



# Genetics Policy: Progress or Paralysis

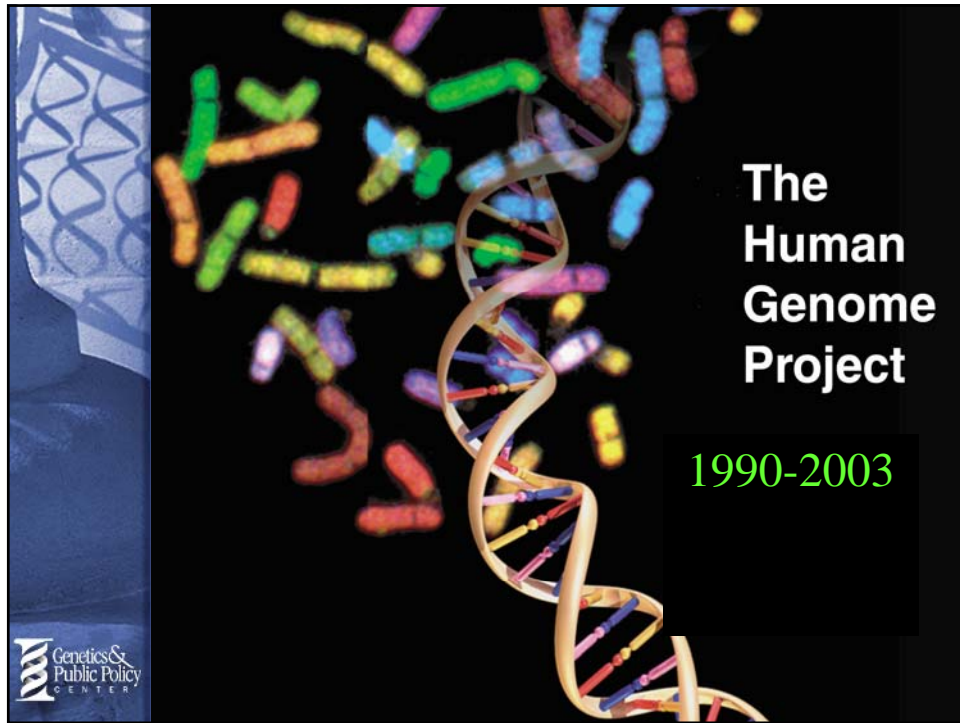
Kathy Hudson, Ph.D.  
Genetics and Public Policy Center  
February 17, 2006

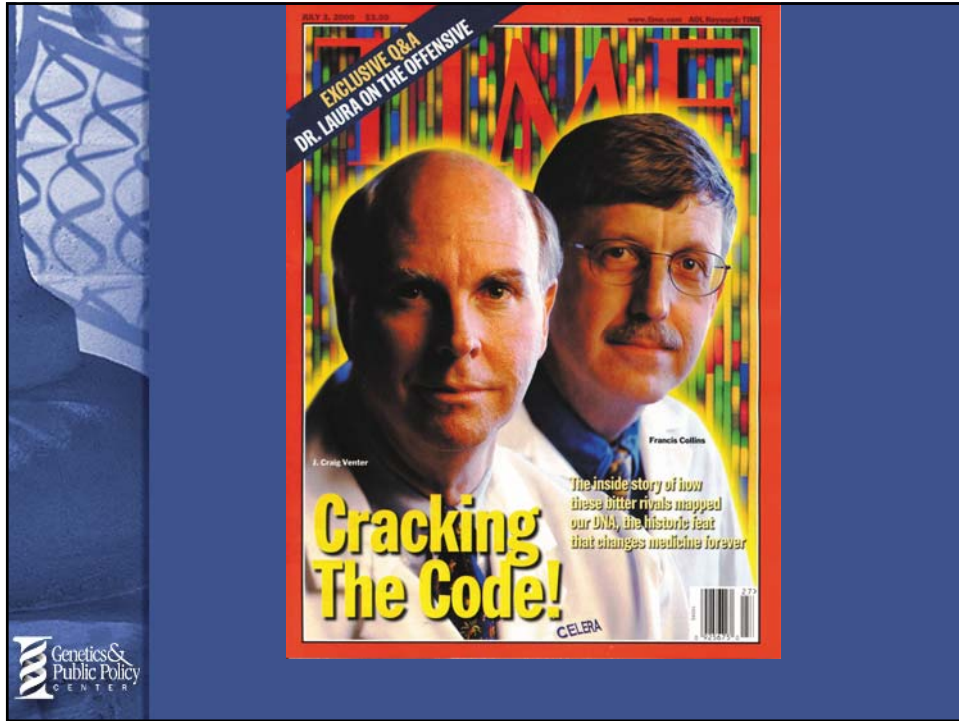


## Our Mission:

To create the environment and tools needed by key decision makers to carefully consider and respond to challenges and opportunities raised by scientific advances in human genetics.







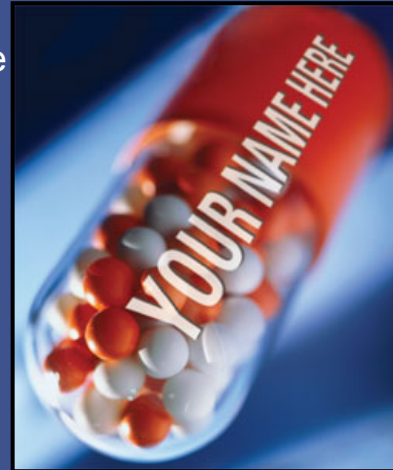
## Benefits of Genetic Testing

- Disease diagnosis
- Early intervention
- Prevention
- Knowledge



# This Drugs For You!!

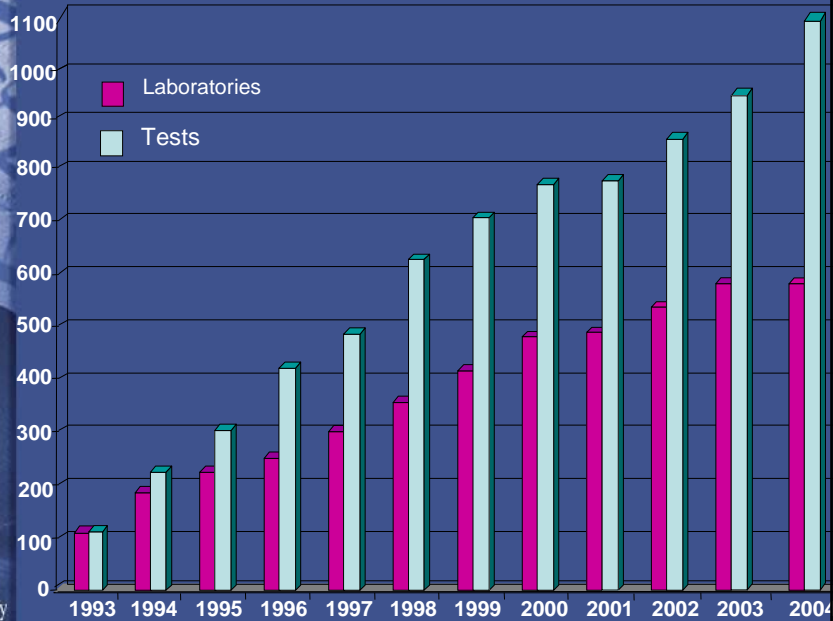
- Personalized Medicine
- Pharmacogenetics
- Pharmacogenomics



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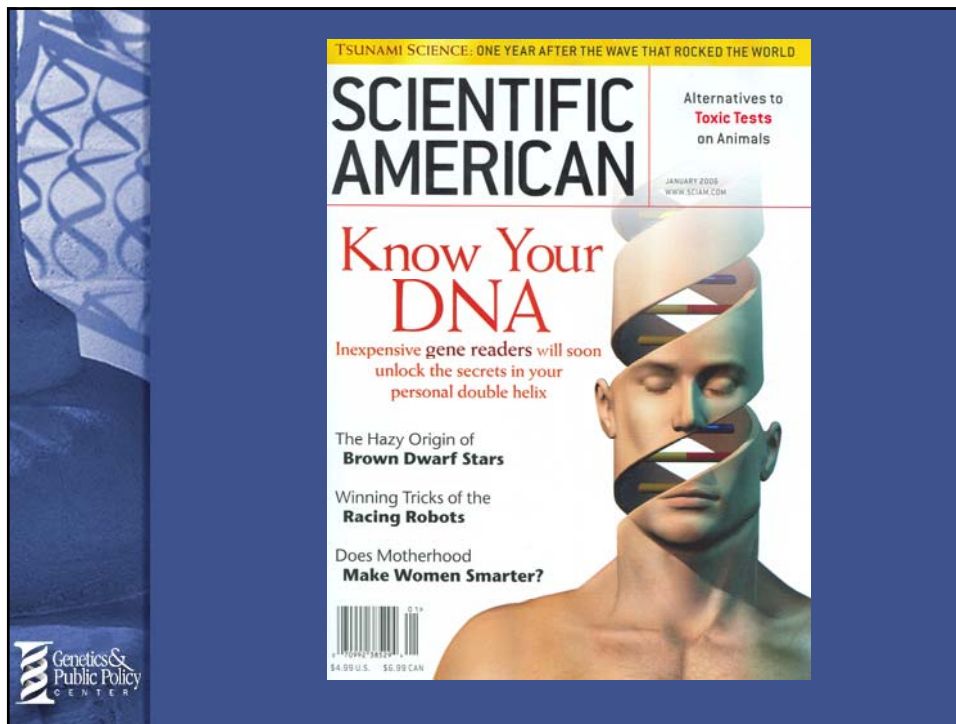
GENETests

## Growth of Genetic Testing



Data source: GeneTests database (2004) / [www.genetests.org](http://www.genetests.org)

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## Cracks in Genetic Testing Landscape

Safety & Accuracy  
of Genetic Tests

Quality  
Laboratories

Knowledgeable  
Health Care Providers



## Genetic Testing Quality Initiative



- *laboratories* must have quality control and personnel standards in place
- *tests* must be valid and reliable
- *health care providers* must understand when to order the tests and how to interpret results
- *uses and outcomes* must be evaluated over time.



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Are you compulsive? Have you ever wondered why you crave certain things and/or act in an irrational manner? Would you like to know if you have the genetic predisposition to abuse drugs and alcohol? Are you concerned about your children's future? Does your child have the genetic trait that leads to disruptive and addictive personalities? DNA testing can help you understand and manage a child's behavior before it gets out of control.

Imagene will test a panel of dopaminergic related Reward Deficiency Syndrome(RDS) genes. This will allow you to know if there is a genetic predisposition towards any of the associated addictions. The Reward product line is then available to treat the genetic predisposition towards RDS.

Imagene is an at home genetic testing kit that is simple to use. Here are the actual

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**Baby Gender Mentor™ Home DNA Gender Testing Kit**

Boy or Girl? Now You Can Know Even Before You Show

As featured on ABC's World News Tonight, CNN, NBC's Today show – watch the TODAY show segment now, and other major news and media outlets! Offered Exclusively Online From PregnancyStore.com! Are you pregnant and want to know if you're having a boy or girl? [Not pregnant yet but know that you want to know the sex of your baby when you are pregnant?](#)

It's estimated that about 50-70% of parents-to-be want to know if they're having a baby boy or girl. Knowing the gender of your unborn baby helps prepare for your baby's birth: you can decorate the nursery, choose your baby's name, and make gender-specific purchases during your pregnancy.

**"We are excited to tell you we are having a baby boy!! My wife and I could not be more happy! Thanks to you and all the great people on your team for rushing our order through so quickly... the suspense was killing us!"** Robert & Ann T.

If you want to know the gender of your unborn baby, forget old wives tales like the Chinese lunar calendar chart, the Wedding Ring Spin, needle on a string test, or how you're carrying your baby. **Now there's a scientific, non-invasive way to predict your baby's gender – with 99.9% accuracy. Are you having twins?** Now you can find out what you're having as early as five weeks after conception!

With a few drops of your maternal blood you can find out your baby's sex AS EARLY AS FIVE WEEKS after conception with the NEW Baby Gender Mentor Home DNA Gender Testing Kit.

**"Wow! That's all I can say about this test. It came very quickly in the mail. Was easy to understand and perform. I had my results within less than 48 hours. And my husband and I were tickled to learn we will be having our first baby girl!! Our two sons are delighted about their baby sister! I wouldn't hesitate to tell women to take this test. Thank you so much for making this kit available to pregnant women everywhere!"** – Paul & Kimberly H.

**How Does the Baby Gender Mentor Home DNA Gender Testing Kit Work?**

This technology has been in trial for 14 years – and works! Baby Gender Mentor Home DNA Gender Test employs cutting-edge, patent-pending technology to attain the **earliest gender detection with unprecedented sensitivity and unsurpassed accuracy.** The technique traces the amount of Y

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**DNAdirect** Your Genes in Context info for healthcare professionals

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Fertility Issues Questionnaire

○ Questionnaire  
● **Results**  
○ Begin Order

**Need more information?**  
Use our decision tools and step-by-step information to help you decide whether to test. (PDF 184K)

**Fertility Issues**

You answered that you and your partner have been trying to get pregnant for several months and that you have had a positive pregnancy test at some time in your reproductive history. If you have experienced 2 or more pregnancy losses (miscarriages, fetal death or stillbirth) the following tests are appropriate for you to consider at this time: **chromosome analysis and factor V testing.**

**Chromosome analysis** looks at the number and structure of your chromosomes. People who have extra, missing, or rearranged chromosomes (or parts of chromosomes) may experience reproductive difficulties.

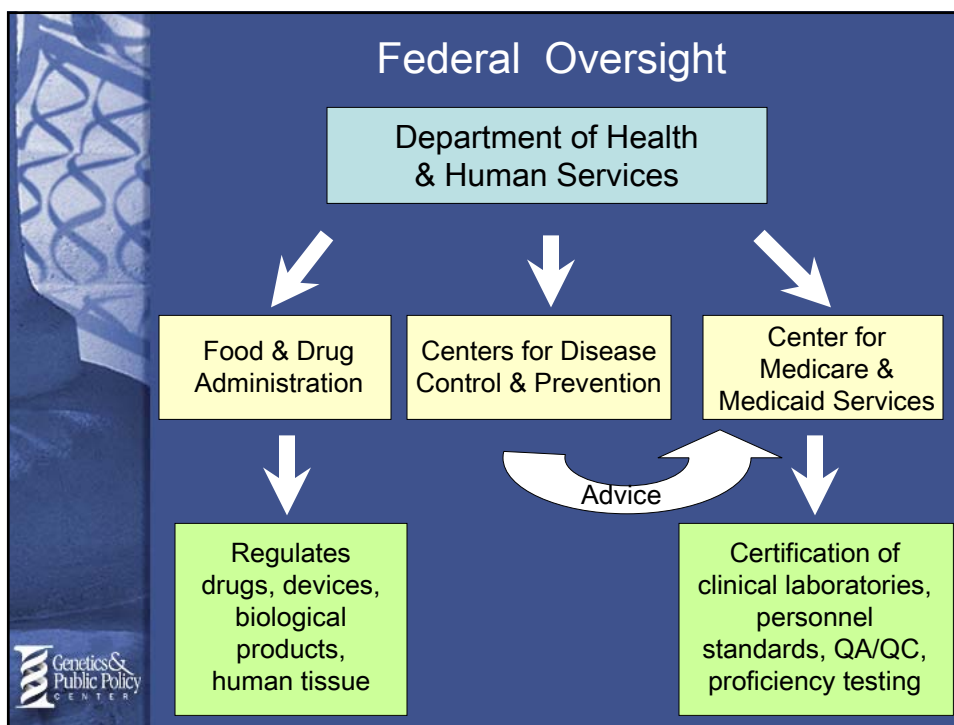
**Factor V testing** identifies a common genetic variation that increases the likelihood of blood clots. Women with recurrent miscarriage may experience pregnancy loss due to blood clot formation in the developing placenta.

**Call for a Free, Personalized Consultation:** Our board-certified genetic experts can discuss your medical and family history and help you make an informed choice

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## Types of Genetic Tests

Home brew



Test kit



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## FDA Regulation of Genetic Tests

- Many members of the public believe government is already regulating the safety and accuracy of genetic tests
- A large majority of the public (>90%) believe that the government should ensure the safety and accuracy of genetic tests
- FDA has reviewed and approved a handful of genetic tests

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## CMS Regulation of Genetic Tests

- CLIA applies to labs doing medical testing, including genetic tests.
- No genetic testing-specific proficiency testing.
- Voluntary proficiency testing for only 17 molecular genetic tests.

## Types of Genetic Tests

Home brew  
home brew

**“Kitchen”  
Inspection  
(CLIA)**

**Chef with  
culinary degree**

Test kit

**“Kitchen”  
Inspection  
(CLIA)**

**Chef with  
culinary degree**

**Cake Mix FDA  
Approved**



Home Brews                      Test Kits



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What is Not Currently Subject to Federal Oversight?

- No premarket approval of most genetic tests.
  - Individual laboratories decide what tests to offer and when.
  - True for genetic tests used in adults, children, prenatally, or in PGD.
- Only handful of tests that have been approved by the FDA
- CMS has not created specialty area for most genetic tests under CLIA, unlike other complex tests

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## Consequences of Inadequate Oversight

1. Unnecessary or harmful treatments undertaken
2. Missed opportunity for early and effective intervention
3. Bad tests=public mistrust  
=thwarted promise of genetics



## Timeline of Inaction

- 1997 NIH/DOE Task Force Recommendations
- 2000 SACGT Recommendations
- 2000 CMS issues Notice of Intent to develop specialty area for molecular and biochemical genetic tests
- January 2001 Secretary Shalala indicates HHS intends to implement enhanced system of oversight for genetic tests
- September 2001 SACGT retracts proposed classification methodology for genetic tests





Why are we immobilized?

Was the 2000 proposal uniformly hated?

We FOIA'd the comments and took a look.

Surprisingly, there were strong areas of consensus about the need for a genetic testing specialty and consensus concern about labs being responsible for what happens in the doctor's office.





11/26/05 – Center sends report on our analysis to McClellan along with a letter asking that CMS expedite a proposed regulation.

1/12/06 – Center receives letter from CMS stating they intend to issue an NPRM “in the coming months.”

2/28/06 – Genetic Alliance sends letter to CMS requesting issuance of NPRM.

4/24/06 – CMS publishes Semiannual Regulatory Agenda stating intention to publish NPRM in 11/06.








What is going on in labs?

What is rate and source of errors?

What do lab directors think about current and proposed oversight?


Survey of genetic testing laboratory directors



Enhanced oversight of genetic testing laboratories:

Necessary but not sufficient.


- Premarket review
- Postmarket surveillance




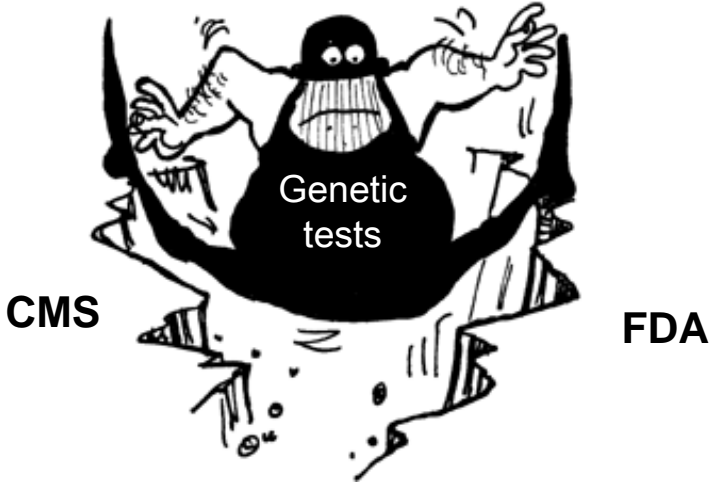


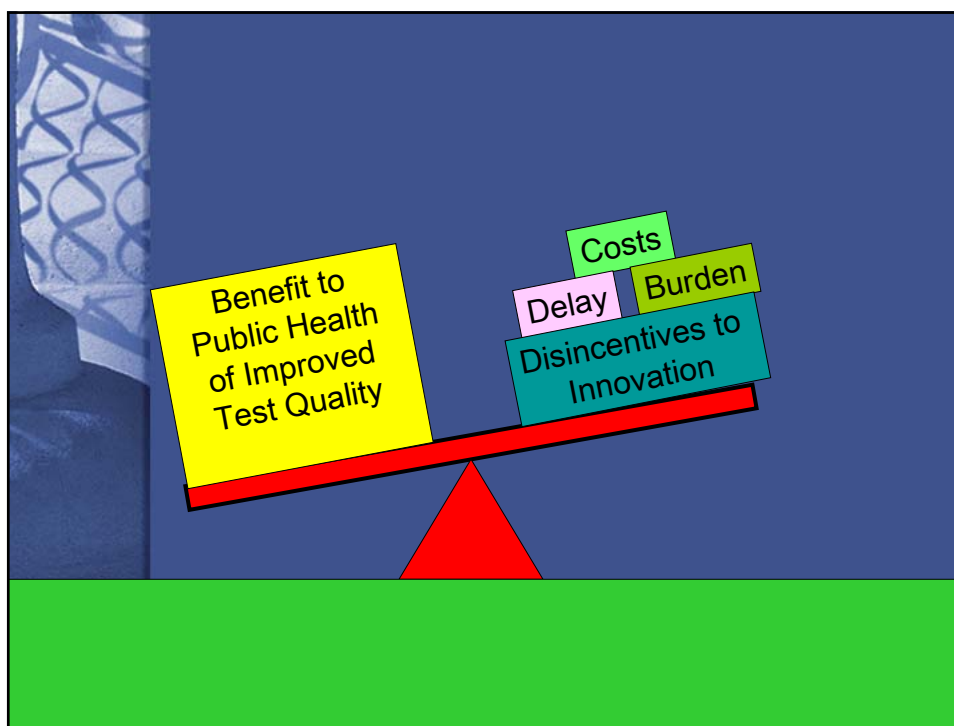
### Timeline of FDA Action on Pharmacogenetics

- November 2003 Guidance for Industry: Pharmacogenomics Data Submissions (Draft)
- March 2005 Guidance for Industry: Pharmacogenomics Data Submissions (Final)
- April 2005 Drug-Diagnostic Co-development Concept Paper
- February 2006 Draft Guidance for Industry and FDA Staff: Pharmacogenetic Tests and Genetic Tests for Heritable Markers



### The quality of genetic tests is falling between regulatory gaps.

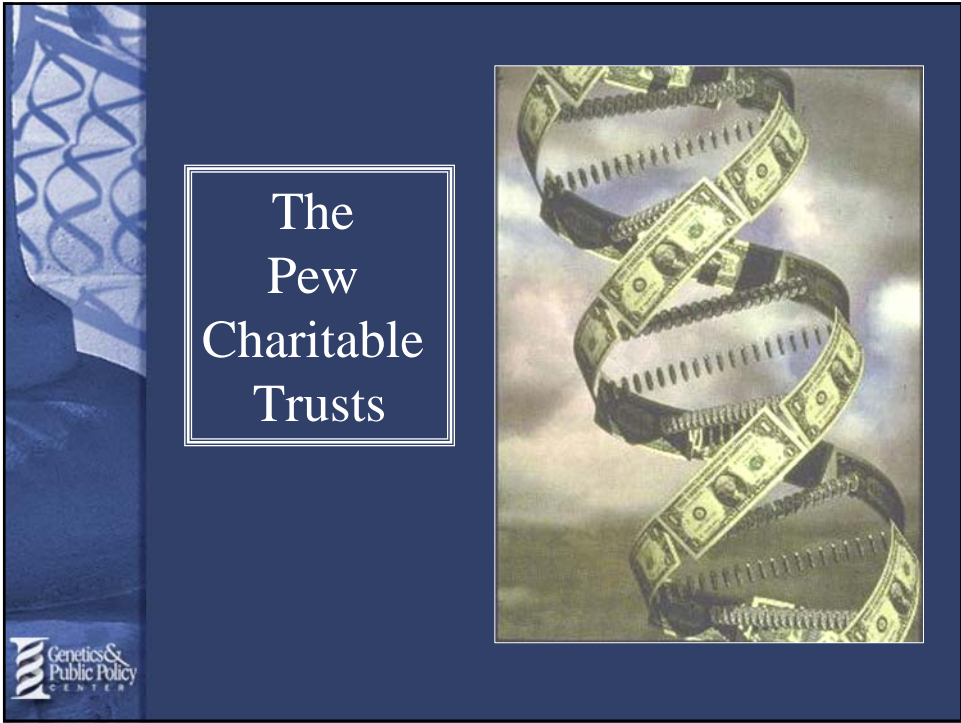





## What is Needed?

1. High quality laboratories-demonstrated ability to get the right answer reliably
2. Validity of tests supported by the science in advance of clinical offering
3. Educated and skilled health care providers
4. "Safety" of test results  
Non-Discrimination legislation





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