HC76A Winter 2004
Professor Bob Goldman

Lecture #11
Genetics & the Law

THEMES/CONCEPTS

1. Human Disease banks - Inborn Errors of Metabolism
2. "Home" DNA testing
3. How is Genetic Information Used? Issues?
4. Chinese Eugenics Law / Buck vs. Bell
5. Federal Laws
6. State Laws
7. Regulation of DNA Tests
8. Newborn, Child, & Adult Testing
9. Family Planning Issues
10. Privacy Issues
11. Employment Issues / Insurance Issues
12. DNA Testing & the Law / Data Banks
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<table>
<thead>
<tr>
<th>Genetic Defect</th>
<th>Locus</th>
<th>Enzyme Deficiency</th>
<th>OMIM Entry</th>
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<td>Acid phosphatase deficiency</td>
<td>3q21–q23</td>
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<td>Alkaptonuria</td>
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<td>Homogentisic acid oxidase</td>
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<td>Ataxia, intermittent</td>
<td>7q31.2</td>
<td>Pyruvate dehydrogenase</td>
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<td>Cystic fibrosis</td>
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<td>Cystic fibrosis transmembrane conductance regulator</td>
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<td>Cataract</td>
<td>9q34</td>
<td>Galactokinase</td>
<td>230200</td>
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<td>Disaccharide intolerance I</td>
<td>3q25–q26</td>
<td>Invertase</td>
<td>213500</td>
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<td>Fructose intolerance</td>
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<td>Gaucher disease</td>
<td>9p13</td>
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<td>Gaucher disease</td>
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<td>Glycogen branching enzyme</td>
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<td>Hemolytic anemia</td>
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<td>Glutathione peroxidase or glutathione reductase or</td>
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<td>Uridine monophosphate kinase</td>
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<td>Intestinal lactase deficiency (adult)</td>
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<td>Kidney tubular acidosis with deafness</td>
<td>2cen–q13</td>
<td>Carbonic anhydrase B</td>
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<td>Leigh's necrotizing encephalopathy</td>
<td>11q13.4–q13.5</td>
<td>Pyruvate carboxylase</td>
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<td>Lesch-Nyhan syndrome</td>
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<td>Hypoxanthine-guanine phosphoribosyltransferase</td>
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<td>Lysine intolerance</td>
<td>2cen–q13</td>
<td>Lysine: NAD-oxidoreductase</td>
<td>247900</td>
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<td>Male pseudohermaphroditism</td>
<td>19q13.1–q13.2</td>
<td>Testicular 17,20-desmolase</td>
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<td>Maple sugar urine disease, type IA</td>
<td>Xp21.2</td>
<td>Keto acid dehydroxylase</td>
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<td>Muscular dystrophy, Duchenne and Becker types</td>
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<td>Dystrophin absent or defective; serum acetylcholinesterase or acetylcholine transferase or creatine phosphokinase elevated</td>
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<td>Niemann-Pick disease</td>
<td>11p15.4–p15.1</td>
<td>Sphingomyelin hydrolase</td>
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<td>Orotidyl decarboxylase and orotidyl pyrophosphorylase</td>
<td>238900</td>
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<td>Phenylketonuria</td>
<td>12q24.1</td>
<td>Phenylalanine hydrolase</td>
<td>261600</td>
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<td>Porphyria, acute intermittent</td>
<td>11q23.3</td>
<td>Uroporphyrinogen III synthetase</td>
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<td>Porphyria, congenital erythropoietic</td>
<td>10q25.2–q26.3</td>
<td>Uroporphyrinogen III synthase</td>
<td>263700</td>
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<tr>
<td>Pulmonary emphysema</td>
<td>14q32.1</td>
<td>α-1-Antitrypsin</td>
<td>107400</td>
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<td>Pyridoxine dependency with seizures</td>
<td>2q31</td>
<td>Glutamic acid dehydroxylase</td>
<td>266100</td>
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<td>Rickets, vitamin D-dependent</td>
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<td>25-Hydroxycholecalcifer 1-hydroxylase</td>
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<td>Tay-Sachs disease</td>
<td>15q23–q24</td>
<td>N-acetylhexosaminidase A</td>
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<td>Thyroid hormone synthesis, defect in</td>
<td>2p25</td>
<td>Iodide peroxidase or deiodinase</td>
<td>274500</td>
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<td>Tyrosinemia, type III</td>
<td>12q24–qter</td>
<td>p-Hydroxyphenylpyruvate oxidase</td>
<td>276710</td>
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*Prenatal diagnosis possible.
### TABLE 18-7  Current Holdings of OMIM (November 2002)


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<thead>
<tr>
<th></th>
<th>Autosomal</th>
<th>X Linked</th>
<th>Y Linked</th>
<th>Mitochondrial</th>
<th>Total</th>
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<td>538</td>
<td>39</td>
<td>37</td>
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<td>phenotype loci (*)</td>
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<tr>
<td>Phenotype descriptions</td>
<td>1,065</td>
<td>90</td>
<td>0</td>
<td>23</td>
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<td>(#)</td>
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<tr>
<td>Other loci or</td>
<td>2,263</td>
<td>158</td>
<td>2</td>
<td>0</td>
<td>2,422</td>
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<td>phenotypes (no prefix)</td>
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<td></td>
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<td>Total</td>
<td>13,120</td>
<td>786</td>
<td>41</td>
<td>60</td>
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**Note:** See Table 18.5 note for definition of the asterisk (*) and number (#) symbols.

**FIGURE 18.6.** Online Mendelian Inheritance in Man (OMIM), accessible via the NCBI website ([http://www.ncbi.nlm.nih.gov/omim](http://www.ncbi.nlm.nih.gov/omim)), allows text searches by criteria such as author, gene identifier, or chromosome.
Figure 1 The functions of the protein products of disease genes. a, The entire disease gene set. b–f, Disease genes stratified according to the typical age of onset of the disease phenotype. The fraction of disease genes encoding transcription factors in the in utero onset disorders (25%) differs from the fraction encoding transcription factors for disorders with onset after birth (6%; $\chi^2 = 46.4, P < 0.001$). Similarly, the fraction of disease genes encoding enzymes causing a disorder with onset in the first year of life (47%) is different from the fraction encoding enzymes causing disorders with other ages of onset (25.8%; $\chi^2 = 35.8, P < 0.001$).

Figure 2 Characteristics of disease arranged by function of the protein encoded by the disease gene. a, Disease genes encoding enzymes; b, disease genes encoding modifiers of protein function; c, disease genes encoding receptors; d, disease genes encoding transcription factors. The columns of disease features are labelled at the top. AR, autosomal recessive; AD, autosomal dominant; early adulthood, puberty to <50 years; late adulthood, >50 years.
Metabolic Diseases - Inborn Errors of Metabolism

Proteins break down in metabolism

Phenylalanine

\[ \text{Phenylpyruvic acid} \]

1. The enzyme that converts phenylalanine to tyrosine is nonfunctional in phenylketonuria (PKU).
2. Because conversion to tyrosine is blocked, phenylalanine and phenylpyruvic acid accumulate in PKU.
3. This compound was detected in the urine test with ferric chloride.
4. Homogentisic acid accumulates in alkaptonuria and turns urine black.
5. The enzyme that catalyzes this conversion is deficient in alkaptonuria.

18.1 One Gene, One Enzyme in Humans
Phenylketonuria and alkaptonuria are both caused by abnormalities in a specific enzyme. Knowing the causes of such single-gene, single-enzyme metabolic diseases can aid in the development of screening tests and treatments.
**FIGURE 14.10** Metabolic pathway involving phenylalanine and tyrosine. Various metabolic blocks resulting from mutations lead to the disorders phenylketonuria, alkaptonuria, albinism, and tyrosinemia.

**Newborn Screens for Compounds that Accumulate Before Block in Metabolic Pathway**
DO-IT-YOURSELF DNA

If you’ve tried and tried but your family tree is still just a seedling, mail-order DNA testing may be for you. Comparing your genetic profile with those of other genealogy buffs—and potential relatives—can provide new leads. For $149 and up, Family Tree DNA will give you a list of 25 markers (or genetic traits) you carry, based on a swab from the inside of your cheek. For a bit more—$220 and up—Oxford Ancestors (oxfordancestors.com) will check 10 markers and tell you which “Seven Daughters of Eve” clan you belong to. If that’s too steep, the Molecular Genealogy Research Project will test 250 markers for free. Run by Brigham Young University, it hopes to create a worldwide database. The catch: the data must be kept anonymous. In other words, the project will create a map of ancestry lines—not an individual report for you.

—MATTHEW MACROBERTS
Screening Newborn Infants for Disease Genes

1. A "heel-stick" blood sample is taken a few days after birth.

2. The sample is dried on blotting paper.

3. The dried spot is cut out and placed on a plate with bacteria that need phenylalanine to grow well.

4. A positive test shows a halo of growing bacteria surrounding spots with excess phenylalanine. A negative test shows limited growth.

18.10 Genetic Screening of Newborns for Phenylketonuria
A simple test devised by Robert Guthrie in 1963 is used today to screen newborns for phenylketonuria. Early detection means that the symptoms of the condition can be prevented by following a therapeutic diet.
Do you want to know your future?

Every state in the country requires that infants be tested for a list of obscure diseases. Before long, some states could move on to DNA testing of all newborns. Now is the time to decide a critical question: How much do we want to know and when do we want to know it?

By Jeff Wheeless
Photography by Catherine Ledner

TESTING YOUR FUTURE
WHAT is Genetic Information Used For?

1. **Medical Uses**
   a. Carrier screening
   b. pre-natal diagnostic testing
   c. newborn screening
   d. presymptomatic testing for predicting adult-onset disorders such as Huntington’s disease
   e. presymptomatic testing for estimating risk of developing adult-onset cancer or Alzheimer’s disease
   f. Continuation of diagnosis of a symptomatic person
   g. pharmacogenetics/drug sensitivity
   h. preventive medicine - potential to develop heart disease, obesity, etc.
   i. population SNPs to associate group with high incidence of genetic disease with gene!

2. **Non-Medical Uses**
   a. Insurance (Life/Health)
   b. Employment (Workplace Hazard Susceptibility)
   c. paternity/kinship/Estate Settlement
   d. Forensics/Identification
   e. Criminal law (Innocence/Suit)
   f. Immigration (kinship)
   g. Schools (Identification) - children
Hypothetical Uses of Genetic Information

1. **Criminal Defendants**
   - Genetically predisposed to violent behavior as a defense to mitigate sentence!

2. **Personal Injury**
   - Compel victims to undergo genetic tests to estimate life expectancy before/at time of accident!

3. **Adoption/Child Custody**
   - Genetic testing prior to placing child for adoption?

4. **Mortgage Company**
   - Genetic test to assess applicant's life expectancy!!

5. **Medical Predisposition to Genetic Disease**
   - Force a relative's donation of DNA for family linkage & testing purposes!

Future - depends upon correlating complex traits (e.g., life expectancy, behaviors) with genes & sifting out environmental components. If tests to be predictive!
Genetic Testing Issues
A Clinically-Oriented List

1. Diagnostic Genetic Testing
2. Predictive/ Presymptomatic Genetic Testing
3. Genetic Testing Of Children -- Diagnostic Vs. Predictive
4. Late-Onset Disorder Pre-Symptomatic Testing
5. Genetic Testing And Privacy
6. Genetic Testing And Insurance
7. Over-The-Counter Testing
8. Population Screening For Carriers
9. Newborn Screening (e.g., Pku)
10. Pre-Natal Genetic Screening
12. Pre-Implantation Genetic Diagnosis (PGD)
11. Outcomes Of Genetic Counseling
Genetic Disease Testing Issues

1. Privacy?
   • Who Should Know -- Spouse, Children, Employer, Insurance
   • Company? Tell If Carrier? Who Should Tell? -- Person, Physician, Testing Lab?
   • Results – When Insurer’s Know/Employer’s Know?

2. Voluntary vs. Mandatory Testing?
   • What Is The Precedent? State Laws?
   • Results - Sickle-Cell, Cystic Fibrosis, Pku, BRCA1?
   • Test For Any Reason-- Information? Eugenics? Forensics?

3. Regulation?
   • Who Monitors Tests? Over-The-Counter?
   • Who Licenses Testing Labs And Ensures Quality/Accuracy?
   • Who Can Have Access To Tests? Costs?

4. Health Insurance?
   • Who Pays For Genetic Diseases? If Known Before Birth? Carriers?
   • How Assess Rates/Risks?
   • Will Society Pay For Genetically "Inferior" If Testing Available?
   • Community Rating? Universal Health Insurance
5. Penetrance/ Expressivity Problem?
   - What To Do If Disease Not Manifested 100%?
   - How To Deal With Information? Insurance Issues?

6. What Laws Govern Parental Rights To Test And Perhaps Use PGD Test To Enhance!
   - Who Protects Genetic Rights Of Zygote? Does It Have Any?
   - Wrongful Birth Suits

7. How Use Association Studies That Test For Complex Traits?
   - How Use Probabilities For "Getting" The Disease? Who Gets Access?
   - We All Carry Genes For Some Disease? Environmental Factors?

8. What Does History Tell Us?
   - Sickle-Cell Testing?
   - Cystic Fibrosis Testing?
   - Tay-Sachs Testing?
   - HMO's? Employers?

9. What Laws Protects Us From Genetic Discrimination?
   - Any? States? Federal?
   - American Disability Act
   - Carriers?
   - Executive Order 2000/ Clinton
   - Genetic Non-Discrimination Act of 2003

10. What About Testing For Enhancement/Eugenics?
    - Culturally Related?
INTRODUCTION

On 1 June 1995, the People’s Republic of China brought into force the first clearly eugenic law that the world has seen since modern genetics began to have impact on medical practice. Innocuously entitled the law on “Maternal and Infant Health Care”, it contains, among other more general and uncontroversial proposals, clauses that are of profound significance for the application and perception of genetics far beyond the boundaries of China itself. Whether this law will lead to what would generally be considered the abuse of genetics remains to be seen, but the rulings would certainly legitimize, in the strict sense of the word, practice that would be unacceptable to the medical genetics community in most of the world.

Because of these wider implications, and because China itself contains one-third of the world’s population, it is worth looking closely at this development; having myself been peripherally involved over a long period, and having found that many professionals in genetics are entirely unaware of the whole topic, I give here some background material that may help to put it into perspective.

First, it is relevant to quote (from the official Chinese translation) [1] some of the clauses in the law that specifically involve genetic disorders.

LAW OF THE PEOPLE’S REPUBLIC OF CHINA IN MATERNAL AND INFANT HEALTH CARE

Adopted at the 10th Meeting of the Standing Committee of the Eighth National People’s Congress on 27 October 1994, promulgated by Order No. 33 of the President of the People’s Republic of China on 27 October 1994, and effective as of 1 June 1995.

Article 8 The pre-marital physical check-up shall include the examination of the following diseases:

(i) genetic diseases of a serious nature;
(ii) target infectious diseases; and
(iii) relevant mental disease.
The medical and health institution shall issue a certificate of pre-marital medical check-up thereafter.

- **Article 10** Physicians shall, after performing the pre-marital physical check-up, explain and give medical advice to both the male and the female who have been diagnosed with certain genetic disease of a serious nature which is considered to be inappropriate for child-bearing from a medical point of view; the two may be married only if both sides agree to take long-term contraceptive measures or to take litigation operation for sterility. However, the marriage that is forbidden as stipulated by the provisions of the Marriage Law of the People's Republic of China is not included herein.

- **Article 16** If a physician detects or suspects that a married couple in their child-bearing age suffer from genetic disease of a serious nature, the physician shall give medical advice to the couple, and the couple in their child-bearing age shall take measures in accordance with the physician's medical advice.

- **Article 18** The Physician shall explain to the married couple and give them medical advice for a termination of pregnancy if one of the following cases is detected in the prenatal diagnosis:

(i) the fetus is suffering from genetic disease of a serious nature;
(ii) the fetus is with defect of a serious nature; and
(iii) continued pregnancy may threaten the life and safety of the pregnant woman or seriously impair her health due to the serious disease she suffers from.

**Supplementary provisions**

'Genetic diseases of a serious nature' refer to diseases that are caused by genetic factors congenitally, that may totally or partially deprive the victim of the ability to live independently, that are highly possible to recur in generations to come, and that are medically considered inappropriate for reproduction;

'Relevant mental diseases', refer to schizophrenia, manic-depressive psychosis and other mental diseases of a serious nature.

It could of course be argued, (and has been within China) that these proposals are simply the practical way of a country with relatively undeveloped services trying to ensure that prenatal diagnosis of genetic disorders and comparable measures are actually made available to its population; also that with a 'one child policy', such as already exists in China, it is important to ensure that the child born does not have avoidable handicap. It is certainly true that the law stipulates that decisions are to be made by appropriately trained people (article 26), while fetal sexing on non-medical grounds is specifically prohibited (article 32).

It is impossible though to deny the directive, even coercive tenor of the genetic clauses in the law, while its linkage with infectious diseases and mental illness makes it clear that genetic disorders are being considered primarily as a public health issue.

Why should China have produced a law of this type at a time when virtually all other countries have moved away from restrictive or eugenic legislation for
genetic disorders? It is difficult for an outsider to be sure on this, but that broader political factors have been involved is clear from the following official commentary on the draft version of the law, produced a year before the final form [2].

HEALTH MINISTER PRESENTS EUGENICS LAW TO NPC STANDING COMMITTEE

(a) Xinhua new agency, Beijing in English 1114 Greenwich Mean Time (GMT), 20 Dec. 93.

Text of report

China is to use legal means to avoid new births of inferior quality and heighten the standards of the whole population. The measures include deferring the date of marriage, terminating pregnancies and sterilization, according to a draft law on eugenics and health protection which was presented to the current session of the Eighth National People’s Congress (NPC) Standing Committee.

Explaining the law to participants at an NPC session that opened here today (Beijing, 20th December) Minister of Public Health, Chen Minzhang, said that the measures will help prevent infections and hereditary diseases and protect the health of mothers and children.

Under the draft law, those having such ailments as hepatitis, venereal disease or mental illness, which can be passed on through birth, will be banned from marrying while carrying the disease. Pregnant women who have been diagnosed as having certain infectious diseases or an abnormal foetus will be advised to halt the pregnancy. Couples in the category should have themselves sterilized, the draft says.

China is in urgent need of adopting such a law to put a stop to the prevalence of abnormal births. Minister Chen explained statistics show that China now has more than 10 million disabled persons who could have been prevented through better controls.

The draft also stipulates that organizations that are engaged in pre-marital checks, eugenics, pre-birth diagnosis or sterilizations should be approved by the authorities at the county level and above. Chen said, “Personnel involved in this area should be subjected to strict training”.

The Minister of Public Health called on medical authorities at various levels to establish a comprehensive network for the implementation of the law.

The draft does not state whether China will adopt euthanasia to eliminate congenitally abnormal children, saying that the international community has not come to a conclusion on that issue. The draft also does not touch on the issues of artificial fertilization or test-tube babies because the effects of these techniques have caused some disputes and because it’s too early to put any limitations into law.
Is China’s law eugenic?

China’s approach to family planning has been attacked in the West as authoritarian and an infringement on individual rights. Below, Chinese Academician Qiu Renzong rejects claims that his country’s Law on Maternal and Infant Health is eugenic. Overleaf, a German Sinologist challenges Qiu Renzong’s position.

1. ‘A concern for collective good’

Qiu Renzong, Bioethics programme director, Chinese Academy of Social Sciences, Beijing.

China’s Law on Maternal and Infant Health (see box opposite page) has attracted considerable criticism in the Western media and scientific circles. Some of the criticism is valid but some is based on misunderstandings caused by linguistic or cultural barriers. Much of the confusion revolves around the word yousheng, which repeatedly occurs in the legal text. A tricky word with dual meanings, it is commonly used to mean “healthy births” in association with child-rearing. However, yousheng can also be used to describe eugenic programmes such as that practised by the Nazis. Unfortunately, English translations of the law tend to reflect this latter meaning.

Is the Maternal and Infant Health Law eugenic? I would argue that for a policy to be eugenic it must first reject individual consent and second, be based on racism. Neither of these conditions applies to China’s law.

While doctors may advise two individuals at risk of passing on hereditary disease to refrain from marrying or to undergo sterilization, the ultimate decision is left to these adults. When prenatal testing reveals genetic disease, a doctor will offer advice—not a directive—concerning abortion.

The way to a higher domain
It is also crucial to recognize that the law is not motivated by racism but by a desire to reduce birth defects. Indeed, there is no racist tradition in China. The Chinese have been the victims of Western imperialism and Japanese militarism. They may have made grave
mistakes, but they have never claimed superiority over another people, and their military actions have never been motivated by racism. Nor is racism part of China's internal policies. The Han, China's dominant ethnic group, do not claim superiority over China's minorities. Westerners are often shocked by Chinese attitudes to defective foetuses because they do not understand the cultural and economic factors involved. The great Confucianist Xun Zi (300-237 BC) said: "Birth is the beginning of a human being, and death is the end of a human being. A human being who has a good beginning and a good end fulfills the Tao [the Way to a higher spiritual domain]." Two major factors shaping genetic policy in China emerge from this Confucian view. First, abortion is morally and socially acceptable because life begins with birth. A foetus is not considered a human being. Second, congenital disease and deformity are considered a sign of sin committed by the parents or ancestors in their previous life. Given that a defective newborn child is traditionally called a "monster foetus", it is not surprising to find little in the way of familial or social support. One of the parents of a deformed baby will usually have to stop working, and the costs of caring for such a child can amount to a third of an average worker's salary.

Poverty
Changing these negative attitudes will take a great deal of time. There are now more than 50 million handicapped people, mostly living in poverty, and it is unreasonable to expect any major improvements in the treatment of handicapped children and their mothers in the near future. In this context, many feel that these children and their mothers would be better off if the handicapped had never been born. In fact, the Chinese Association of the Handicapped formally urged the government in 1989 to speed up legislation to prevent the birth of deformed babies, given their suffering and the burden they represent for society.

The concern for the collective good has at times led geneticists and others in China to infringe upon individual autonomy. They have confused what is technologically possible (genetic testing) with what is ethically permissible. However, I feel that the law is a positive step towards guaranteeing everyone access to genetic counselling and to prohibiting sex-selection. Chinese geneticists and bioethicists have criticized some articles of the law. Their suggestions include more explicit recognition of the principle of informed consent. Last year, the authorities consulted leading Chinese bioethicists and geneticists and will make the needed changes at an appropriate time. Meanwhile, I
STATEMENT OF THE BOARD OF DIRECTORS OF THE AMERICAN SOCIETY OF HUMAN GENETICS:
Eugenics and The Misuse of Genetic Information to Restrict Reproductive Freedom

Approved by the ASHG Board of Directors, October 1998

Introduction

The global scientific community is making extraordinary advances in understanding the human genome. This knowledge has contributed many important medical benefits. Yet, concern about the possibility of misuse of genetic concepts and genetic information may be as great today as at any time since World War II. Many fear that as we learn more about how genes vary and function, some individuals or institutions may be tempted to ascribe an overly deterministic influence to their role in shaping human health and potential and pursue social policies that limit or constrain reproductive freedom.

Therefore, the Board of Directors of the American Society of Human Genetics reaffirms its commitment to the fundamental principle of reproductive freedom and unequivocally declares its opposition to coercion based on genetic information.

Statement

The American Society of Human Genetics recognizes that genetic variation can significantly influence risk for disease and the nature of an individual's future health and that many human capacities and talents are influenced by genes.

The American Society of Human Genetics deplores laws, governmental regulations and any other coercive effort intended to restrict reproductive freedom or constrain freedom of choice on the basis of known or presumed genetic characteristics of potential parents or the anticipated genetic characteristics, health or capacities of potential offspring.

The American Society of Human Genetics recognizes the need for international cooperation to protect reproductive freedom and stands ready to work with colleagues in and outside the field of human genetics to achieve this goal.

The American Society of Human Genetics believes that the best way to prevent genetic information from being used to restrict reproductive freedom is to educate the public (in particular those directly involved in setting public policy) about the scope and limitations of our understanding of genetics and genetic tests. It is especially important that individuals be educated about how to ask for and obtain appropriate genetic information and that health care providers be educated to assist them.

Background

A Note on Language
The drafting of this document was complicated by the substantial variations in meaning given to the word "eugenics". Ultimately, the drafters decided to de-emphasize that word. Yet, because on many occasions during this century scientifically unsound and socially harmful policies have been implemented in many nations in the name of eugenics, a comment on the term is warranted.
When Francis Galton (1883) coined the term eugenics, he took it from the Greek; eu means "good" and genic derives from the word for "born". Galton defined it as "the science of improvement of the human race germ plasm through better breeding." At the height of the eugenics movement in the 1920s, the Encyclopedia Britannica (1926) entry on eugenics emphasized that the term connoted a "plan" to influence human reproduction. A typical modern dictionary definition is "a science that deals with the improvement (as by control of human mating) of hereditary qualities of a race or breed" (Webster's 1983). Although it is not apparent from the dictionary definition, the word has a pejorative connotation, and is frequently used in reference to governmentally driven policies to limit reproductive freedom. Knowledge-based decisions made by individuals or couples to avoid the birth of a child with disease or disability, so long as they are not unduly influenced by coercive governmental, institutional, or other policies, are acceptable. Many public health practices to improve the health of living or future people have been implemented to achieve laudable goals. Examples include newborn screening programs to identify infants with disorders for which early treatment is beneficial, the provision of prenatal diagnostic services, maternal vaccination for rubella, addition of folic acid to food to reduce the risk of certain birth defects, and warnings on alcohol or cigarette labels about the potential for damage to the fetus. The American Society of Human Genetics views prenatal screening and diagnostic programs, including those undertaken with the knowledge that an individual who chooses to be tested may seek selective termination of pregnancy, as acceptable so long as individuals are not coerced.

Historical Note

Many nations have a history of eugenic thought or practice based on perceived genetic risks. It is important to note that such practices were based on little or no scientifically defensible beliefs. Some have tried to keep gene pools separate by forbidding unions between members of different social groups. For example, the caste system in India may represent the largest such eugenic program ever, spanning almost 2500 years (Dobzhansky 1973). Anti-miscegenation laws in the United States, which appeared as early as 1630 in the colonies and existed until they were struck down as unconstitutional in 1967, were premised in part on the erroneous notion that interracial marriage produced children of reduced genetic quality.

Galton used the word eugenics to characterize efforts to produce children who would be well born. However, he did not merely desire that as many infants as possible be born healthy. His real goal was to insure that as large a fraction as possible of each generation be the offspring of what he considered the best "stock." By 1883 Galton, who then had been studying human heredity for almost 20 years, was convinced that the British upper classes were having too few children to maintain what he considered their crucially important contribution to the gene pool of Victorian England. He exhorted the upper classes to have more children. Over the next 30 years this idea garnered much interest. Among its most famous proponents in the United States was President Theodore Roosevelt, who warned that the failure of couples of Anglo-Saxon heritage to have large enough families would lead to "race suicide" (Reilly 1991). Roosevelt's support of eugenic ideals reflects the popular appeal of eugenics during the first half of this century. Adherents included liberals and conservatives, progressives and libertarians. In the early decades of this century the emphasis on encouraging reproduction among those assumed to possess a superior genetic endowment became known as "positive eugenics."

The term immediately suggests a contrasting policy, "negative eugenics", which emerged at about the same time. The goal of negative eugenics is the restriction of parenting by "undesirable" individuals, presumably because of a strong likelihood that their children would be "unfit". During the first half of the twentieth century, the United States, implemented two "negative eugenics" programs. The United States immigration policy that was erected in the 1920s and dismantled in 1968 favored immigrants from northern and western Europe over other peoples. It was rationalized during Congressional testimony by a self-described eugenics
expert who strongly favored the quota system that became the centerpiece of the law (Reilly 1991). The United States never enacted a federal sterilization statute, but about 30 states did, many after the Supreme Court upheld a Virginia law that permitted state officials to sterilize institutionalized retarded persons whom a physician determined likely to become the parent of children with similar deficits (Buck v. Bell 1927). Between 1907 and 1960 in the United States at least 60,000 people were sterilized without their consent pursuant to these state laws. During the 1930s, the heyday of these programs, about 5,000 persons were sterilized each year. The majority were young women for many of whom the evidence of genetically caused mental retardation was poor or non-existent (Reilly 1991). Geneticists were not active participants in these programs; with few exceptions, however, neither were they public critics.

England never enacted an involuntary sterilization law, nor launched a coercive private effort. In Canada, the Province of Alberta was strongly influenced by sterilization programs in the United States. Alberta had an active program from 1928 until 1960, pursuant to which several thousand people were sterilized (Caulfield and Robertson 1996). A class action lawsuit by many of the surviving individuals was recently settled with the government (Muir 1996).

Although arguments for maintaining racial purity abound in nineteenth century German literature, the Nazis were also influenced by events in the United States. The 1934 German racial hygiene law relied on a model bill written by the American eugenicist, Harry Hamilton Laughlin, who for three decades directed the Eugenics Record Office at Cold Spring Harbor. In its first full year of operation the Nazi program dramatically eclipsed activities in the United States, sterilizing about 80,000 persons without their consent. The much grander scope was achieved because the Nazi law applied to the entire population (rather than institutionalized persons), created a system of "hereditary health courts" designed exclusively to hear and process petitions for sterilization, and permitted petitions proposing that an individual should be sterilized to be filed by a broad range of citizens.

The German sterilization program quickly evolved to target and eliminate retarded and epileptic children, the mentally ill, and other groups. The program has been called a precursor to the gas chambers. During the early years (1934-38) the Nazi sterilization program was not primarily an attempt to improve the gene pool. It focused on eliminating "useless eaters" - persons who would consume resources without contributing to their production. One exception was persons with Huntington disease. It was a stated goal of the Nazis to sterilize as many persons at risk for this disorder as possible. The Nazi sterilization program owed part of its success to the efficiency with which the government maintained patient registries which made it comparatively easy to locate persons with various disorders (Burleigh 1994).

Often overlooked in discussions of Nazi eugenic practices are the sterilization programs that were implemented during the 1930s in other European countries (Adams, 1990) as well as in other nations around the globe. In smaller nations (for example, Sweden, which had an active eugenic sterilization program until the 1960s), the impact of the programs was proportionately larger than in the United States.

After World War II (1948) Japan passed a Eugenic Protection Law that permitted the sterilization of persons who had even distant relatives with any one of about 30 (presumably and, in most cases, erroneously) inherited conditions (Tsuchiya 1997). Japan's law was amended in 1996, in part to remove the term eugenic. We know of no firm evidence that it was applied coercively.

Over the last 20 years a few governmentally supported public health programs have focused on reducing the number of births of children with specific disorders. In some cases voluntary public response to these programs has led to a substantial reduction. Examples include the rapid decline in the United Kingdom in the number of children born with neural tube defects (Cuckle and Wald 1987) and the public health campaigns to reduce the
number of children born with beta-thalassemia in Sardinia (Cao et al 1989) and Cyprus (Angostiniotis et al 1986).

**Current Programs that May Restrict Reproductive Freedom**

There are few public health programs operating in the world today that may be said to use genetic information to restrict reproductive freedom.

Singapore has implemented a policy of using economic incentives to encourage reproduction by educated women and to encourage sterilization among uneducated, poor women, but it does not rely on genetic information and is not mandatory (Chan 1985).

China's Maternal and Infant Health Care Law (1994) has aroused concern because it appears to require medical counseling before marriage for people whose families have a relative with one of a listed group of conditions (including mental illness, epilepsy, and mental retardation) that the law presumes (with little or no scientific basis) are hereditary. The law (the official translation of which involves nuances of language that complicate analysis) also has been construed to require sterilization or long-term contraception as a precondition of marriage if a person is determined by the doctor to be at risk for bearing an affected child. Another section of the Chinese law appears to require that couples at risk for certain disorders must undergo prenatal diagnosis and follow the directive of the attending physician.

However, the law includes no penalty for non-compliance and (to the best of our knowledge) is not enforced. It seems to represent a "standard of care," albeit highly directive, to which the government aspires rather than a rule of conduct that must be obeyed. The official English translation of the law uses the word "shall" in a manner that connotes compulsion, but some Chinese bioethicists insist that it is meant to connote "ought", e.g. an ethical obligation, rather than a legal rule (Qiu 1998). China's human geneticists, recognizing the importance of even symbolic language that seems to embrace eugenics, have requested that the central government change the law to comply with international concern, and to acknowledge the centrality of voluntary choice in genetic testing and counseling (Yang 1998). Taiwan has had a similar law (Sung 1998). on its books for several years, which has neither been enforced nor drawn international criticism.

Many governments support programs in the interests of improving the odds that children will be healthy. Some are mandatory. In our view, none involve the misuse of genetic information. Examples include: 1) programs to encourage or discourage the number of births among the entire population, 2) laws that try to protect the fetus from environmental harm (e.g. warnings on cigarette packages about the risk of smoking during pregnancy), 3) laws that implement newborn genetic screening programs, 4) laws or regulations that fund genetic services, including genetic counseling, genetic testing, prenatal diagnosis, and the provision of special diets for newborns with certain inborn errors of metabolism (Cunningham 1998), and 5) laws forbidding first cousin marriages and other consanguineous unions.

**Conclusion**

Efforts to implement programs that restrict reproductive freedom based on genetic information are scientifically and ethically unacceptable and should be challenged. While it is sometimes possible to ascertain the risk of bearing a child with a genetic disorder, for the majority of pregnancies it is not possible to make predictions about a future child's health or other capacities. Misguided efforts to do so devalue humanity.

**Acknowledgments**

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Key words: eugenics, genetics, reproductive freedom

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better "left behind in the cast-off junk of ignorant efforts, with which the past is filled."

By the outbreak of the First World War, sterilization laws were in such dispute as to have been de facto suspended in their operation in a number of states. The courts had also declared unconstitutional not only the stringent Iowa statute but less sweeping measures in six other states. Advocates of eugenic sterilization, frustrated at the legal impasse, wanted to take the issue to the Supreme Court. In Virginia, eugenicists helped draw up a sterilization statute, passed by the legislature in March 1924, that was designed to meet the constitutional objections. The opportunity to press a test case arose that June, when a seventeen-year-old girl named Carrie Buck, who seemed definable as a "feebleminded," was committed to the Virginia Colony for Epileptics and Feebleminded, in Lynchburg.24

Carrie's mother, Emma, had lived at the Colony since 1920 and was also certified to be feebleminded. Carrie herself had conceived a child out of wedlock, and shortly before her commitment, she gave birth to a daughter, Vivian. Carrie was given the Stanford revision of the Binet-Simon test and was found to have a mental age of nine years, well within Henry Goddard's definition of "moron." Carrie's mother was found to have a mental age of slightly under eight years. Thus, according to these results, there was mental deficiency in two successive generations. If Vivian could be shown to be feebleminded too, Carrie would be a perfect subject for a test of the Virginia sterilization statute. In September 1924, the Colony's board of directors ordered Carrie Buck sterilized, and a court-appointed guardian initiated legal proceedings by appealing the order in a suit on Carrie's behalf against the superintendent of the Colony, Albert S. Frick.25

In preparing their case, Virginia officials consulted Harry Laughlin at the Eugenics Record Office. Laughlin examined the pedigrees of Carrie, her mother, and her daughter, and information about them given by Colony officials, and—without ever having seen them in person—provided an expert deposition that Carrie's alleged feeblemindedness was primarily hereditary. Carrie and her forebears, Laughlin submitted, "belong to the shiftless, ignorant, and worthless class of anti-social whites of the South." At the time of Laughlin's deposition, however, there was no evidence at all that Vivian was mentally deficient. To clarify the matter, Caroline E. Wilhelm, a Red Cross worker who had placed Vivian in a foster home, was prevailed upon to examine her there. At the initial hearing, in the Circuit Court of Amherst County, she testified that there was "a look about Vivian (who at the time of the visit was seven months old) which was not quite normal."26 Evidence also came from Arthur Estabrook of the Eugenics Record Office, who had subjected Vivian to a mental test for an infant and concluded that she was below average for a child her age. In the court proceeding, Estabrook testified that the feeblemindedness in the Buck line conformed to the Mendelian laws of inheritance, and the judge upheld the sterilization order.27

The case—now known as Buck v. Bell, because Priddy had in the meantime died and been replaced as the defendant by the Colony’s new superintendent, John H. Bell—was carried to the Virginia Supreme Court of Appeals in 1925, and the sterilization order was again upheld. In April 1927 it was argued before the United States Supreme Court. Carrie's defense counsel, I. P. Whitehead, a sometime member of the board of directors of the Colony, attacked the sterilization statute, warning that under this type of law "a reign of doctors will be inaugurated and in the name of science new classes will be added, even races may be brought within the scope of such a regulation and the worst forms of tyranny practiced." Nevertheless, the Court was persuaded not only that Carrie Buck and her mother were "feebleminded" but also—because Vivian was, too (or so all the experts said)—that the feeblemindedness was heritable. The Court, whose membership ranged in political conviction from William Howard Taft to Louis D. Brandeis, upheld the Virginia statute by a vote of eight to one. The sole dissent was Justice Pierce Butler, a conservative, and he kept his minority opinion to himself. The decision declared that sterilization on eugenic grounds was within the police power of the state, that it provided due process of law, and that it did not constitute cruel or unusual punishment.

The Court's opinion was written by Justice Oliver Wendell Holmes, an enthusiastic science as a guide to social action, who managed to find a link between eugenics and patriotism: "We have seen more than once that the public welfare may call upon the best citizens for their lives. It would be strange if it could not call upon those who already sap the strength of the State for these lesser sacrifices . . . in order to prevent our being swamped with incompetents. . . . The principle that sustains compulsory vaccination is broad enough to cover cutting the Fallopian tubes." With deliberate panache Holmes asserted: "Three generations of imbeciles are enough."28

Eugenacists naturally rejoiced at Buck v. Bell. For some years prior to the decision, the American Eugenics Society had promoted what it thought might be a constitutional revision of the faulty sterilization statutes. Apart from procedural and technical changes, the revisions centered on making the laws eugenic rather than punitive in intent. After Buck v. Bell, what was constitutional was clear. By the end of the nineteen-twenties, sterilization laws were on the books of twenty-four states, with the South no longer a regional exception. (Though now severely restricted by federal regulation, they are still on the books of twenty-two states today.) The laws were not uniformly enforced, but Carrie Buck was sterilized soon after the Court's
Is Genetic Information Unique and Different from Other Medical Information?

1. Reveals Parentage
2. Reveals Sex
3. Reveals presence of Disease Genes
4. Can Reveal Potential Future Health Risks
   e.g., early onset Alzheimer's disease, Huntington's disease, inherited forms of cancer
5. Can Reveal Potential Health Risks of Family Members
6. Can Reveal Future Reproductive Options
   e.g., if carrier of genetic disease gene - prenatal testing or pre-implantation genetic diagnosis on embryo cells
7. Can be regarded as "Unique" by third parties who might want to misuse it!

Should Genetic Information be Protected Separately?

1. Recent legislation suggests yes - genetic information needs to be protected specifically - unique vs. predictive is distinct from normal medical records
2. Contrary Argument - not possible to separate them - clinical records: genetic data similar to other medical information
I. FEDERAL POLICY HISTORY

No federal legislation has been passed relating to genetic discrimination in individual insurance coverage or to genetic discrimination in the workplace. Several bills were introduced during the last decade. Some of these bills attempted to amend existing civil rights and labor laws, while others stood alone. The primary public concerns are that (1) insurers will use genetic information to deny, limit, or cancel insurance policies or (2) employers will use genetic information against existing workers or to screen potential employees. Because DNA samples can be held indefinitely, there is the added threat that samples will be used for purposes other than those for which they were gathered.

Executive Order Protecting Federal Employees
On February 6, 2000, U.S. President Clinton signed an executive order prohibiting every federal department and agency from using genetic information in any hiring or promotion action. This executive order, endorsed by the American Medical Association, the American College of Medical Genetics, the National Society of Genetic Counselors, and the Genetic Alliance.

- Prohibits federal employers from requiring or requesting genetic tests as a condition of being hired or receiving benefits. Employers cannot request or require employees to undergo genetic tests in order to evaluate an employee's ability to perform his or her job.
- Prohibits federal employers from using protected genetic information to classify employees in a manner that deprives them of advancement opportunities. Employers cannot deny employees promotions or overseas posts because of a genetic predisposition for certain illnesses.
- Provides strong privacy protections to any genetic information used for medical treatment and research. Under the EO, obtaining or disclosing genetic information about employees or potential employees is prohibited, except when it is necessary to provide medical treatment to employees, ensure workplace health and safety, or provide occupational health and safety data. In every case where genetic information about employees is obtained, it will be subject to all Federal and state privacy protections.

U.S. House of Representatives Committee on Energy and Commerce

Senate Committee on Health, Education, Labor, and Pensions
Hearing on Genetic Information in the Workplace. July 20, 2000

II. STATE POLICY HISTORY

States have a patchwork of genetic-information nondiscrimination laws, none of them comprehensive. Existing state laws differ in coverage, protections afforded, and enforcement schemes. Some of the first state laws enacted to address this issue prohibited discrimination against individuals with specific genetic traits or disorders. Other state laws regulate both the use of genetic testing in employment decisions and the disclosure of genetic test results. These state laws generally prohibit employers from requiring workers and applicants to undergo genetic testing as a condition of employment. Some states permit genetic testing when it is requested by the worker or applicant for the purpose of investigating a compensation claim or determining the worker's susceptibility to potentially toxic chemicals in the workplace. These statutes often require the employer to provide informed written consent for such testing, contain specific restrictions governing disclosure, and prevent the employer from taking adverse action against the employee.

[See charts of state genetics laws and information on genetics legislative activity on the National Conference of State Legislatures Web site. See the NIH NHGRI chart of all genetics insurance discrimination legislation and the NIH NHGRI chart of all genetics workplace discrimination legislation that has been enacted at the state level as of April 29, 2002.]

State Genetics Reports
- HI: The Challenges of Human Cloning for Public Policy in Illinois (February 2001)
- OR: Assuring Genetic Privacy in Oregon (November 2000)
- KY: Genetic Testing in Health Care and Disability Insurance in Kentucky (January 2000)
- MI: Report of the Michigan Commission on Genetic Privacy and Progress (February 1999)
- WA: Genetic Privacy, Discrimination, and Research in Washington State (October 2002)
- WI: Genetic Services Plan for Wisconsin

III. EXISTING FEDERAL ANTI-DISCRIMINATION LAWS AND HOW THEY APPLY TO GENETICS

Although no specific federal genetic nondiscrimination legislation has been enacted, some believe that parts of existing nondiscrimination laws could be interpreted to include genetic discrimination. Here is a brief overview of these laws and how they apply to genetics.

Americans with Disabilities Act of 1990 (ADA)
The most likely current source of protection against genetic discrimination in the workplace is provided by laws prohibiting discrimination based on disability. Title I of the Americans with Disabilities Act (ADA), enforced by the Equal Employment Opportunity Commission (EEOC), and similar disability-based antidiscrimination laws such as the Rehabilitation Act of 1973 do not explicitly address genetic information, but they provide some protections against disability-related genetic discrimination in the workplace.

http://www.ortho.gov/TAI/ResourcesHuman_Genome/Policy/legislative.html
Prohibits discrimination against a person who is regarded as having a disability.

- Protects individuals with symptomatic genetic disabilities the same as individuals with other disabilities.
- Does not protect individuals with expressed or unexpressed genetic conditions.
- Does not protect potential workers from requirements or requests to provide genetic information to their employers after a conditional offer of employment has been extended but before they begin work. (Note: this is a heightened concern because genetic samples can be stored.)

- Does not protect workers from requirements to provide medical information that is job related and consistent with business necessity.

In March 1995, the EEOC issued an interpretation of the ADA. The guidance, however, is limited in scope and legal effect. It is policy guidance that does not have the same legal binding effect on a court as a statute or regulation and has not been tested in court. According to the interpretation:

- Entities that discriminate on the basis of genetic predisposition are regarding the individuals as having impairments, and such individuals are covered by the ADA.
- Unaffected carriers of recessive and X-linked disorders, individuals with late-onset genetic disorders who may be identified through genetic testing or family history as being at high risk of developing the disease are not covered by the ADA.

**Health Insurance Portability and Accountability Act of 1996 (HIPAA)**

The Health Insurance Portability and Accountability Act (HIPAA) applies to employer-based and commercially issued group health insurance only. HIPAA is the only federal law that directly addresses the issue of genetic discrimination. There is no similar law applying to private individuals seeking health insurance in the individual market. HIPAA:

- Prohibits group health plans from using any health status-related factor, including genetic information, as a basis for denying or limiting eligibility for coverage or for charging an individual more for coverage.
- Limits exclusions for preexisting conditions in group health plans to 12 months and prohibits such exclusions if the individual has been covered previously for that condition for 12 months or more.
- States explicitly that genetic information in the absence of a current diagnosis of illness shall not be considered a preexisting condition.
- Does not prohibit employers from refusing to offer health coverage as part of their benefits packages.

**HIPAA National Standards to Protect Patients' Personal Medical Records, Dec. 2002**

This regulation would protect medical records and other personal health information maintained by health care providers, hospitals, health plans and health insurers, and health care clearinghouses. The regulation was mandated when Congress failed to pass legislation as directed by HIPAA by 1999. The new standards: limit the non-consensual use and release of private health information; give patients new rights to access their medical records; and to know who else has accessed them; restrict most disclosure of health information to requirements for access to records by researchers and others. They are not specific to genetics, rather they are sweeping regulations governing all personal health information.

For more on the standards, see:

- U.S. Department of Health and Human Services (DHHS) Announces Final Regulation Establishing First-ever National Standards to Protect Patients' Personal Medical Records: DHHS Press Release
- Summary of the Final Regulation: DHHS Fact Sheet

**Title VII of the Civil Rights Act of 1964**

An argument could be made that genetic discrimination based on racially or ethnically linked genetic disorders constitutes unlawful race or ethnicity discrimination.

- Protection is available only where an employer engages in discrimination based on a genetic trait that is substantially related to a particular race or ethnic group.
- A strong relationship between race or national origin has been established for only a few diseases.

**IV. RECOMMENDATIONS FOR FUTURE LEGISLATION**

**Workplace Discrimination**

Based on previous recommendations from the National Action Plan on Breast Cancer (NAPBC) and the NIH-DOE Working Group on the Ethical, Legal, and Social Implications (ELS) of human genome research, in 1998 report the Clinton Administration announced recommendations for future legislation to ensure that discoveries made possible by the Human Genome Project are used to improve health and not to discriminate against workers or their families. These recommendations are:

- Employers should not require or request that employees or potential employees take a genetic test or provide genetic information as condition of employment or benefits.
- Employers should not use genetic information to discriminate against, limit, segregate, or classify employees in a way that would deprive them of employment opportunities.
- Employers should not obtain or disclose genetic information about employees or potential employees under most circumstances.

Genetic testing and the use of genetic information by employers should be permitted in the following situations to ensure workplace safety and health and to preserve research opportunities. However, in all cases where genetic information about employees is obtained, the information should be maintained in medical files that are kept separate from personnel files, treated as confidential medical records, and protected by applicable state and federal laws.

An employer should be permitted to monitor employees for the effects of a particular substance found in the workplace to which continued exposure could cause genetic damage under certain circumstances. Informed consent and assurance of confidentiality should be required. In addition, employers may use the results only to identify and control adverse conditions in the workplace and to take action necessary to prevent significant risk of substantial harm to the employee or others.

The statutory authority of a federal agency to promulgate regulations, enforce workplace safety and health laws, or conduct occupational or other health research should not be limited.

An employer should be able to disclose genetic information for research and other purposes with the written, informed consent of the individual.

These recommendations should apply to public and private-sector employers, unions, and labor-management groups that conduct joint apprenticeship and other training programs, employment agencies and licensing agencies that issue licenses, certificates, and other credentials required to engage in various professions and occupations also should be covered.

Individuals who believe they have been subjected to workplace discrimination based on genetic information should be able to file a charge with the Equal Employment Opportunity Commission. Department of Labor, or other appropriate federal agency for investigation and resolution. The designated agency should be authorized to bring lawsuits in the federal courts to resolve issues that would not settle amicably. The courts should have the authority to halt the violations and order relief, such as hiring, promotion, back pay, and compensatory

http://www.oml.gov/Test/Resources/Human_Genome/ase/legisl.html
and punitive damages to the individual. Alternatively, an individual should be able to elect to bring a private lawsuit in federal or state court to obtain the same type of relief plus reasonable costs and attorney's fees. To enforce these protections, the designated enforcement agency must be given sufficient additional resources to investigate and prosecute allegations of discrimination.

**Insurance Discrimination**

In 1995, the NIH-DOE Joint Working Group on Ethical, Legal, and Social Implications of Human Genome Research (ELSI Working Group) and the National Action Plan on Breast Cancer (NAPBC) developed and published the following recommendations for state and federal policymakers to protect against genetic discrimination (Science, vol. 270, Oct. 20, 1995):

**Definitions**

- "Genetic information" is information about genes, gene products, or inherited characteristics that may derive from the individual or a family member.
- "Insurance provider" means an insurance company, employer, or any other entity providing a plan of health insurance or health benefits, including group and individual health plans whether fully insured or self-funded.

**Recommendations**

- Insurance providers should be prohibited from using genetic information or an individual's request for genetic services to deny or limit any coverage or establish eligibility, continuation, enrollment, or contribution requirements.
- Insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information or an individual's request for genetic services.
- Insurance providers should be prohibited from requiring collection or disclosure of genetic information. Insurance providers and other holders of genetic information should be prohibited from releasing genetic information without the individual's prior written authorization. Written authorization should be required for each disclosure and include to whom the disclosure would be made.

**Sample Genetic Privacy Act and Commentary**

A draft bill (Genetic Privacy Act) was written in 1995 by George Annas of the Boston University School of Public Health to assist legislators. This sample bill proposed that access to information in genetic data banks should be regulated during sample collection, storage, disclosure, and use. Several state lawmakers adapted language and concepts from the draft bill to write proposals for legislation in their own states.

**V. WHY LEGISLATION IS NEEDED NOW**

1. Based on genetic information, employers may try to avoid hiring workers they believe are likely to take sick leave, resign, or retire early for health reasons (creating extra costs in recruiting and training new staff), file for workers' compensation, or use healthcare benefits excessively.

2. Some employers may seek to use genetic tests to discriminate against workers—even those who do not and may never show signs of disease—because the employers fear the cost consequences.

3. The economic incentive to discriminate based on genetic information is likely to increase as genetic research advances and the costs of genetic testing decrease.

4. Genetic predisposition or conditions can lead to workplace discrimination, even in cases where workers are healthy and unlikely to develop disease or where the genetic condition has no effect on the ability to perform work.

5. Given the substantial gaps in state and federal protections against employment discrimination based on genetic information, comprehensive federal legislation is needed to ensure that advances in genetic technology and research are used to address the health needs of the nation—and not to deny individuals employment opportunities and benefits. Federal legislation would establish minimum protections that could be supplemented by state laws.

6. Insurers can still use genetic information in the individual market in decisions about coverage, enrollment, and premiums.

7. Insurers can still require individuals to take genetic tests.

8. Individuals are not protected from the disclosure of genetic information to insurers, plan sponsors (employers), and medical information bureaus, without their consent.

9. Penalties in HIPAA for discrimination and disclosure violations should be strengthened in order to ensure individuals of the protections afforded by the legislation.

**VI. MORE INFORMATION**

- NIH/NHGRI has a legislative policy page with details of previous legislation attempts and recommendations
- National Conference of State Legislatures' Genetics Legislation Project and Genomics Technologies Project
- Genetic Alliance Statement on Genetic Discrimination in Health Insurance and Employment Act, June 21, 2000
- UNESCO Universal Declaration on the Human Genome and Human Rights, November, 1997
- Freedom of Information Center article explains the latest rates for HIPAA. HHS Issues Privacy Rules for Use of Health Records, August, 2002
- Understanding HHS December 2002 HIPAA Privacy Guidance
- Privacy Rights Clearinghouse on How Private is My Medical Information?, October, 2002
- Health Privacy Project
POLICY ISSUES:
Preliminary Issues in the Genome Era

James M. Jeffords and Tom Daschle

The sequencing of the human genome heralds a new age in medicine, with enormous benefits for the general public. For example, it will allow scientists to identify all of the genes contributing to a given disease state, leading to a more accurate diagnosis and precise classification of disease severity. In addition, healthy patients can know the diseases for which they are at risk, giving them the opportunity to make beneficial lifestyle changes or to take preventive medications to protect their health. Understanding the genetic bases of heritable diseases also will allow researchers to develop therapeutics at the molecular level, resulting in better treatments with fewer side effects.

Despite the potential benefits, many ethical, legal, and social concerns exist. The U.S. Congress recognized this early in the development of the publicly funded human genome project and set aside approximately 5% of the budget, starting in 1990, to fund the ELSI program (Ethical, Legal, and Social Implications of Human Genomics Research) (1). Initially, the ELSI program focused efforts on four areas: Privacy and fair use of genetic information, clinical integration of genetic technologies, issues surrounding research ethics, and public and professional education. Later these goals were expanded to include studies of the societal impact of knowing the complete human genome sequence, the interpretation of genetic variations among individuals, integration of genetic technologies into clinical and nonclinical settings, and the implications of genetic technologies for religious, philosophical, ethical, and socioeconomic concerns.

One of the most difficult issues is determining the proper balance between privacy concerns and fair use of genetic information. The growing number and use of genetic tests has many worried about discrimination due to inappropriate access to, and use of, private genetic information. A survey in 1998 by the Institute for Human Rights and Freedom and in 1997 by the Human Rights Watch indicated that 86% of U.S. adults over the age of 18 believe that physicians should obtain permission before doing any genetic testing beyond routine testing (2). Similarly, 93% of adults believe that their permission should be granted before researchers use their genetic information. Francis Collins, Director of the National Human Genome Research Institute (NHGRI), has written, "It is estimated that all of us carry dozens of glitches in our DNA--so establishing principles of fair use of this information is important for all of us" (2). Without adequate safeguards, the genetic revolution could mean one step forward for science and two steps backwards for civil rights. Misuse of genetic information could create a new underclass: the genetically less fortunate.

Many Americans are concerned about potential genetic discrimination by their employers. In 1998 the National Center for Genome Resources (NCGR) surveyed 1000 American adults, and found that the majority (85%) believed that employers should not have access to a patient's genetic information, and 63% indicated they "probably" or "definitely" would not undergo genetic testing if they knew that insurers or employers could discover the results (4). However, members of the business community report that employment discrimination based on genetic information is currently very rare. The American Management Association surveyed 2133 employers this year, and of those surveyed, only 7 indicated that they used genetic testing, either for testing job applicants or employees (5).

However, it is important that this situation not become more prevalent, and even a perception of genetic discrimination can seriously impede future progress. Craig Venter put it succinctly: "...there are more barriers to achieving that era [of personalized and preventive medicine] than the scientific ones that have now been overcome. A key barrier is the fear that is pervasive in our society that genetic information will be used to deny health insurance or a job.... Without the enactment of legislation, I fear that this new era will be delayed" (6).

In the United States, federal laws such as the Americans with Disabilities Act and the Rehabilitation Act provide some protections against genetic discrimination in the workplace, but the scope of that coverage has not been tested in the courts (2). Former President Clinton recently signed an executive order barring genetic discrimination against employees in federal executive departments and agencies (4). Just this past November, the Society for Human Resource Management (SHRM) issued a policy position that stated, in part, "For this reason, the SHRM would oppose employment policies that permit employment decisions to be made based on an individual's genetic information" (2).

U.S. federal law does provide some protection against discrimination in health insurance. Specifically, the Health Insurance Portability and Accountability Act of 1996 (HIPAA) bars a group health plan, or an issuer of a group health plan, from using genetic information as a basis for implementing rules of eligibility for the plan or for setting premiums (6). But it does not cover people who buy insurance as individuals, or limit collection and disclosure of genetic information by issuers of individual policies (6). For example, definitions vary from state to state. One state may protect only DNA and RNA; another may extend protection to family history data and other medical information that could offer genetic clues. In addition, because of federal law preemptions, state laws do not protect the nearly one-in-three Americans who get their health insurance through their employer.

Ethical ambiguities are not limited to how genetic information will be made available and applied, but extend to the research methods used to gather the data in the first place. For example, in large community studies, obtaining informed consent from every community member is often impractical. Furthermore, studying groups of people with relatively small gene pools may have an unintentional stigmatizing effect. Policies protecting confidentiality in research are crucial both to guard individual privacy and to promote advancement of the science. Some organizations have published guidelines in this area. For example, general recommendations to protect privacy in genetic research have been published by members of the Privacy Workshop Planning Subcommittee of the National Action Plan on Breast Cancer (13).

Genetic information has begun to be catalogued and maintained in many different forms, such as pathology specimens, blood bank donations, newborn screening samples, and research collections. In addition, the U.S. Armed Forces require all members to donate a sample of their DNA for future casualty identification. Many countries including the United States maintain forensic DNA databases for use in criminal courts as well as commercial (6). Outside the United States, there have been efforts to create national genetic databases. For example, in December 1999, Iceland's parliament passed a bill allowing Decode Genetics, a biotechnology company, to combine all Icelanders' genetic, medical, and genealogical information into one database to be sold to customers. Critics of this research have expressed concerns over the "ownership rights" of genetic information, especially when a profit is to be made from the information (12). Estonian scientists are working to create a similar genetic database while also addressing concerns regarding access (12). Their goal is to provide the genetic information, as well as other health and lifestyle data, on more than 70% of the Estonian citizens. If established, the participants will receive access to their own genetic profiles in exchange for their contribution.

One of the most challenging areas of policy development involves genetic testing in the reproductive sciences. Research advances in this area have been remarkable, but are fraught with controversy. Couples considering pregnancy now have many options for genetic screening. In fact, those undergoing in vitro fertilization may now opt to have their embryos genetically screened before implantation (14). This can be helpful to couples whose offspring are known to be at risk for an inherited disease. Although some view this technology as a wonderful breakthrough, critics argue that it borders on eugenics.

In our lifetime, we have watched with amazement the progress of this field from the initial discovery of the structure of DNA in 1953 by Watson and Crick (15), to the present-day
sequencing of the human genome. Increased understanding of the human genome may ultimately result in the eradication of common diseases, but in the meantime we need to be on guard against potential misuse of genetic information. This is an emerging technology, and we should proceed with caution. The science is expanding at a breathtaking pace, and the overwhelming amount of new information puts governments under increasing pressure to pass legislation.

Eventually every country must decide what genetic information should be protected, who will have access to it, and how it may be used. In addition, governments must ensure that the public realizes practical gains from their investment in genetic technology, because much of the research is made possible by taxpayer-supported federal enterprises in partnership with academic and industrial institutions. Further, for this partnership to continue, the public must understand the new technologies so that unfounded fears will not develop and slow progress. Ultimately, the greatest difficulty will be for policy-makers to strike a balance between timely promotion and use of the best genetic research and careful protection of people from genetic discrimination.

Editor’s note: The authors have chosen to express their individual views about future directions for legislation in the United States separately.

Senator Jeffords:
As chairman of the U.S. Senate Committee on Health, Education, Labor, and Pensions, Senator Jeffords held a hearing on Genetic Information in the Workplace during the 106th Congress and a hearing on Genetic Information and Health Care during the 105th Congress. During the 106th Congress, Senator Jeffords joined with Senators Snowe and Frist in cosponsoring the Genetic Information Non-discrimination in Health Insurance Act. The bill is designed to protect American consumers from discrimination by health insurance companies based on predictive genetic information or the use of genetic services. It prohibits the use of this information by health insurers to set eligibility requirements or premium rates. It clearly specifies the very limited conditions under which a company may request genetic information from individuals. Furthermore, it calls for the establishment of safeguards within the insurance companies to protect the confidentiality of the individual’s genetic information. On 29 June 2000, the Senate adopted the measure as an amendment to the Labor/Health and Human Services Appropriations bill. It was subsequently removed by the Conference Committee. This bill will be reintroduced during the 107th Congress. Senator Jeffords’ Committee will continue its examination of issues surrounding the use of genetic information and workplace discrimination.

Senator Daschle:
I believe that Congress must pass strong federal laws against genetic discrimination. I believe that the United States should develop legislation that conforms to the Universal Declaration of the Human Genome and Human Rights: “No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.”

Thus, I believe that employment and health insurance discrimination on the basis of predictive genetic information should be firmly prohibited. Further, I believe that limits must be placed on the collection and disclosure of individuals’ genetic information. In crafting these protections, lawmakers should actively solicit opinions from others, including—at a minimum—scientists, geneticists, ethicists, consumers, employees, and employer groups, and insurers.

References and Notes
THE GENETIC PRIVACY ACT AND COMMENTARY

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INTRODUCTION

The Genetic Privacy Act is a proposal for federal legislation. The Act is based on the premise that genetic information is different from other types of personal information in ways that require special protection. The DNA molecule holds an extensive amount of currently indecipherable information. The major goal of the Human Genome Project is to decipher this code so that the information it contains is accessible. The privacy question is, accessible to whom?

The highly personal nature of the information contained in DNA can be illustrated by thinking of DNA as containing an individual’s “future diary.” A diary is perhaps the most personal and private document a person can create. It contains a person’s innermost thoughts and perceptions, and is usually hidden and locked to assure its secrecy. Diaries describe the past. The information in one’s genetic code can be thought of as a coded probabilistic future diary because it describes an important part of a unique and personal future.

Genetic information is powerful and personal. As the Genetic Code is deciphered, genetic analysis of DNA will tell us more and more about a person’s likely future, particularly in terms of physical and mental well-being. The search for genetic information often involves locating predictors of undesirable and stigmatizing conditions—such as cancers, and conditions that lead to mental illness and dementia. This information is uniquely sensitive for a number of reasons. First, unlike ordinary diaries that are created by the writer, the information contained in the genetic code is largely unknown to the person in whose genetic material it is found. Therefore, if this information is obtained by someone else without the individual’s permission, another person would learn intimate details of the individual’s likely future life. A stranger could, in effect, read the future diary of an individual without the individual even knowing that the diary exists. There are many people, including insurers and employers, to whom information about an individual’s likely health future would be useful.

Second, deciphering an individual’s genetic code also provides the reader of that code with probabilistic health information about that individual’s family, especially parents, siblings and children. Third, since the DNA molecule is stable, once removed from a person’s body and stored, it can become the source of an increasing amount of information as more is learned about how to read the genetic code. Finally, genetic information (and misinformation) has been used by governments to viciously discriminate against those perceived as genetically unfit.

DNA Databases

We originally proposed drafting legislation to regulate DNA databases. We thought of DNA databases as entities that collected, stored, analyzed and controlled DNA samples and information derived from DNA samples, although the term could also include entities that either only stored DNA samples or only stored information derived from genetic analysis. We thought of such databases as holders of genetic information, like computerized medical records, James Watson has said, “The idea that there will be a huge database of genetic information on millions of people is repulsive.”

Dr. Watson’s statement expresses the concern of many people who distrust both computer technology and large, bureaucratic record-keeping systems, and perceive private genetic information as uniquely personal. Such distrust also flows from the realization that current confidentiality policies and practices, which supposedly safeguard personal medical information, are inadequate to protect private genetic information. New rules for DNA databases are needed to minimize the potential harm to individual privacy and liberty that the collection, storage and dissemination of genetic information could produce, and to foster personally and socially useful applications of genetic information. As the U.S. House of Representatives Committee on Government Operations rightly concluded in its study of genetic information, such rules will be more effective and less expensive to implement if established in advance.

Our own analysis of the privacy issues implicated by DNA databases has persuaded us that it is not feasible to protect genetic privacy by limiting regulation to places called DNA databases. One reason is that it is difficult even to define precisely a DNA database. Entities that only store medical records seem to qualify, but are not the major focus of concern regarding the new genetics. There are already many entities that store genetic materials, including the FBI and individual state programs that store DNA samples from convicted sex offenders and other criminals, the U.S. Army’s DNA sample storage program, and private medical research projects. The FBI is primarily interested in using DNA to identify criminal suspects, while medical research programs might conduct future analysis of DNA samples to further decipher the genetic code. Other entities could qualify as DNA banks because they collect and store large amounts of biological material, even though they have no current intent to conduct genetic analysis. Such programs include the Red Cross and other blood banks, private sperm, ovum and embryo banks, and state facilities that store blood samples that have been used for phenylketonuria (PKU) testing.

Collection, Analysis and Storage of DNA and Genetic Information

Focusing solely on any or all of these types of DNA databases assumes that the DNA samples have been legitimately obtained and analyzed, and the only issues are the proper storage of genetic information, and rules governing the disclosure of the genetic information by DNA databanks. But meaningful privacy protection must regulate the collection, analysis and storage of DNA samples, as well as the storage and disclosure of the genetic information derived from the analysis of these samples, no matter who performs that analysis. It is, after all, the DNA samples that contain the individual’s private genetic information. Control of these samples enables the custodian to analyze and reanalyze them to derive increasing amounts of genetic information as new tests are developed. It is also possible to obtain biological material for the purpose of DNA analysis without the person knowing that such material was obtained or analyzed. For example, DNA can even be obtained from hair samples left on a barber’s floor or from saliva found on a licked stamp.

Therefore, to effectively protect genetic privacy unauthorized collection and analysis of individually identifiable DNA must be prohibited. As a result, the overarching premise of the Act is that no stranger should have or control identifiable DNA samples or genetic information about an individual unless that individual specifically authorizes the collection of DNA samples for the purpose of genetic research, authorizes the creation of that private information, and has access to and control over the dissemination of that information.

The rules protecting genetic privacy must be clear and known to the medical, scientific, business and law enforcement communities and the public. The purpose of the Genetic Privacy Act is to codify these rules. It has been drafted as a federal statute to provide uniformity across state lines. However, the Act could be adopted by individual states and used as guidelines by professional societies, at least until such time as Congress acts.

Under the Act, each person who collects a DNA sample (e.g., blood, saliva, hair or other tissue) for the purpose of performing genetic analysis is required to:

- provide specific information verbally prior to collection of the DNA sample;
- provide a notice of rights and assurances prior to the collection of the DNA sample;
- obtain written authorization which contains required information;
- restrict access to DNA samples to persons authorized by the sample source;
- abide by a sample source’s instructions regarding the maintenance and destruction of DNA samples.

Special rules regarding the collection of DNA samples for genetic analysis are set forth for minors, incompetent persons, pregnant women, and embryos. DNA samples may be collected and analyzed for identification for law enforcement purposes if authorized by state law, and for identifying dead bodies, without complying with the authorization provisions of the Act. Research on individually identifiable DNA samples is prohibited unless the sample source has authorized such research use, and research on nonidentifiable samples is permitted if this has not been prohibited by the sample source. Pediatric research and research involving DNA from minors are also governed by specific provisions of the Act.

Individuals are prohibited from analyzing DNA samples unless they have verified that written authorization for the analysis has been given by the sample source or the sample source’s representative. The sample source has the right to:

- determine who may collect and analyze DNA;
- determine the purposes for which a DNA sample can be analyzed;
- know what information can reasonably be expected to be derived from the genetic analysis;
- order the destruction of DNA samples;
- delegate authority to another individual to order the destruction of the DNA sample after death;
- refuse to permit the use of the DNA sample for research or commercial activities; and
- inspect and obtain copies of records containing information derived from genetic analysis of the DNA sample.

written summary of these principles and other requirements under the Act must be supplied to the sample source by the person who collects the DNA sample. The Act requires that the person who receives speculative genetic information in the ordinary course of business keep such information confidential and prohibits the disclosure of private genetic information unless the sample source has authorized the disclosure in writing or the disclosure is limited to access by specified researchers for compiling data.

The Genetic Privacy Act protects individual privacy while permitting medical uses of genetic analysis, legitimate research in genetics, and genetic analysis for identification purposes.

Acknowledgements

This project had its genesis at a meeting in Cold Spring Harbor in November 1989 at which one of the drafters (GIA) gave a presentation on the privacy issues involved in DNA banking. Fourteen months later, he and Dr. Sherman Elias co-hosted an NIH-sponsored workshop in Bethesda, Maryland the purpose of which was to suggest a prioritized research agenda for the Ethical, Legal & Social Implications (ELSI) program of the Human Genome Project. Protecting genetic privacy was ranked as one of the two highest priority issues at that workshop (regulating the introduction of new genetic tests into clinical practice was ranked slightly higher). Shortly thereafter the Director of the ELSI program for the U.S. Department of Energy, Michael Yealy, asked us to draft guidelines to protect the privacy of individuals whose DNA was stored at DNA banks. We agreed, and began this project in June of 1993. With Dr. Daniel Drell of the U.S. Department of Energy (Health Effects and Life Sciences Research Division, Office of Health and Environmental Research, Office of Energy Research) as the project monitor.

In the course of the first year of research we concluded that it was necessary to broaden the scope of the project, and presented the rationale for this change to the ELSI Working Group in June of 1994. They concurred. The first draft of the Genetic Privacy Act was completed in late September 1994, and presented to the ELSI Working Group in December 1994.

Many people, in addition to the members of the ELSI Working Group, contributed in substantial ways to the final product. These included our research assistants, Nan Elster, Sue Yeu, Chris Hager, and Alex Klickstein, as well as our support staff, especially the Administrative Coordinator of the Health Law Department, Marilyn Ricciardelli, and the Department's Secretary, Deborah Darling. The Director of the Boston University School of Public Health, Dr. Robert F. Moen, was especially supportive of our work. We are grateful for the generous and thoughtful comments of our colleagues who reviewed drafts and provided needed insight to both legal and genetic issues. Sherman Elias was our primary genetics consultant, and his advice was invaluable. Robert Gellman's thoughtful comments and advice helped us to avoid many legislative drafting pitfalls. Lori Andrews worked especially hard to make sure we had taken all of the genetic privacy issues into account.

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