB0teMQo0> (Part 3)

<http://www.youtube.com/watch?v=Kc7LXkuebCl> (Part 4)

<http://www.youtube.com/watch?v=pEGDaBcplIQ> (Part 5)

## CHAPTER 7

# THE GENOMICS HEALTH POLICY PROCESS: A MODEL OF DELIBERATIVE DEMOCRACY

*Dr. Greg Fowler*

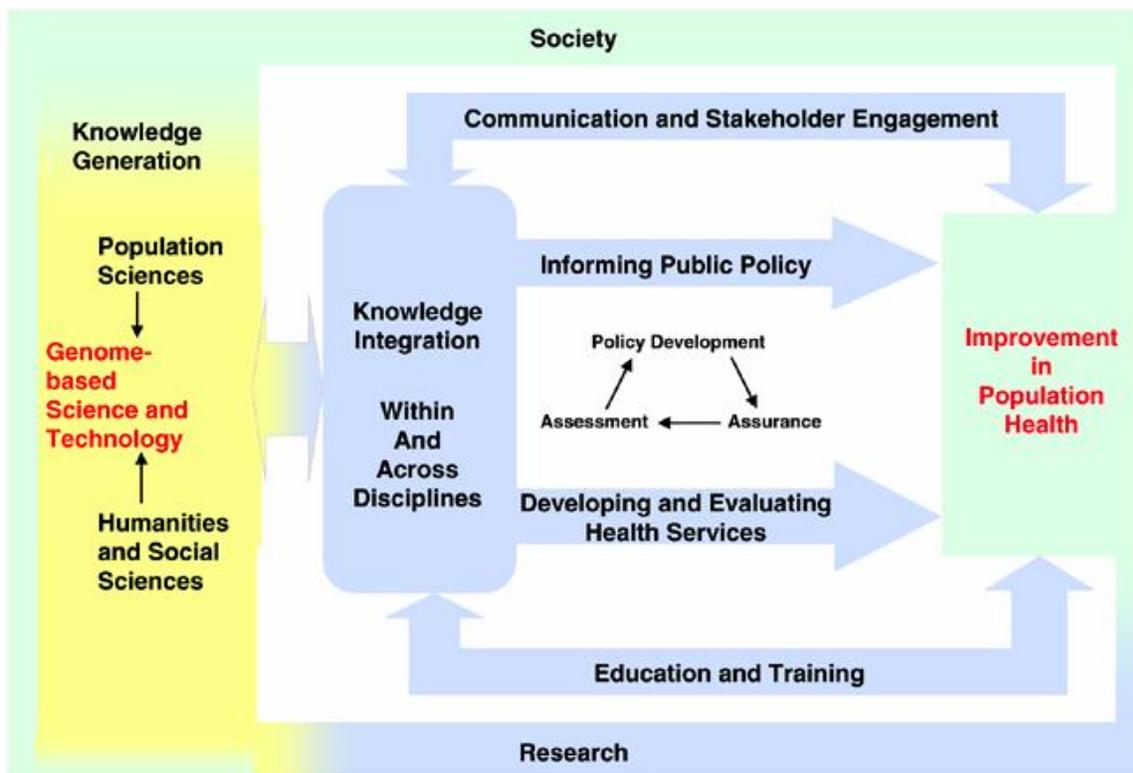
### **Why *Genomics* Health Policy?**

The last eleven years has witnessed a breathtaking acceleration in genome science, the process of looking at hundreds, if not thousands, of genetic interactions simultaneously in order to understand the root causes of diseases and to better understand how an organism works. Genomics is a synthesis of many disparate and diverse fields, including biology, public health, engineering, computer science, and mathematics, all working to map the interactions between thousands of genes and their cellular and extracellular environment.

The social sciences and humanities are also integral components of the genomics revolution as ethicists and legislators create policies and laws that will guide the integration of genomics into scientific practice and health care. Today, genomics has yielded the richest source of [biological data](#) we have ever known.

Public mistrust of the scientific establishment tells us that attention must be paid to the way in which genomic knowledge and expertise is expressed, heard and acted upon. There is concern that new genomic technologies might be used in ethically and socially unacceptable ways with the public left powerless to resist their advance. On the other hand, advances in genomics offer the potential to transform health care. Such tensions at the intersection of core human values and technology, if unaddressed, impede research and reduce the ability of the scientific enterprise to serve society.

In April 2005, an international expert workshop convened in Bellagio, Italy to “translate genome-based science and technology into improvement in population health.” In their final report, the conferees recommended promoting a public policy development process that uses structured activities involving both the general public and scientific experts, an international and interdisciplinary partnership approach that “will offer unprecedented opportunities for improved disease prevention and therapy.”



Specifically, the key outcomes of this international forum for dialogue and collaboration are five-fold: 1) to promote relevant research; 2) to support the development of an integrated knowledge base; 3) to promote education and training; 4) to encourage communication and engagement with the public and other stakeholders; and 5) to inform public policy.

### **Why a *public policy* approach?**

Policy makers who seek public input should understand the nature and limits of the information they seek. At different points in the policy development and decision making process, they will want to...

- Assess a spectrum of community values;
- Understand technical data about the options under consideration;
- Provide rational structure to the policy problem;
- Evaluate alternative policies under consideration; and
- Determine how the power forces in society are aligning on the issues being considered

Involving the general public in complex public policy issues works best when, as a matter of empirical fact, policy makers, the public, and technical experts engage with one another as *partners in a common endeavor*. All three are members of the same community. But they have different roles to play in serving the common good.

- The general public articulates community values.
- Experts identify relevant facts and probabilities.
- Policy makers combine community values and the best available data in choosing among various alternatives.

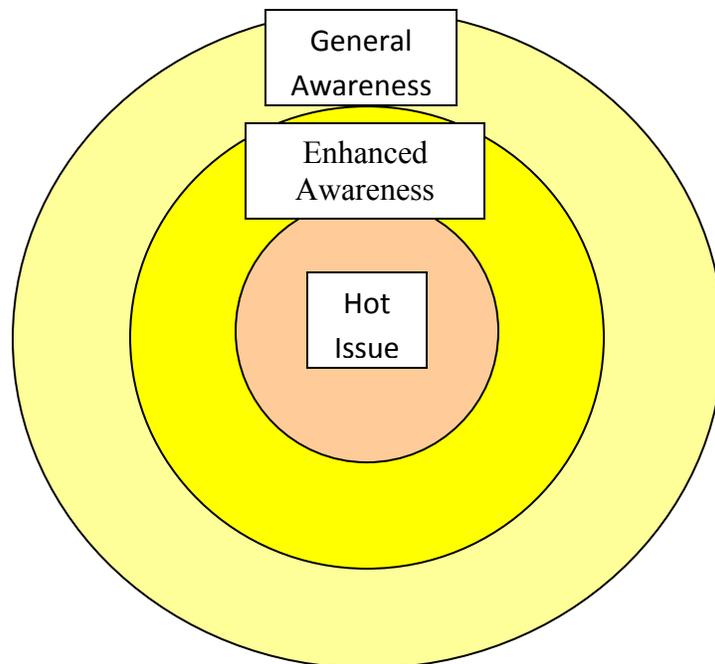
Specifically, citizen participation contributes to the realization of three important goals:

- The normative rationale of deliberation gives meaning to democracy in policy formation and implementation.
- Value input from a broad range of the public can get decision makers to think outside the box and enhance the quality of the decision process.

- Participation plays an important educational and psychological role in the social development of the individual citizen.

The lack of scientific knowledge amongst the general public often leads policy makers to rely solely upon expert input and omit or trivialize the ordinary citizen’s role in policy development. Indeed, public perceptions and beliefs that run counter to *de facto* expert knowledge are not acceptable justifications for public policies. However, citizens do not need sophisticated scientific knowledge about technical genomics in order to play a valuable consultative role in the policy process. Their role is to articulate the values that inform the common good of their community. Moreover, there is evidence that exclusive use of expert input can actually *exacerbate* public conflicts.

**How might one think about “public engagement” (citizen input) in genomic (health) policy formation?**



Health policy formation moves through a cyclic process. Most of the time, only minor policy issues are being worked on. This can be thought of as the “**General Awareness**” phase.

During this period, public engagement focuses mostly on general education and advocacy by interest groups with a mission of public engagement on behalf of the common good.

Such organizations can also create vehicles for building general public awareness about the health care system's positive and negative aspects. Conferences, regional meetings, surveys, focus group, media campaigns, blogging, and other information exchange methods are all useful for this work. The focus is on helping develop general awareness that aids members of the community to function more successfully in the health care arena as *consumers*--and in the public policy arena as *citizens*--who interpret political rhetoric and who vote for or against people, parties, and proposals.

Specific advocacy groups also have an ongoing need for strategizing, message framing, and networking among potential collaborators. Public engagement at this level is indirect and preparatory. It can improve consumer behavior, advocacy, and voter behavior.

Democratic systems often call on citizens to volunteer their time on committees and task forces related to the functioning of the health care system, or to work on specific health policy problems. Civic organizations with a mission of public engagement in health policy serve a valuable role by organizing opportunities for community members to gain a higher level of understanding (“**Enhanced Awareness**”) of the structure and functioning of the health care system, how it can be evaluated and improved, and how various political ideologies interact with the balance of stability and change. Public engagement at this level is also indirect and preparatory.

Occasionally, major changes are “on the table” (or being contemplated, designed, implemented, or evaluated by specific public policy entities), indicated as the “**Hot Issue**” zone.

The effort to accomplish serious change creates the opportunity to structure a direct link between members of the public and policy leaders in the very process of designing, implementing, or evaluating a major new policy-rich arena of science and technology (e.g., genomics).

Community groups seeking to facilitate this interplay between the public and policy makers use conferences, community meetings, focus groups, surveys, and town hall events. These methods are designed to gather information for policy makers who actively seek input from the community relative to specific policy related questions.

### **Does Oregon have a history of *public* policy decision-making?**

In Oregon, [Geneforum](#) and [Oregon Health Decisions](#) have acted in consulting roles with state policy makers to generate citizen input about values for public policy decision-making. In the process, both organizations (for 10 and 30 years, respectively) have explored the full continuum of possibilities for incorporating public value judgments into public policy decision making, from simply providing a list of values for stimulating decision makers to think outside the box to building a decision-analytic model for combining public values and expert value judgments to yield evaluations of policy alternatives.

We call it the “[Public Policy Partnership Model](#).” The essential feature of this model is the separation of value judgments from factual data. Deciding *what* is important—and *why* it is important--requires value judgments. Value judgments are necessary to construct the fundamental objectives of public policy.

Deciding how to achieve a higher-level objective requires factual knowledge. Expert knowledge of facts is critical for designing the means to achieve the valued outcome.

Both of these kinds of knowledge need to be used by the policy maker to fashion appropriate public policy in the common good.

### **How does the model work?**

The model can be combined with various group process techniques, such as focus groups, town hall meetings, or community meetings.



These various approaches yield qualitative data to aid the policy maker's intuitive understanding of the values information relevant to the issue at hand. The model can also be combined with quantitative survey methods to assess the distribution and intensity of values held by members of a given community.

### **How does the policy maker get the ball rolling?**

On the public side of the discourse, the process begins with the policy maker's desire to

understand the community values relevant to the particular policy issue at stake. The key to this discourse mode is the question, “What is important to you about X?” followed by “Why?” asked recursively to move from superficial preferences to more stable core values.

Properly designed meetings can generate public input that flows from questions that encourage wise discussion among a broad cross-section of the public.

For example, with regard to the many social and ethical issues emerging from the application of new discoveries in genomics--

- How private should genetic information be?
- Should it be exclusively under the control of the individual? The family? The human race? Pharmaceutical companies?
- Who should have access to information about a person’s genome? Private individuals or families? The State?

These are not factual matters to be decided by experts, but rather are matters related to the values held by the community.

### **Why is “expert input” a necessary part of the partnership model?**

Expert information, on the other hand, is evidence-based data about issues. Technical discourse relies on established norms for determining the most plausible evidence. Here the policy maker needs to hear and understand the strength of the evidence that a given explanation of an issue is the best one available. It is a common practice for the policy maker to commission technical reports from appropriate expert consultants.

A difficult and critically important task of the policy maker is to guide the technical experts to focus on those specific aspects of their field of expertise relevant to the issue at hand and not

simply impose their own values in place of technical analysis.

**In what sense is a policy maker a “receptor site”?**

In this partnership model the policy maker is a “receptor site.” The policy decision maker—a committee, commission, legislature, board of directors, or negotiation team—must be committed to combine the public’s input about values with expert information about facts into a coherent policy. A well-prepared receptor site will optimize this outcome by--

- Involving the decision makers in the design of the information-gathering process, and--
- Coming to agreement with them on how they can best bring the information obtained to bear upon deciding among policy alternatives.



In Oregon, Geneforum has used the partnership process described above to work with the [Oregon Advisory Committee on Genetic Privacy and Research \(ACGPR\)](#), a permanent advisory committee of citizens, experts, and legislators charged with monitoring genetic research and privacy throughout the state.

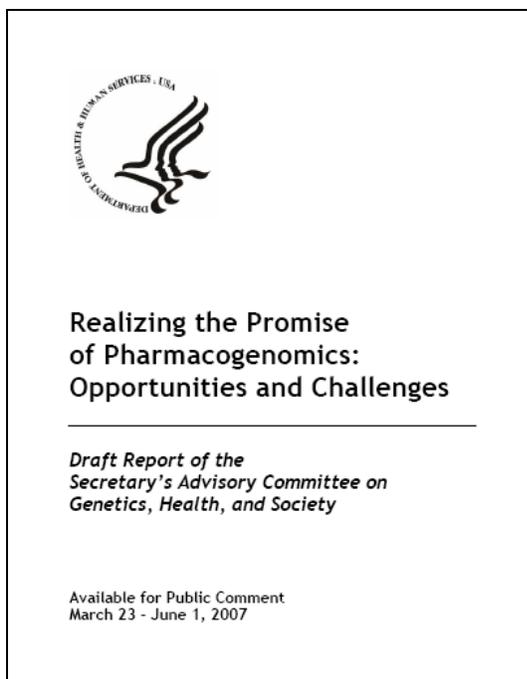
The ACGPR fits the definition of a “policy maker” in the public policy partnership model. Its current mandate includes the obligation “to create opportunities for public education and elicit public input on the scientific, legal, and ethical development within the fields of genetic privacy and research.”

### **Does the partnership model work?**

Using focus group design and community meeting formats, qualitative data about the values held by Oregonians about genetic privacy and an assortment of related issues, has been incorporated into a number Oregon revised statutes (e.g., ORS 192.531-540; ORS 743.730) and administrative rules (e.g., OAR 333-025-0155 and OAR 333-025-0165).

In 2008, Oregon Health Decisions arranged meetings with more than 1,000 Oregonians in 15 communities around the state to hear what Oregonians think and value about health and health care. This notwithstanding, the public consultation process we see as a valuable response to an objective need has not become habitual among policy makers, in general, in particular outside of Oregon and at the Federal level. The most common approach is to consult with experts, select among policy options, and invite public comment on a near final draft of a policy statement.

As a case-in-point, in March 2007—and acting on its mandate—the [Secretary’s Advisory Committee on Genetics, Health and Society \(SACGHS\)](#) solicited public comments on its draft report on the promise, opportunities and challenges of pharmacogenomics, a branch of genetics which deals with the genetic variability in individual responses to drugs and drug metabolism



The committee received 58 comments: 53 from subject matter experts and only five from the public at large. While there may be many reasons for this disparity, the figures nonetheless suggest that U.S. policy decisions regarding genome-based research and applications reside distantly from the partnership advocated in Bellagio, and isolated still largely within the domain of experts.

More immediately, on June 21, 2011, the Centers for Disease Control of the Department of Health and Human Services issued a call for public comment on “Assessing the Current Research, Policy and Practice Environment in Public Health Genomics” with the purpose of assessing “the most important steps for public health genomics in the next five years “ (2012-2017) with public comments solicited through August 1, 2011.

The CDC established an Office of Public Health Genomics in 1997 to focus on:

*“Conducting population-based genomic research, assessing the role of family health history in disease risk and prevention, supporting a systematic process for evaluating genetic tests, translating genomics into public health research and*

*programs, and strengthening capacity for public health genomics in disease prevention programs.”*

The [order and scope of queries in the call for public comment](#) is characteristic:

- 1) Identifying the most important activities to be carried out by the public health system to apply genomic knowledge to public health goals;
- 2) Identifying public health-specific outcomes to be achieved by this;
- 3) Identifying policies needed to achieve these outcomes;
- 4) “What institutions, organizations, and agencies need to participate in achieving these outcomes and what roles should they play?” and
- 5) barriers anticipated to achieving those outcomes and how to overcome them.

Is anything missing in this approach?

While some public engagement initiatives have been funded by HHS, here engagement with “outside entities” (lay citizens notably *unmentioned*) is framed in terms of those entities being instruments of the already established goals, targeted outcomes, and policies.

In this regard, Oregon has been far ahead of HHS in seriously engaging— and more importantly—benefitting from the input of citizenry in considering genomic policy as a good for and of the public.

## What is needed to insure equitable health care policy in the Age of Genomics?

Regularizing an alternative model of public consultation is critically needed, one in which citizens, experts, and policy makers are partnered in a coordinated process that can effectively implement 1) the appropriate impact of *expert* input; 2) the appropriate impact of *public* input, and 3) a process by which expert input, public input, and policy perspectives are combined to yield a choice among policy alternatives.

Achieving this three-pronged goal will be possible when the genomic policy process ensures that citizen input is reflective of the larger community, not simply the aspirations of vocal interests.

By using both public values and expert knowledge in an intentional, collaborative, open, and transparent environment, genomic scientists, the public, and policy makers can reasonably hope to create effective genomics policy in the future.

Good policy decisions will be crucial to reaping the benefits that should flow from the coming revelations about the genome by addressing important issues with prudence. These will include—

- Protection of individual and family privacy;
- Effective education of health-care providers and the public

Informed dialogue leads to better understanding, consensus building and a more stable environment for commerce.

Public policy decisions will result in better outcomes if they are based on both public values and technical knowledge.

Common citizens must become more involved in debating matters of great civic concern—as well as engaged in developing policies by which they are affected and governed.

Practicing genomics in the real world means thinking about the outcomes from the start, so that science and society can pull together to optimize the benefits of this new knowledge to human welfare and opportunity.

about genomic medicine; and—

- Appropriate health-care system reimbursement for the cost of validated preventive measures.

Genomics has had an exceptionally powerful facilitating role in biomedical advances over the past decade. Only time will tell how deep and how far that power potential will take us. With fair and equitable public policy decisions in place to help guide the translation of genomics discovery into the practice of genomic medicine and public health, the best is yet to come.

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