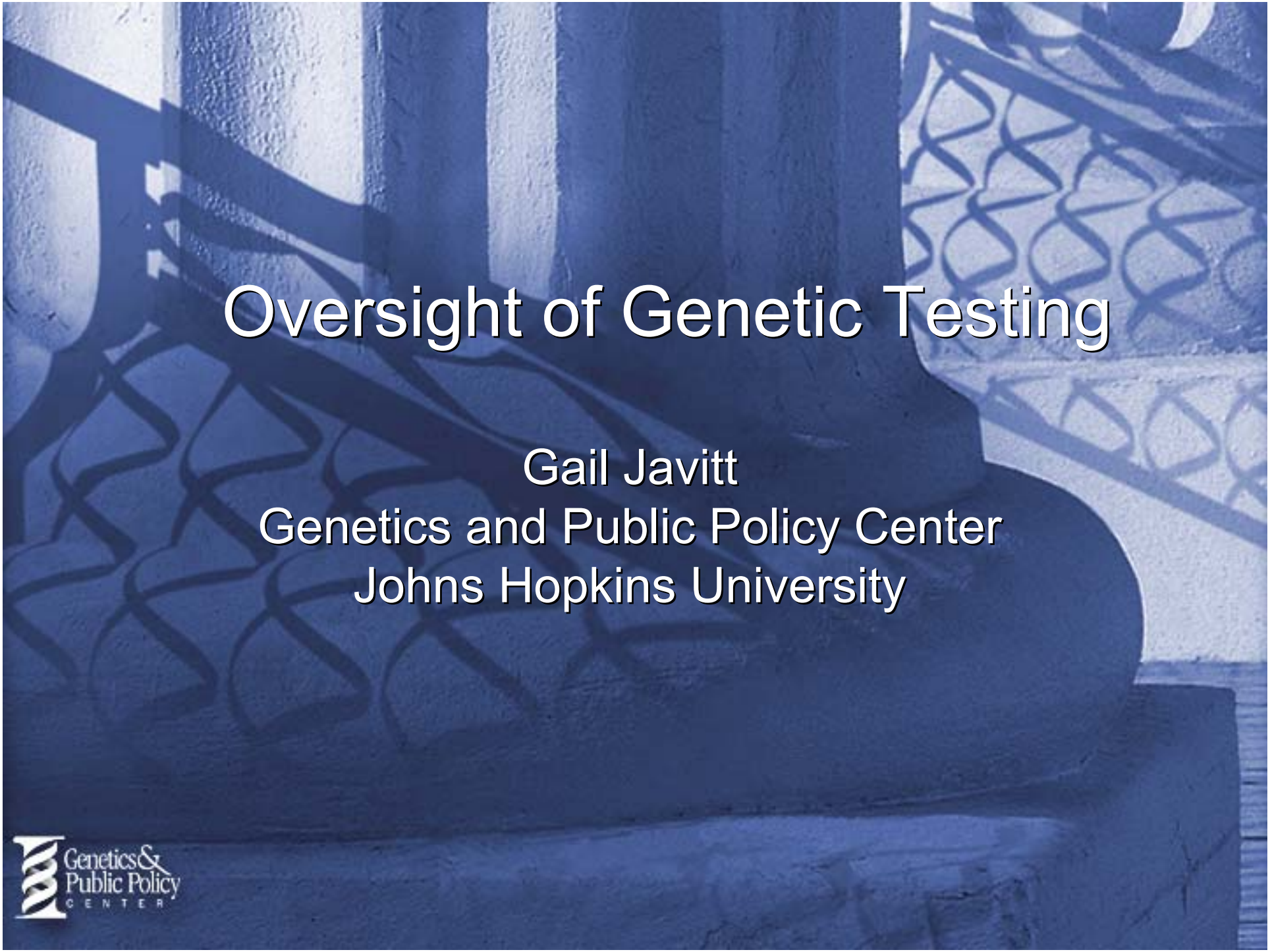

Oversight of Genetic Testing

Gail Javitt
Genetics and Public Policy Center
Johns Hopkins University

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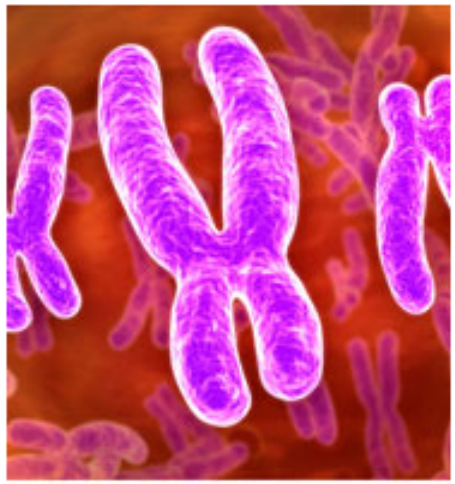
NEWS

XML

05/28/08 **Statement of Director Kathy Hudson regarding announced resignation of NHGRI's Francis Collins**
It is no coincidence that Francis Collins's tenure at the National Human Genome Research Institute marked one of the most extraordinary periods in modern science. full story

05/21/08 **President Bush signs long-awaited Genetic Information Nondiscrimination Act**
Signed into law today, GINA provides vital protection for Americans against the misuse of genetic test results by heath insurers and employers. full story

05/15/08 **Center to hold personal genomics seminar June 10**
The seminar, "The Molecular Full Monty: Personal Genomes, Personal Health," will take place at the National Press Club at full story



Internet

start | Microsoft O... | https://mail.... | - Genetics &... | Session 5 | Microsoft Po... | 9:12 AM

Mission

- The Center works to help policy leaders, decision makers, and the public better understand the rapidly evolving field of human genetics and its application to healthcare. . . . To inform genetic policy decisions, the Center surveys public attitudes about genetics issues, conducts analyses of the existing regulatory landscape, monitors the transition of genetic applications into clinical practice, and posits options and likely outcomes of key genetics policies.

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Goals of Regulation

- Protect the public
- Foster innovation
- Create uniform rules to prevent market inequalities (“level playing field”)

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Mechanisms of Regulation

- Many “tools” in the toolbox
- The right “tool” depends on the problem one is trying to fix or prevent, or the good one is trying to maintain or achieve

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Tools of Regulation

- Statute
- Regulation
- Incentives/Penalties (e.g., taxes, subsidies)
- Tort law
- Private mechanisms (e.g., professional societies)

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Goals of Genetic Testing Oversight

1. High quality laboratory testing
2. Clinically valid tests
3. Truthful, non-misleading claims about test benefits and limitations
4. Continued development of new tests
5. Appropriate and timely translation into clinical practice

Public confidence

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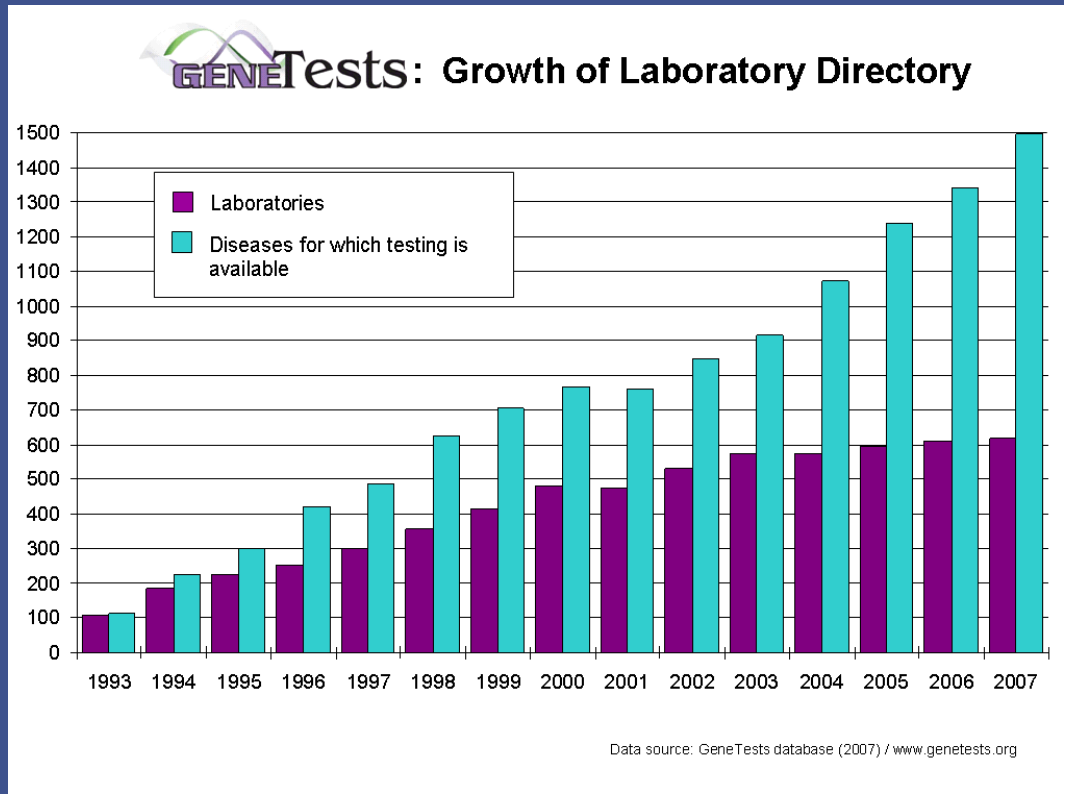
Challenges to Oversight of Genetic Tests

- Rapidly moving technology
  - tests for more than 1500 diseases



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Challenges to Oversight of Genetic Tests

- Old laws, new situations
  - need for continued updating of regulations to respond to new technologies
  - the laws may not “fit” the new context neatly

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Challenges to Oversight of Genetic Tests

- Prospective v. reactive
- Many government actors potentially involved

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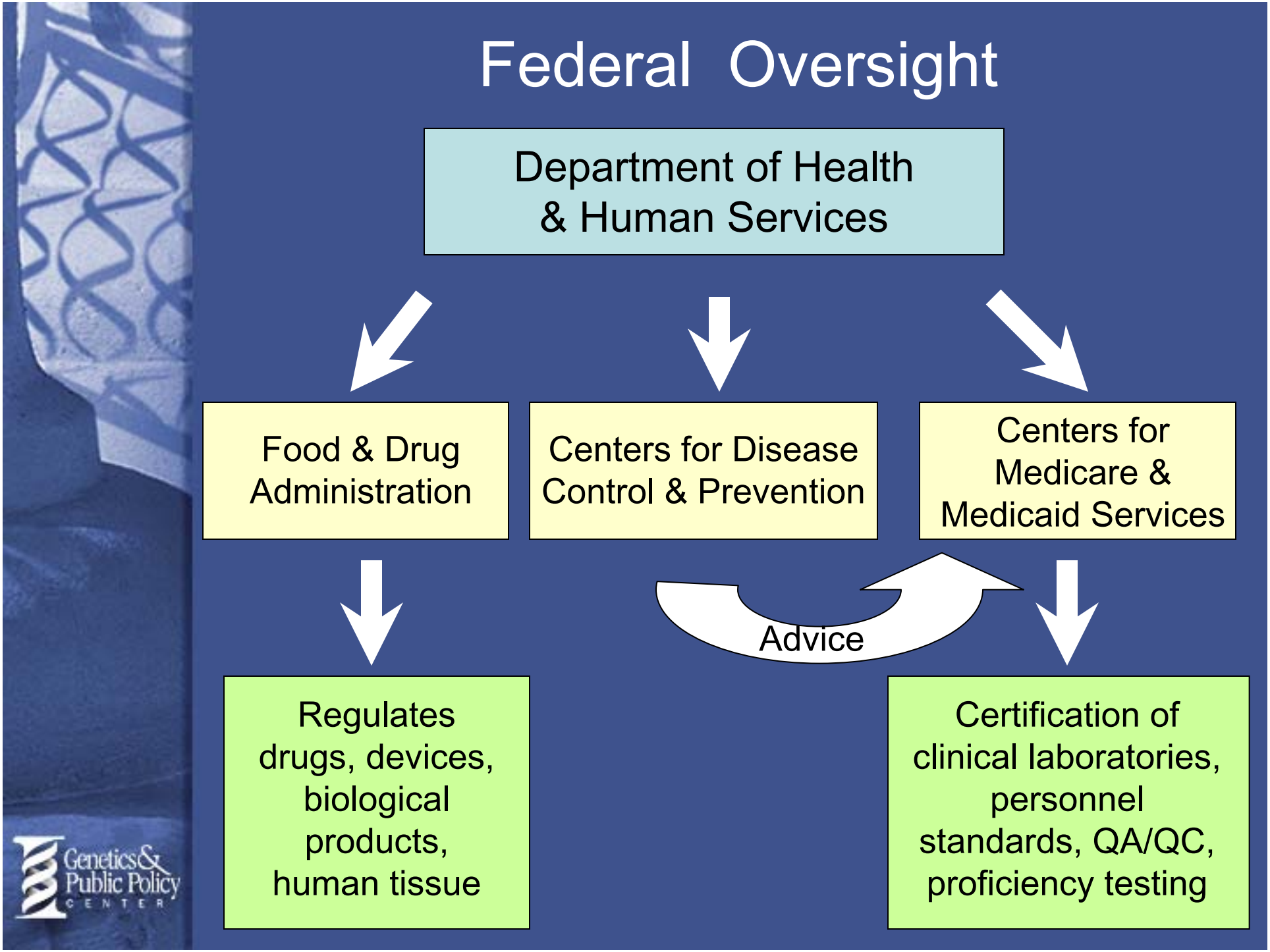
Federal Oversight



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CLIA

Definition of a Clinical Laboratory:

A laboratory that examines materials "derived from the human body" in order to provide "information for the diagnosis, prevention, or treatment of any disease or impairment of, or the assessment of the health of, human beings."

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Components of Laboratory Oversight

- Certification
- Inspection
- General requirements relating to QC, personnel, documentation, etc.
- More specific and tailored requirements for laboratories performing “high complexity” tests are set forth through the creation of “specialty areas”

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CLIA and Genetic Tests

- Law passed in 1988, regulations implemented in 1992
- Not specific to genetic tests
- Need to ensure that laboratory requirements appropriately include genetic testing laboratories
  - Proficiency testing
- Need to ensure that public has necessary information about laboratory quality

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FD&C Act

- Gives FDA authority to approve pharmaceuticals and medical devices before they are marketed

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FDA and Genetic Tests

- Regulatory status of genetic tests depends on how the laboratory develops and performs the test
  - Test kit/test system = regulation as medical device
  - Laboratory-developed = no FDA regulation
- Most genetic tests are laboratory developed
- Most genetic tests not required to demonstrate clinical validation

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IVDMIA Draft Guidance (July 2007)

- Defines new category of laboratory test -- “in vitro diagnostic multivariate index assays”
- Hallmarks of IVDMIAs
  - measure multiple analytes simultaneously (e.g., multiple gene or gene expression products)
  - use proprietary algorithm to calculate patient-specific result
  - provide result that cannot be independently interpreted by physician
- Require premarket clearance/premarket review by FDA
- Must demonstrate analytic and clinical performance of test

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Challenges to Oversight of Genetic Tests

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Meanwhile, on the Internet…

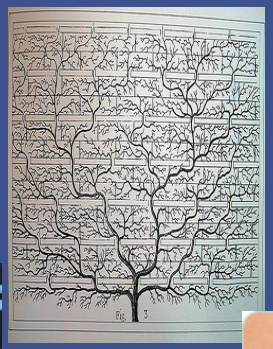
Fetal Gender

Inherited

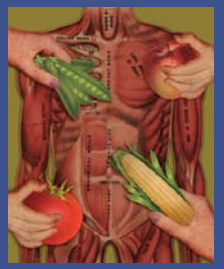
Pharma

Complex Disorders

Paternity



Ancestry



Nutrition

Athletic perf.

Complex Conditions

Infidelity

Skin care

Recreational

Infertility

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Companies offering health-related tests DTC

2/27/2008

Genetic Testing Company	Personal Genome Service	Addiction	Alzheimer	Arthritis	Ashkenazi Jewish Screening	Asthma	Athletic Performance	Bipolar / Depression	Cancer	Cardiovascular Disease	Celiac Disease	Cystic Fibrosis	Drug Response	Fanconi Anemia	Fetal Gender	Fragile X Syndrome	Gastrointestinal Disease	Gaucher Disease	General Nutrition	Glaucoma / AMD	Glycogen Storage Diseases	Hair Loss	Hemochromatosis	HIV Progression	Infertility	Metabolic Health	Multipe Sclerosis	Narcolepsy	Osteoporosis	Parkinson's	Periodontal Disease	Recurrent Pregnancy Loss	Skin Profile	Spinal Muscular Atrophy	Tay-Sachs Disease	Thrombosis	Type 1 / 2 Diabetes, Obesity
23andMe	X		X						X	X							X			X							X		X							X	X
Acu-gen Biolabs															X																						
Consumer Genetics													X		X																						
CyGene Direct							X													X					X			X							X	
DeCODE	X			X		X			X	X	X									X						X									X	X
Dermagenetics																																X					
DNA Direct	X				X				X			X	X					X					X		X						X			X	X	X
DNA Traits					X							X		X		X		X			X												X	X	X	
G-nostics		X																																		
GATC Biotech	X																																			
Genelex										X	X		X						X				X				X	X		X						
Genetwist	X																																			
Graceful Earth			X																																	
Hair DX																						X														
Health Check USA											X												X												X	
Health Tests Direct											X	X																							X	
Holistic Heal																			X																	
Knome	X																																			
MediChecks			X						X																										X	
Medigenomix																							X						X						X	
My Genome			X							X			X																X			X			X	
Navigenics	X																																			
Proactive Genomics									X																											
Psynomics								X																												
Quixtar - Interleukin Genetics										X									X																	
Salugen - DNA Services of America		X																																		
Scientific Match																															X					
Sciona										X															X			X								
SeqWright	X																																			
Signpost										X																										X
Smart Genetics			X																					X												
Suracell										X																										

July 27, 2006:

Senate Hearing, Special Committee on Aging, “At Home DNA Tests: Marketing Scam or Medical Breakthrough”

GAO
Accountability • Integrity • Reliability

Highlights

Highlights of GAO-06-977T, testimony before the Special Committee on Aging, U.S. Senate

July 27, 2006

NUTRIGENETIC TESTING

Tests Purchased from Four Web Sites Mislead Consumers

Why GAO Did This Study

Scientists increasingly believe that most, if not all, diseases have a genetic component. Consequently, genetic testing is becoming an integral part of health care with great potential for future test development and use. Some genetic tests are sold directly to the consumer via the Internet or retail stores, and purport to use genetic

What GAO Found

The results from all the tests GAO purchased mislead consumers by making predictions that are medically unproven and so ambiguous that they do not provide meaningful information to consumers. Although there are numerous disclaimers indicating that the tests are not intended to diagnose disease, all 14 results predict that the fictitious consumers are at risk for developing a range of conditions, as shown in the figure below. However, although some types of diseases, such as cystic fibrosis, can be definitively diagnosed by looking at certain genes, the experts GAO spoke with said that the medical predictions in the tests results can not be medically proven at this time.

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"The results from all the tests GAO purchased mislead consumers by making predictions that are medically unproven and so ambiguous that they do not provide meaningful information to consumers."

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Why GAO Did This Study

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GAO was asked to investigate the "legitimacy" of these claims. This testimony reflects the findings of GAO's investigation of a nonrepresentative selection of genetic tests. Specifically, GAO purchased tests from four Web sites and created "fictitious consumers" by submitting for analysis 12 DNA samples from a female and 2 samples from an unrelated male, and describing this DNA as coming from adults of various ages, weights, and lifestyle descriptions. GAO also consulted with experts in genetics and nutrition.

www.gao.gov/cgi-bin/getrpt?GAO-06-977T.

To view the full product, including the scope and methodology, click on the link above. For more information, contact Greg Kutz at 202-512-7455 or kutzg@gao.gov.

July 27, 2006

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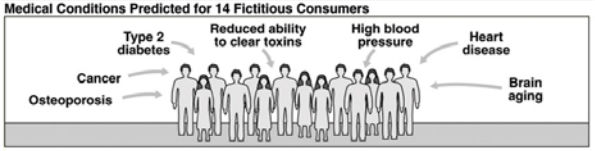
Medical Conditions Predicted for 14 Fictitious Consumers



Source: GAO.

Even if the predictions could be medically proven, the way the results are presented renders them meaningless. For example, many people "may" be "at increased risk" for developing heart disease, so such an ambiguous statement could relate to any human that submitted DNA.

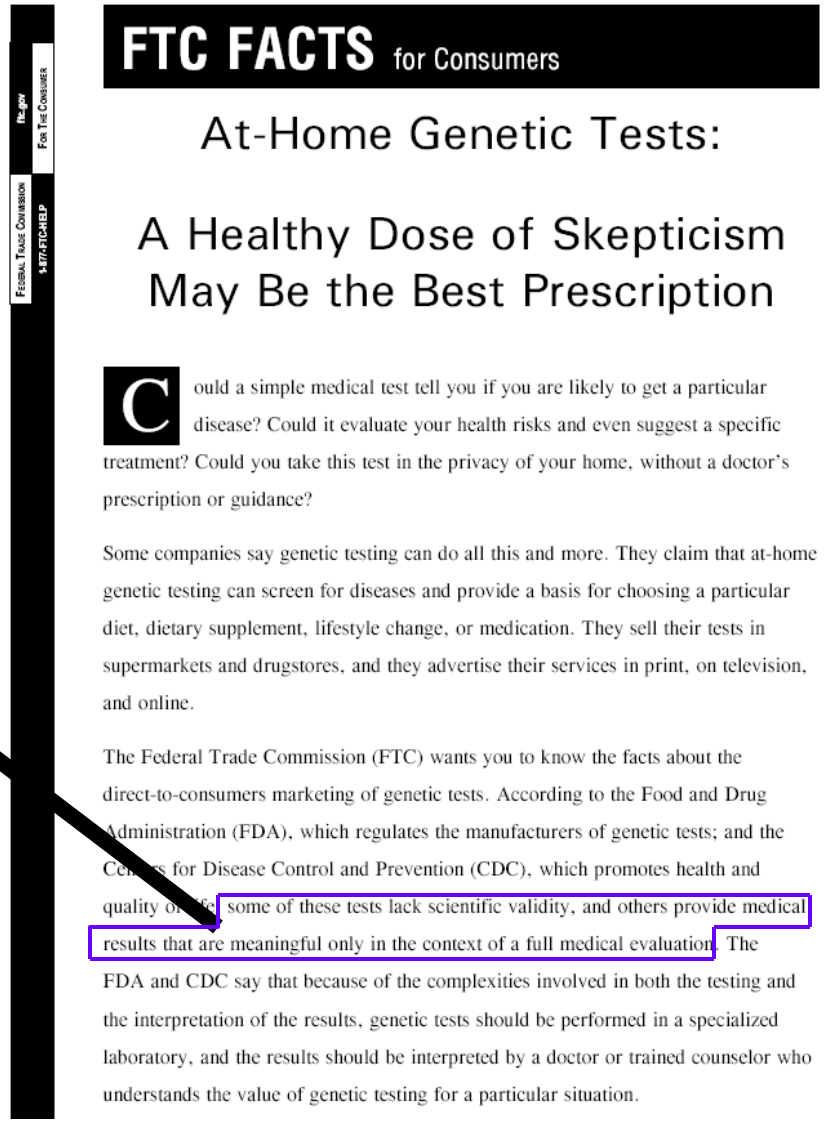
Results from the tests that GAO purchased from Web sites 1 and 4 further mislead the consumer by recommending costly dietary supplements. The results from the tests from Web site 1 suggested "personalized" supplements costing approximately $1, 200 per year. However, after examining the list of ingredients, GAO found that they were substantially the same as typical vitamins and antioxidants that can be found in any grocery store for about $35 per year. Results from the tests from Web site 4 suggested expensive products that claimed to repair damaged DNA. However, the experts GAO spoke with stated that there is no "pill" currently available that has been proven to do so. The experts also told us that, in some circumstances, taking supplements such as those recommended may be harmful.

In addition, results from the tests that GAO purchased from Web sites 1, 2, and 3 do not provide recommendations based on a unique genetic profile as promised, but instead provide a number of common sense health recommendations. If the recommendations were truly based on genetic analysis, then the 9 fictitious consumers that GAO created for these sites using the female DNA should have received the same recommendations because their DNA came from the same source. Instead, they received a variety of different recommendations, depending on their fictitious lifestyles. For example, when GAO created lifestyle descriptions stating that the consumers smoked, they received recommendations to stop smoking. In contrast, if GAO said the consumers never smoked, they received recommendations to continue to avoid smoking.

United States Government Accountability Office

Federal Trade Commission releases consumer advisory, "At Home Genetic Tests: A Healthy Dose of Skepticism May Be the best Prescription" (July 2006)

"…some of these tests lack scientific validity, and others provide medical results that are meaningful only in the context of a full medical evaluation."



Federal Trade Commission
1-877-FTC-HELP
ftc.gov
For The Consumer

FTC FACTS for Consumers

At-Home Genetic Tests:

A Healthy Dose of Skepticism May Be the Best Prescription

Could a simple medical test tell you if you are likely to get a particular disease? Could it evaluate your health risks and even suggest a specific treatment? Could you take this test in the privacy of your home, without a doctor's prescription or guidance?

Some companies say genetic testing can do all this and more. They claim that at-home genetic testing can screen for diseases and provide a basis for choosing a particular diet, dietary supplement, lifestyle change, or medication. They sell their tests in supermarkets and drugstores, and they advertise their services in print, on television, and online.

The Federal Trade Commission (FTC) wants you to know the facts about the direct-to-consumers marketing of genetic tests. According to the Food and Drug Administration (FDA), which regulates the manufacturers of genetic tests; and the Centers for Disease Control and Prevention (CDC), which promotes health and quality of life, some of these tests lack scientific validity, and others provide medical results that are meaningful only in the context of a full medical evaluation. The FDA and CDC say that because of the complexities involved in both the testing and the interpretation of the results, genetic tests should be performed in a specialized laboratory, and the results should be interpreted by a doctor or trained counselor who understands the value of genetic testing for a particular situation.

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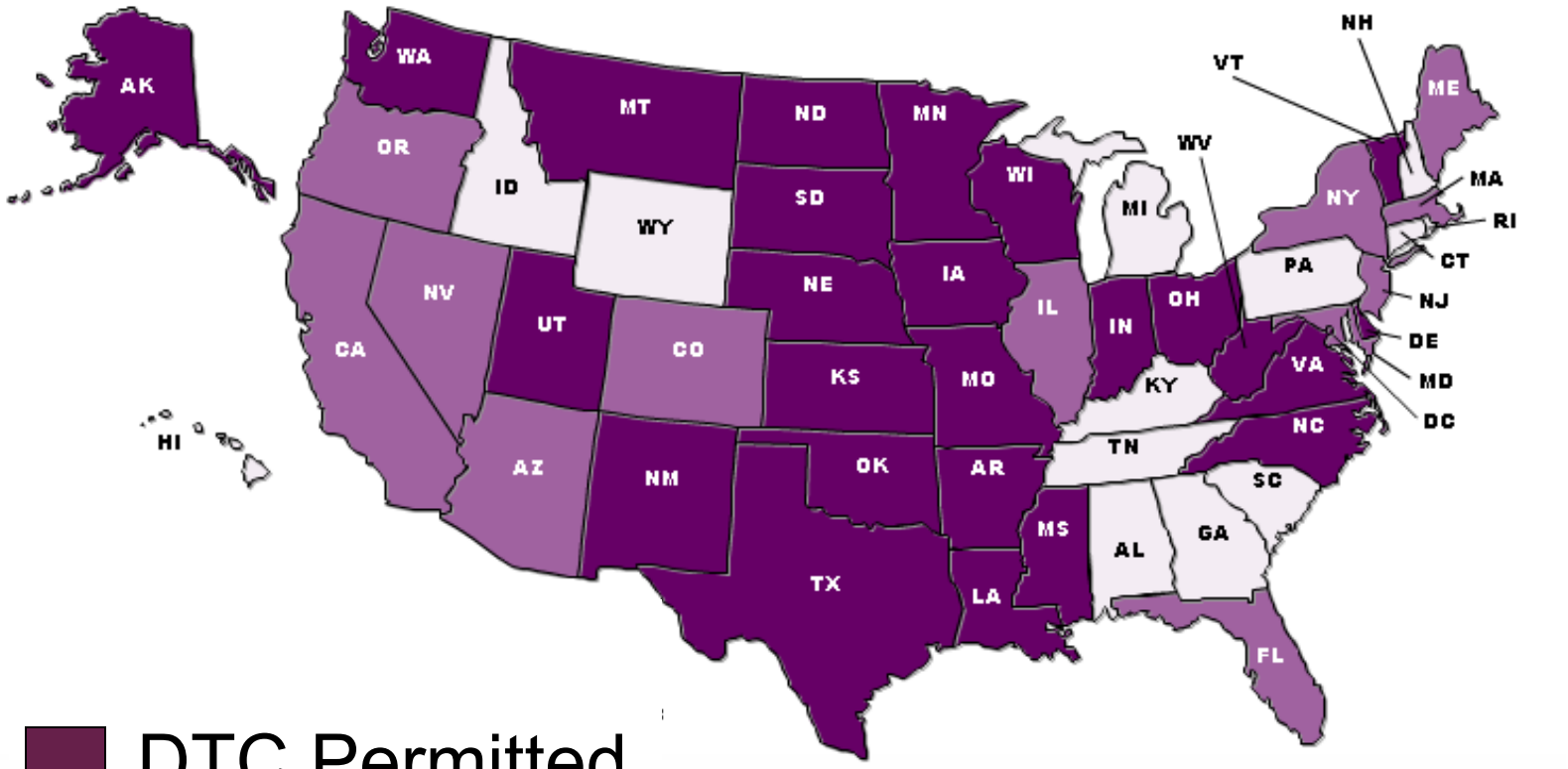
State DTC Testing Statutes and Regulations



DTC Permitted

Limited

DTC Not Permitted

Source: Genetics and Public Policy Center, http://www.dnapolicy.org/resources/DTCStateLawChart.pdf

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ACMG Statement on DTC

April 7. 2008

- A knowledgeable professional should be involved in the process of ordering and interpreting a genetic test.
- The consumer should be fully informed regarding what the test can and cannot say about his or her health.
- The scientific evidence on which a test is based should be clearly stated.
- The clinical laboratory must be accredited by CLIA, the state and/or other applicable accrediting agencies.
- Privacy concerns must be addressed.

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ASHG Statement on DTC

1. DTC companies should provide accurate information
2. Government actors should provide adequate oversight
3. Not all tests the same – DTC may be appropriate for some but not others

**K. Hudson, G. Javitt, W. Burke, P. Byers, with the ASHG Social Issues Committee, Am. J.. Hum. Genetics, Sept. 2007*

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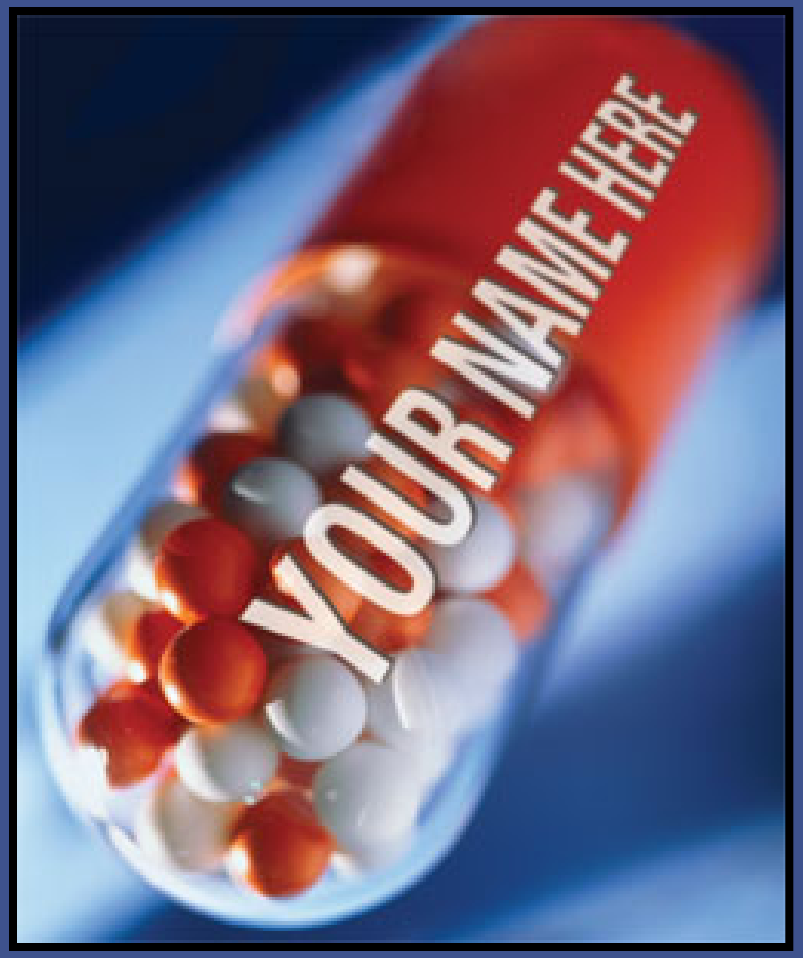
Pharmacogenetics: The Promise



↑Safety

↑Effectiveness

↓Cost

= Better patient care

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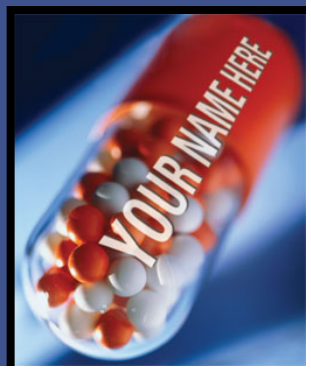
Pharmacogenetics: The reality

A few examples:

- Her2/neu and Herceptin
  - Only “co-developed” drug and test
- UGT1A1 and Irinotecan
- EGFR and Iressa
- HLA B and Abacavir
- CYP450 and Warfarin?????



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Prerequisites for Pgx Success

1. Robust and responsive research enterprise
2. Regulatory system that encourages development of safe and effective tests
3. Mechanism for evidence development and translation to clinical practice
4. Fair reimbursement
5. Safeguards for genetic information

Public confidence

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- Policy development is still evolving
- Legislation pending
- Recent recommendations by the SACGHS provide direction for future government activities

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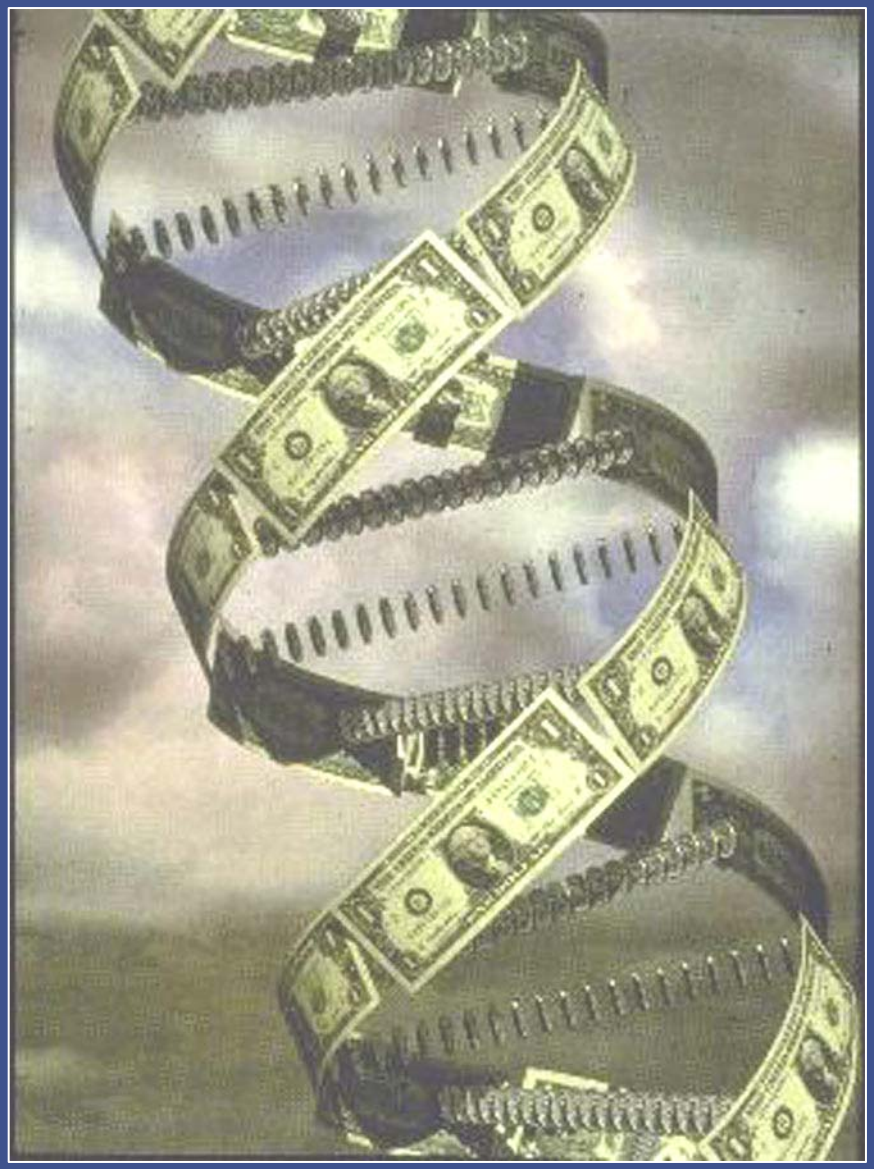
The Pew Charitable Trusts



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