HC 70 A

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Learning Unit # 8

Genetics & The Law

This is 1.25 hr lecture

THEMES / CONCEPTS

1 hr lecture 3/11/03

+

1 hr lecture 3/13/03

1. How is genetic information used?
2. Is genetic information unique & given special status?
3. Federal & State Genetic legislation?
4. What are the issues with respect to Genetic Testing?
5. What agencies regulate genetic testing?
6. What legal issues are there with respect to Genetic testing?
7. Newborn Screening?
8. Adult Testing? Child Testing?
9. Family Planning Issues? Stop.
10. Privacy Issues?
11. Employment & Insurance Issues?
12. Legal & Judicial Issues in law enforcement
13. Genetics in other legal areas/laws?

WHAT is Genetic Information Used for?

1) Medical Uses

a. Carrier screening
b. pre-natal diagnostic testing
c. new born screening
d. presymptomatic testing for predicting adult-onset disorders such as Huntington's disease
e. presymptomatic testing for estimating risk of developing adult-onset cancers + Alzheimer's disease
f. Confirmational diagnosis of a symptomatic person

future:
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 / Guilt)
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 expentancy 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 & sorting out environmental components IF tests to be predictive!

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, 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 of embryo cells –
7. CAN be regarded as "Unique" by third parties who might attempt to misuse it!

Should Genetic Information be Protected Separately?

1. Recent legislation suggests yes – genetic information needs to be protected specifically – unique & predictive ∴ distinct from normal medical records
2. Contrary Argument – not possible to separate from clinical records; genetic data similar to other medical information –

Genetics Privacy and Legislation

Quick Links for this page:

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 8, 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 and health esearchers access to 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
Hearing on Potential for Discrimination in Health Insurance Based on Predictive Genetic Tests, July 11, 2001

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 worker 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

- IL: 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, Life, and Disability Insurance in Kentucky (January 2000)
- MI: Report of the Michigan Commission on Genetic Privacy and Progress (February 1999)
- NE: Report of the Nebraska Commission on Human Genetic Technologies Commission (December 1998)
- NY: Genetics Testing and Screening in the Age of Genomics Medicine (November 2000)
- WA: Genetic Privacy, Discrimination, and Research in Washington State (October 2002)
- WA: Washington State Genetics Education Plan (1997)
- 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.

- 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 against discrimination based on 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.
- Doesn't 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 comprehensive privacy legislation (as required 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 the minimum needed for the intended purpose; establish new criminal and civil sanctions for improper use or disclosure; and establish new 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 (ELSI) of human genome research, in a 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 a 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 or contractor 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

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 requesting or 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 HIPPA 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 Genetics 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 rules 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

Science 291, 1249-1250 (2001)

POLICY ISSUES:

Political Issues in the Genome Era

James M. Jeffords and Tom Daschle*

(1)

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 so set aside approximately 5% of the budget, starting in 1990, to fund the ELSI program (Ethical, Legal, and Social Implications of Human Genetics 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 Gallup poll by the Institute for Health Freedom released this past September revealed that 86% of U.S. adults 18 years of age or older 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" (3). 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 all 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 (7). Former President Clinton recently signed an executive order barring genetic discrimination against employees in federal executive departments and agencies (8). 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" (9).

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 (10). But it does not cover people who buy insurance as individuals, nor limit collection and disclosure of genetic information by insurers.

Most protections, whether in terms of employment or health insurance discrimination, are at the state level. At present, 37 U.S. states have laws regarding genetic discrimination and health insurance; 24 states have laws regarding genetic discrimination and employment. Although this patchwork of state laws affords some protections, it also contains loopholes. 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 within 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 (11).

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 banks for use in criminal courts as well as commercial DNA banks. 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 researchers. 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 trying to create a similar genetic database and also to address concerns regarding access (13). Their goal is to include 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

(6)

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 Nondiscrimination 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 also 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, employee and employer groups, and insurers.

References and Notes

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A Proposed Federal Law

THE GENETIC PRIVACY ACT AND COMMENTARY

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PART I - - EFFECTIVE DATE; APPLICABILITY; AND RELATIONSHIP TO OTHER LAWS

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."[1] 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.[2]

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 Databanks

We originally proposed drafting legislation to regulate DNA databanks. We thought of DNA databanks 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.[3] Thinking of such databanks as holders of genetic information, like computerized medical records, James Watson has said, "The idea that there will be a huge databank of genetic information on millions of people is repulsive."[4]

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.[5] New rules for DNA databanks are needed to minimize the potential harm to individual privacy and liberty that the collection, storage and distribution of genomic information could produce, and to foster personally and societally 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."[6]

Our own analysis of the privacy issues implicated by DNA databanks has persuaded us that it is not feasible to protect genetic privacy by limiting regulation to places called DNA databanks. One reason is that it is difficult even to define precisely a DNA databank. 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 databanks 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 analysis, 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.[7]

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. Pedigree 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.

A 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 holds private 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 (GJA) 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 Yesley, 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 Engergy 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. Meenan, 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.

Others who provided valuable comments and input include Wendy Mariner, Michael Grodin, Philip Reilly, Jean McEwen, Wendy Parmet, Bernard Dickens, Margaret Somerville, Alan Westin, Judy Garber and Margaret Dreyfus. The final product, of course, is our responsibility.

George J. Annas
Leonard H. Glantz
Patricia Roche

Boston
February, 1995

Proceed to next section.

File posted May 1995.

Return to Home Page.

New Jersey outlaws genetic discrimination

Washington. The New Jersey state legislature last week gave near-unanimous approval to the most sweeping bill outlawing genetic discrimination yet passed in any of the 50 states.

But a controversial clause giving an individual property rights to genetic information was dropped after pressure from the biotechnology and pharmaceutical industries that are well represented in the state.

The bill is expected to be signed into law by New Jersey Governor Christine Todd Whitman (Republican) this week. It not only outlaws the use of genetic information to deny individuals jobs or health insurance, but also restricts how life and disability insurers may use such information.

In its revised form, the bill has received the backing of a wide range of interest groups, including the two industries involved, labour unions and the Roman Catholic church. The only obvious dissent has come from insurance groups, which testified against the bill last spring. They argue that insurers need all the information possible about applicants to accurately assess risk and avoid driving up rates on individual policies. They are said to have been "reluctantly coerced" into supporting the legislation.

Whitman had vetoed the bill in September, after both houses of the legislature had passed it unanimously (see *Nature* **383**, 367; 1996). Her position reflected the concerns of the biotechnology and pharmaceutical industries, which had objected to a statement in the bill declaring genetic information to be an individual's private property.

The governor and the industries argued that this could have a 'chilling' effect on research, by exposing companies to law-suits for royalties by those whose DNA had been used to develop new products.

The property right declaration was subsequently removed from the bill. Supporters of the clause say that the political power of the pharmaceutical and biotechnology industries left them with little choice — but that the issue was a relatively minor concern when compared with the bill's broad anti-discrimination provisions.

Even in its modified form, the bill is "absolutely more far-reaching than any other", says Generosa Grana, a breast cancer specialist at Cooper Hospital in Camden, New Jersey, and an adviser to the New Jersey Cancer Commission, who helped draft the bill.

None of the advocacy groups fought Whitman's demanded change "because there was so much [else] to lose", adds Karen Rothenberg, director of the Law and Health Care Program at the University of Maryland School of Law, and an expert on state genetic discrimination laws.

Not everyone agrees. George Annas, a lawyer and professor of public health at Boston University School of Public Health, says that "gutting" the property right clause has turned the bill into "an anti-genetic privacy act". He called it "bizarre" that "other people can own your genetic information, but you can't". And State senator Robert Martin (Republican), a law professor who was the lone Senate opponent of the revised bill, argues that Whitman's concern to protect industry may not have given enough protection to ordinary citizens.

The strength of the bill lies in its prohibition of discrimination not only on the basis of genetic tests, but of genetic information — a far broader term which includes family history, and can include individual history, physical examination and the results of other tests.

The bill is also broad in scope. It imposes restraints on life and disability insurers, in addition to employers and health insurers. Laws in other states have been narrower.

Under the bill, neither genetic information, nor an individual's refusal to submit to a genetic test or provide test results, can be used in decisions on hiring, firing and health insurance. Life and disability insurers may demand and use genetic information in underwriting, but must not use it 'unfairly'.

A life insurer, for example, could not use the fact that a woman is carrying a *BRCA1* mutation to decide whether to issue a policy, or what rate to charge, because this fact is no guarantee that she will develop cancer.

But the same insurer could legally refuse cover or charge higher premiums to somebody who carries the gene for Huntington's disease, as that person has a 100 per cent chance of developing the disease. In such a case, the insurer would have to base rates on actuarial data for Huntington's patients.

Only one state — Oregon — of the 12 others that have passed laws dealing with genetic discrimination includes a property right. An official now implementing the Oregon law says that the property right does not seem to have had any immediate impact. Michael Skeels, director of the state's Public Health Laboratory, adds that its implications for research will "take years" to become clear.

Earlier this year, the US Congress passed a law merely forbidding health insurers from using genetic information to discriminate against people who change or lose jobs. Pressure is growing for a broader federal law, and the issue may be addressed in the next legislative session. **Meredith Wadman**

Gene tests 'need research protocols'

Washington. An advisory committee to the National Institutes of Health (NIH) has recommended that genetic testing for breast and ovarian cancer be conducted only within strictly defined research protocols. This reverses an earlier position encouraging wider use of testing (see *Nature*, **380**, 573; 1996).

Last week, the Advisory Committee on Research on Women's Health unanimously passed a resolution urging that genetic tests for breast and ovarian cancer be conducted only within "hypothesis-driven protocol studies" endorsed by NIH-approved institutional review bodies.

Typical studies, says the resolution, might address questions such as the positive predictive value of tests, and the appropriate medical management of those carrying mutations. The advice represents a refusal to endorse commercial genetic testing that does not incorporate hypothesis-driven research.

Last April the committee refrained from calling for testing to be confined to research protocols. One dissenter at the time was Linda Burhansstipanov, director of the Native American Cancer Research Program at the AMC Cancer Research Center in Denver, Colorado, who called the resolution "paternalistic". But last week she supported the revised resolution, after the committee added a new, lengthy preamble. It includes a call for research on how poor, non-white and rural women can be guaranteed access to testing under research protocols.

Vivian W. Pinn, director of the NIH's Office of Research in Women's Health, says she agrees with the advisory committe. Access to genetics testing is important, but women "should know what it means", and such information is more likely to be both gathered and imparted in the research setting.

In adopting its position, the advisory committee joins the American Society of Human Genetics, the Advisory Council of the National Center for Human Genome Research, and the National Breast Cancer Coalition. In contrast, the American Society of Clinical Oncology has called for genetic testing to be made available outside research settings "as part of the preventive oncologic care of families".

The new recommendation comes two weeks after Myriad Genetics of Salt Lake City introduced commercial full-sequence testing of *BRCA1* and *BRCA2* genes, mutations which can confer a predisposition to breast and ovarian cancers. The company is charging $2,400 for initial testing, and $395 for tests of additional family members. **M.W.**

From Learning Unit #7

Genetic Testing Issues

1. **Privacy?**
   - Who should know – spouse, children, employer, insurance company? Tell if carrier?
   - Who should tell – person, Dr., testing Lab.
   - Results when insurers know/employers know?
2. **Voluntary vs. Mandatory Testing?**
   - What is precedent? State Laws?
   - Results – Sickle cell / Cystic Fibrosis PKU?
   - Test for any reason – Information? Eugenics? Forensics?
3. **Regulation?**
   - Who monitors tests? Over-the-counter
   - Who licenses testing labs & 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 "interim" if testing available? Community Rating? Universal Health Insurance.
5. **Penetrance/Expressivity Problem?**
   - What to do if disease not manifested 100%? How deal with information? Insurance issues?

Genetic Testing Issues Continued

(6) What Laws govern Prenatal Rights to Test & Perhaps use BABI 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 disease? Will carry gene 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 Protect us From Genetic Discrimination? Any? Americans w Disabilities Act. Carriers? Executive order 2000 / Clinton

(13) What about testing for Enhancement / Eugenics?

Culturally Related?

What Agencies & Laws Regulate Genetic Testing Products & Services?

Police Powers – "promote the general welfare"

(1) Test Products

Example: PATHWAY® – Her2 test for breast cancer

- The FDA regulates genetic testing kits, reagents, & machinery under the Medical Devices Act of 1976 & the Safe Medical Device Amendments of 1990.
- Intent is to provide safety & effectiveness – e.g., give proper results & distinguish between high risk, low risk, & false positives
- Premarket Approval (PMA) is required of all Medical devices Marketed for in vitro diagnosis

(2) Laboratory Services

- Clinical Laboratory Improvement Act of 1967 (CLIA67) –
- Department of Human & Health Services (HHS)
- Clinical Laboratory Improvement Amendments of 1988 (CLIA88)

Materials from human body for purposes of diagnosis, prevention, or treatment of disease

WHAT LEGAL ISSUES ARE THERE in Genetic Testing?

1. Health Care & Reproduction?
2. Workplace?
3. Insurance?
4. Law Enforcement & Judicial Applications?

Legal Issues in Genetic Testing
J.A. Robertson
in
The Genome, Ethics, & the Law

MEDICAL CARE & HEALTH PROMOTION

NEWBORN SCREENING

1960's
Robert Guthrie
PKU screening test

(1) LAWS in MOST States REQUIRE **Mandatory** newborn screening IF disease CAN BE TREATED in newborn baby to Prevent Disease or Mitigate More Serious Aspects of Disease — MANDATORY PUBLIC HEALTH APPROACH!

(2) PKU, Galactosemia, Primary Congenital Hypothyroidism, Sickle Cell and other Hemoglobin diseases (California Newborn Screening Program).

(3) Only Legal ground for refusal is religious beliefs & practices in CA. Ethical?!

(4) New Mass Spectroscopy Screening in CA and other States have added 20 more tests for metabolic disorders voluntarily in a trial research period.

(5) State pays although fee can be charged.

MANDATORY testing! ⇐

Laws Governing
- **Police Powers**
  "promote general welfare"
- **"Parens patriae"**
  authority to intervene to prevent person at risk from self-inflicting harm

(6) If Risks/Benefits of TREATMENT NOT clear, then CAN'T be MANDATORY & Left to PARENTAL DISCRETION — OR NO TREATMENT

(16)

Genetic Technologies Project
Health Care Program

Newborn Genetic and Metabolic Disease Screening

Updated January 2003

Source: kidshealth.org

State newborn screening systems were the first and remain the largest genetic programs for children. Nationwide, state public health programs screen an estimated 4 million infants annually for genetic disorders. Undetected and untreated abnormalities can result in severe problems, mental retardation or even death. Although funding newborn screening programs requires expenditures by the states, proactively treating congenital abnormalities may save states money by avoiding more financially burdensome medical costs and state institutional services. Comprehensive state newborn screening programs involve testing, follow-up, diagnosis, treatment and evaluation.

Even though newborn screening became available to infants through state programs in the 1960s and all states screen for some conditions, the extent of screening varies throughout the states. Some 700 genetic tests are available; however, not all the tests are recommended. For example, some conditions are so rare, testing is not cost-effective; in other cases no treatment exists for the conditions. State experiences vary regarding laws or regulations, specific tests, oversight responsibilities, state advisory boards, processes for informing parents, exemptions, storage policies and use of blood samples and payment for newborn screen procedures.

A Newborn Screening Task Force, co-sponsored by the American Academy of Pediatrics and the Maternal and Child Health Bureau, made a series of recommendations with regard to state newborn screening programs. Their report calls for states to: use a comprehensive systems approach; follow accepted national guidelines; coordinate infant programs and data; pilot new tests and technologies before adopting major policy changes and new mandates; monitor and evaluate program performance; involve and inform families; establish a state advisory group that has a diverse representation; set state-level policies for the use and storage of residual newborn screening blood samples; and assure adequate financing for a whole system using state newborn screening fees and other funds.

State laws on genetic screening relate to diseases and disorders such as adrenal hyperplasia, biotinidase deficiency, branched-chain ketonuria, cystic fibrosis, galactosemia, homocystinuria, hypothyroidism, maple syrup urine disease, phenylketonuria (PKU) and sickle cell anemia. Many state laws include exemptions for parents who object to genetic testing for religious or other reasons. During the 2002 legislative session, at least three states--**Mississippi**, **Nebraska** and **Virginia**--enacted laws related to newborn genetic screening. Other states have created laws related to newborn screening privacy issues.

California

Cal. Health & Safety Code € 1374.56 and Insurance Code € 10123.89 (1999) requires health plans to offer coverage for the testing and treatment of PKU. [Cal. Stats., Chap. 541 (SB 148)]

Cal. Health & Safety Code € 125000 and 125001 (1998) requires the Department of Health Services to establish a program to detect PKU and other preventable heritable or congenital disorders. The law requires the department to establish a genetic disease unit to promote a statewide program of information, testing, and counseling services. The law directs the department to charge a fee for tests. The law does not apply if a parent or guardian of the newborn objects to a test on religious grounds. [Cal. Stats., Chap. 1011 (S 537)]

Newborn Genetic Screening Privacy Laws

Health Programs

Updated July 2002

Currently, **28 states** require consent to either perform or require genetic testing or to obtain, retain or disclose genetic information through genetic-specific privacy laws. In addition, Washington includes genetic information in the definition of protected health information under the state's health privacy statute. Many of the states with genetic privacy laws exempt newborn screening from consent provisions, including Delaware, Illinois, Louisiana, Massachusetts, Michigan, Nevada, New Hampshire, New Jersey, New Mexico, New York, Oregon and Vermont. The chart below does not address consent requirements or exemptions for newborn screening that may be found in state administrative codes.

At least **23 states** have laws that allow for an exemption to the newborn genetic screening requirements if parents object on religious grounds (Alabama, Arkansas, California, Colorado, Connecticut, Delaware, Georgia, Illinois, Indiana, Kentucky, Louisiana, Massachusetts, New Jersey, New York, North Dakota, Ohio, Rhode Island, South Carolina, Texas, Utah, Virginia, Washington and Wisconsin). Two states--Florida and Wyoming--allow for an exemption to the newborn genetic screening requirements if parents object on any grounds.

At least **12 states** have confidentiality requirements related to newborn screening laws (Arizona, Colorado, Florida, Hawaii, Iowa, Louisiana, New Jersey, North Dakota, Ohio, South Carolina, Virginia and Wisconsin).

At least **six states** and the District of Columbia have laws related to obtaining consent from the parents of children before performing genetic tests (Hawaii, Ohio, Nebraska, Texas, Wisconsin and Wyoming). Kansas requires informed consent in order to monitor infants with genetic disorders.

Many states have laws regulating newborn hearing screening, but these laws do not necessarily apply to newborn genetic screening.

State	Newborn Genetic Screening Privacy Laws	Law Allows for a Religious Exemption	Genetic Privacy Law Allows for an Exemption for Newborn Screening
California	Cal. Health & Safety Code § 124975 clarifies that participation of people in hereditary disorders programs should be wholly voluntary, except for initial screening for phenylketonuria (PKU) and other genetic disorders treatable through the California newborn screening program. All information obtained from people involved in hereditary disorders programs in the state should be held strictly confidential. Cal. Health & Safety Code § 124980 prohibits tests from being performed on any minor over the objection of the minor's parents or guardian. Tests may not be performed unless the parent or guardian is fully informed of the purposes of testing for hereditary disorders and is given reasonable opportunity to object to the testing. No testing, except initial screening for phenylketonuria (PKU) and other diseases that may be added to the newborn screening program, shall require mandatory participation. The law requires all testing results and personal information generated from hereditary disorders programs to be made available to individuals over 18 years of age, or to the individual's parent or guardian. All testing results and personal information from hereditary disorders programs shall be held confidential and be considered a confidential medical record except for information that the individual, parent, or guardian consents to be released.	Cal. Health & Safety Code § 125000	

The California Newborn Screening Program

The NBS Program has several mechanisms in place to ensure testing of all babies born in California. State NBS Regulations specify reporting requirements for both licensed perinatal health facilities and county registrars to ensure testing. All newborns must be tested; **the only legal ground for refusal** is a conflict with religious beliefs and practices. The following procedures and forms are utilized to ensure testing.

The NAPS laboratories enter demographic data and test results on terminals linked to a Genetic Disease Branch central computer in Berkeley. A computer-generated printed report of all test results, referred to as a "result mailer," is mailed to the hospital where the specimen was collected. Another copy is mailed to the physician of record as reported on the specimen collection form.

Perinatal facilities must review each newborn's medical record within 14 days from the date of discharge to determine that the NBS results are filed in it, or that a parent's or legal guardian's signed refusal is present. If it has been determined that a newborn was not tested, the facility must notify the infant's physician and the NBS Program. If a specimen was collected (as indicated by the presence of the goldenrod copy of the specimen collection form) but there is no NBS Results Mailer in the chart, the facility must complete a Missing Result Form (see below) and submit it to the State within five days. Most often the State has a record of the baby having been tested and a duplicate result mailer is forwarded to the hospital. Occasionally, a baby is not tested or the specimen is lost between the hospital and lab, and these are followed up. It is the responsibility of the pediatrician who provides comprehensive care for the child to ensure that a newborn screening test has been done and that the results have been reviewed and noted in the patient's chart. Pediatric care providers who do not have a copy of the NBS Result Mailer can request a duplicate from the State or regional ASC.

It is essential that the NBS-NO and NBS-OH forms be mailed **promptly** to the State NBS Program. The state follows up on each of these forms to make sure the baby has been tested. Unless there is a record of parent refusal on file, the State refers all untested babies under one year of age to the Newborn Screening Follow-Up Coordinators for assistance in obtaining the test. If you delay in sending us the forms, we are delayed in getting the babies tested, which in turn could delay treatment if a baby has one of the disorders for which the newborn screening panel tests.

Information for County Birth Registrars

County birth registrars are required to notify persons registering the birth of a baby born outside of licensed perinatal health facilities of newborn screening. The birth registrar must provide the person registering the birth with the pamphlet "Important Information for Parents About the Newborn Screening Test" and information about how to have the baby tested. The registrars are also required to notify the NBS Program of these births and must complete and send the NBS-OH form ("Notification of Registration of Birth Which Occurred Out of a Licensed Health Facility") to GDB.

Notification of Registration of Birth Which Occurred Outside of a Licensed Health Facility (NBS-OH)

Used by county birth registrars to report babies born outside of a licensed health facility

Important Information for Parents About the Newborn Screening Test (IIP):

Birth registrars are required to give this pamphlet to the person registering the birth of a baby born outside of a licensed health facility and not admitted to a hospital within 30 days of the birth.

To order these forms, please call (510) 540-3302.

The California Newborn Screening Program

Phenylketonuria (PKU)

Cofactor Variants

Galactosemia

Primary Congenital Hypothyroidism

Sickle Cell Disease and Other Hemoglobinopathies

Thalassemias

Alpha Thalassemia

Beta Thalassemia

The California Newborn Screening Program

In California, the prevalence of:

PKU is 1 in 27,000 births (classical PKU only)

Approximately 15-18 cases are detected annually through the mandated Newborn Screening Program. Over 350 children have been identified with classical PKU since 1980.

Galactosemia is 1 in 73,000 births

Approximately four to eight cases are identified in California every year

Primary congenital hypothyroidism is 1 in 2,700 births

Approximately 200 cases a year are identified in California.

Sickle cell disease is about 1 per 4,400. The Newborn Screening Program detects approximately 125 cases each year.

In addition, Beta thalassemia major and hemoglobin E/Beta thalassemia are detected, occurring in about 1 in 27,000 newborns in the State.

About 5 cases of E/Beta thalassemia, 4 cases of Beta thalassemia major and 1 each of C, D and E/Beta thalassemia are identified annually.

In California the incidence of Hemoglobin H disease is about 1 in 15,000 births, or about 35 to 40 cases per year are detected.

The California Newborn Screening Program

Newborn Screening is recognized nationally as an essential preventive public health measure. All states in the nation and the District of Columbia have established newborn screening programs. The State of California began its Newborn Screening Program in 1966 with the testing for phenylketonuria (PKU). In October 1980, the program was expanded to include galactosemia, primary congenital hypothyroidism, and a more comprehensive follow-up system. In 1990, screening for sickle cell disease was added to the State's existing program. This also allowed for the identification of some of the related non-sickling hemoglobin disorders, including beta^0 thalassemia major, and Hb E-Beta Thalassemia. In 1999, the Program implemented screening for hemoglobin H and hemoglobin H - Constant Spring disease.

Very early detection permits the metabolic disorders PKU and galactosemia to be treated with a diet, and hypothyroidism with thyroid hormones, thus preventing the development of mental retardation and other severe health problems. Detection of sickle cell disease in newborns makes possible early entry into comprehensive care, which includes the initiation penicillin prophylaxis and parent education (e.g., identification of early warning signs and preventive health measures), factors which have been shown to reduce morbidity and mortality. Early detection of thalassemia disorders allows for close monitoring for infections and anemia. Ongoing health care and close monitoring help children with hemoglobin disorders stay as healthy as possible.

Informing Parents of the Test

State regulations (17 CCR 6500) require that prenatal care providers give pregnant women informational material about the newborn screening program. Because some women do not receive prenatal care, the same informational material, **Important Information for Parents about the Newborn Screening Test (IIP)**, is also distributed upon admission to a licensed perinatal health facility for delivery. The State Newborn Screening Program supplies copies of this pamphlet at no cost to all health professionals who serve maternity patients, to hospitals that provide maternity and/or newborn care, to local health departments, and county birth registrars.

How to Order IIP

Benefits of the Newborn Screening Program

The program screened 10,065,506 babies from October 1980 to June 2000 and identified the following disorders:

Disorder	Cases
PKU	358
Primary Congenital Hypothyroidism	3,236
Galactosemia	132
Sickle Cell Disease* and other clinically significant Hemoglobinopathies* (Beta^0 Thal Major, E-Beta Thal, etc.)	1,229
Hemoglobin H Disease**	91
Total	**5,046**

* from 2/27/90
** from 7/96

Based on the known occurrence rates of these disorders, the number of diagnosed cases has been within the expected frequency rate. Efficient processing of test results and program monitoring have resulted in the initiation of treatment of these babies at a very early age.

Median Age of Treatment

1980 - 2000

Disorder	Age (days)
PKU	10
Galactosemia	5
Primary Congenital Hypothyroidism	9
Sickle Cell Disease*	55

The Original Four Genetic Diseases Screened For

The California Newborn Screening Program

High blood phenylalanine levels are indicative of one of the following categories of disorders in its metabolic pathway: classical PKU, hyperphenylalaninemia, and co-factor variant defect.

Classical PKUis an inherited recessive autosomal disorder (chromosome 12) with an incidence of **1:27,000** in California (1:15,000 in Caucasians, less common in other races). California's lower birth prevalence is due to the preponderance of non-Caucasian births. Since 1966, when PKU screening began, more than 500 cases have been detected.

PKU

The disorder is due to a lack of phenylalanine hydroxylase; this is an enzyme needed to metabolize the amino acid phenylalanine to tyrosine (another amino acid); tyrosine is a precursor for such important biochemical products as serotonin, catecholamines, thyroid hormone, and melanin. This enzyme deficiency leads to high levels of phenylalanine and low levels of tyrosine, causing:

- mental retardation
- seizures
- decreased growth rate
- poor motor skills
- hypopigmentation

Dietary restriction of phenylalanine (phe), begun within the first few weeks of life, will result in normal development. This is accomplished by replacing most dietary protein with a supplementary formula containing adequate amounts of essential amino acids other than phenylalanine. Phenylalanine can be found in all foods containing protein. By eliminating overtly proteinaceous foods, aspartame (NutraSweetÓ), and wheat products containing gluten, blood phe levels can be significantly reduced. Since phenylalanine is an essential amino acid, it should not be totally omitted from the diet: too low a phe level is not healthy, either. The recommended phe-level range is 120 to 360 mmol/L (2 to 6 mg/dl) for children.

Frequent monitoring of the blood phe level and adjustment of the diet is necessary to ensure both adequate nutrition and safe levels of phenylalanine. This strict diet should be followed indefinitely rather than discontinuing it at eight or nine years of age (which was the standard in the past). The current recommendation of diet for life was developed based on studies which indicated that maintaining low phe levels seemed to result in individuals with PKU being able to concentrate better, do better in school, are able to do more complex math problems than when they are not on a low-phe diet.

Severe mental retardation is the rule for individuals with untreated classical PKU. With early adequate treatment, mental retardation is totally preventable. If treatment is delayed for some weeks, the results are more variable. Children who are not treated until after six months of age may show some improvement in IQ, but they will be retarded. Those who are not treated until they are even older usually show little change in IQ, but a phe-restricted diet may help control seizures and/or serious behavioral problems. A mousy or musty odor in older, inadequately treated-individuals is frequently noticed.

What is primary congenital hypothyroidism?

Primary congenital hypothyroidism is an endocrine condition present at birth that occurs when the thyroid gland does not produce enough thyroid hormone to meet the body's needs. Typically, the thyroid gland makes thyroid hormones, such as thyroxine (T4), which are necessary for brain and central nervous system development as well as muscle and bone growth. These hormones help to maintain body temperature and assist with intestinal movements. They also keep the chemical changes which occur in various tissues of the body going at a constant rate.

Hypothyroidism

When primary congenital hypothyroidism occurs, it is usually caused by an undeveloped thyroid gland. The gland is either too small, located in the wrong place, or was never formed. An undeveloped thyroid gland either makes small amounts of thyroid hormone or none at all. If primary congenital hypothyroidism is untreated, it can lead to severe mental retardation and growth retardation. Early identification and treatment of hypothyroidism will prevent severe mental retardation and other health problems.

What are the symptoms?

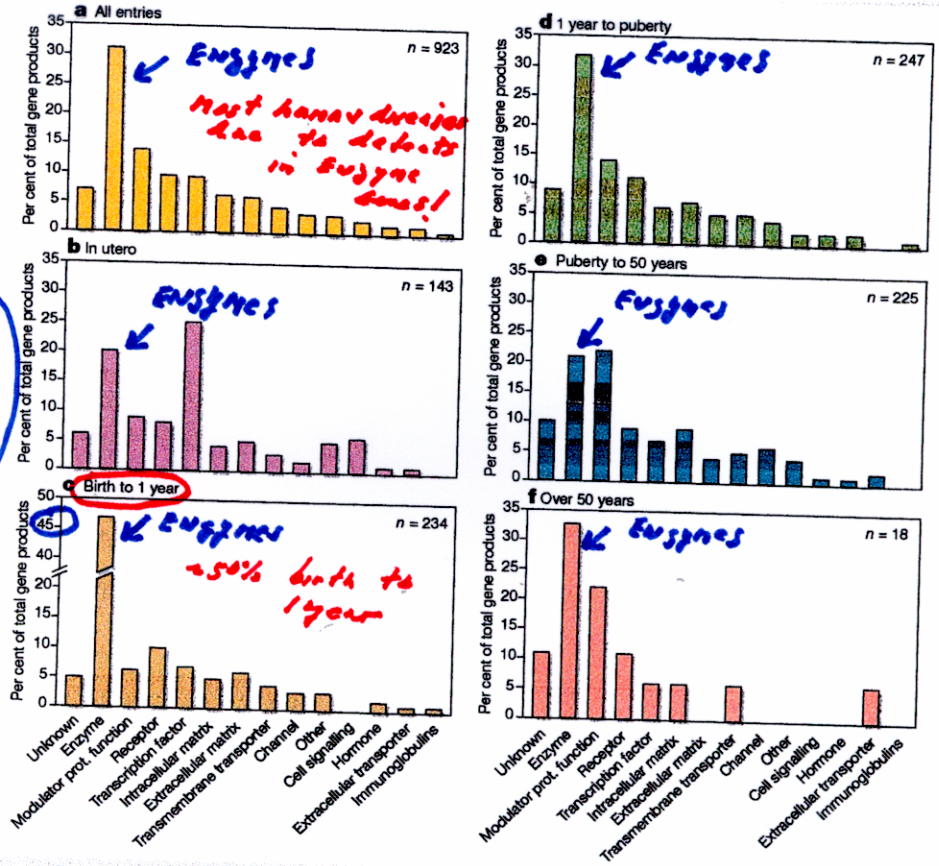
The characteristic features include puffy eyes, thick tongue, coarse facial features, a hoarse cry, skin mottling (spotting of different coloring on the skin), and lethargy (extreme drowsiness or sluggishness).

What is the treatment?

The treatment requires taking a daily pill of thyroid hormone called thyroxine. You should always consult your doctor regarding any treatment recommended.

Recall:
1.2% of live births have gene defects due to mutations!

Human disease genes



Age of Disease Onset

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%; $\chi^2 = 49.4$, $P < 0.001$). 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%; $\chi^2 = 35.8$, $P < 0.001$).

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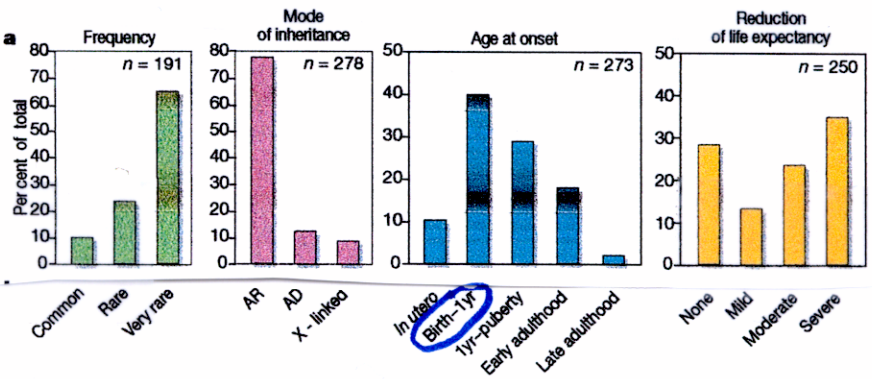
ENZYME ENCODING GENES



Affect metabolism

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.

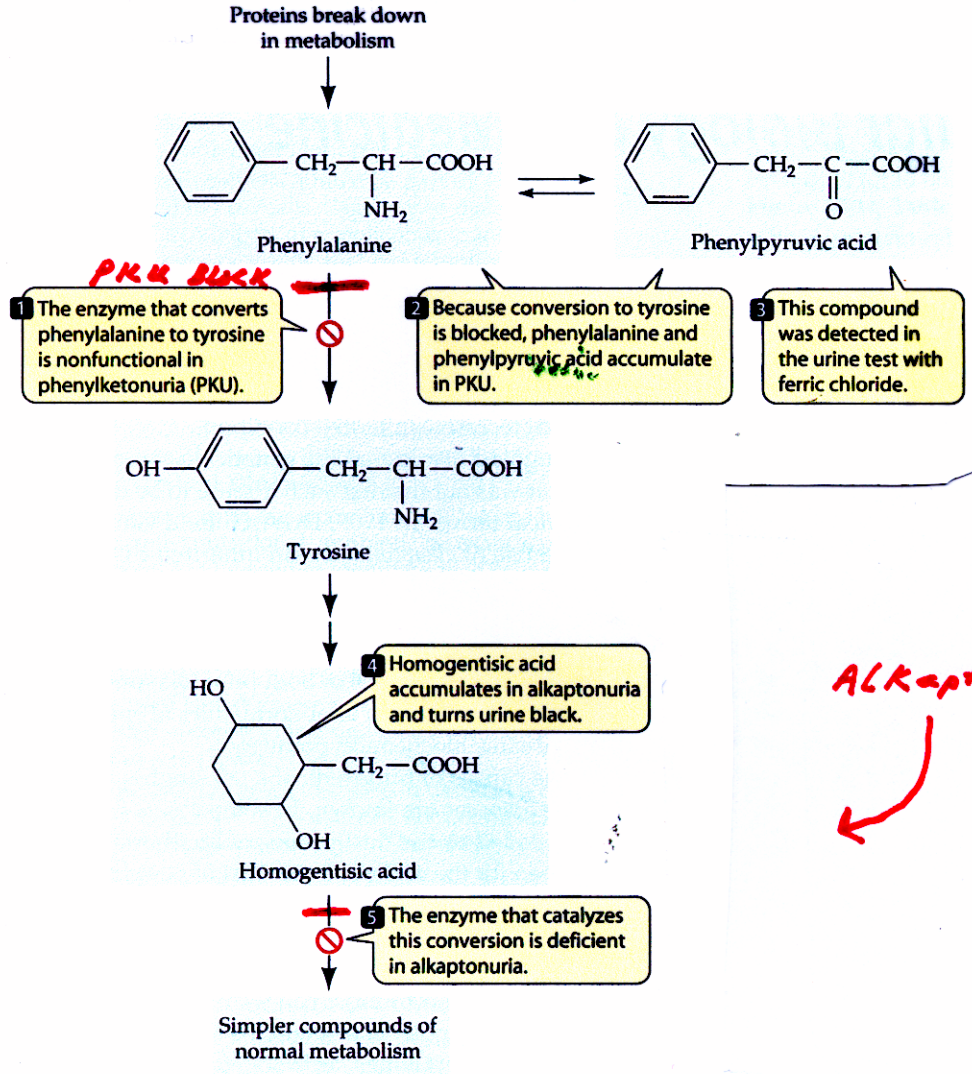
Metabolic Diseases - Inborn Errors of Metabolism

[Phenylalanine] ↑

Block

PKU →

∴ need a no phenylalanine diet to overcome!



Alkaptonuria Block ↓

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.

Mutations in Specific Gene of Pathway Block pathway leading to Accumulation of toxic precursor compounds

INBORN ERRORS of Metabolism

Protein degradation

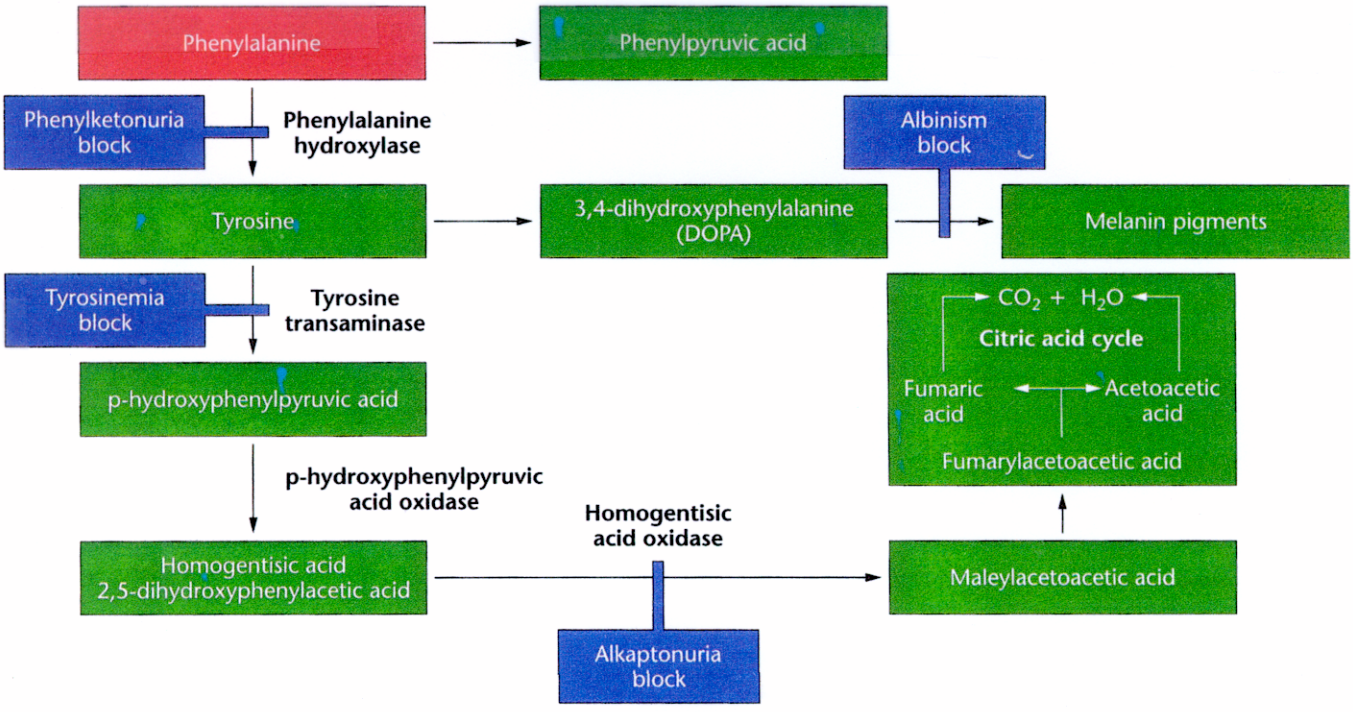
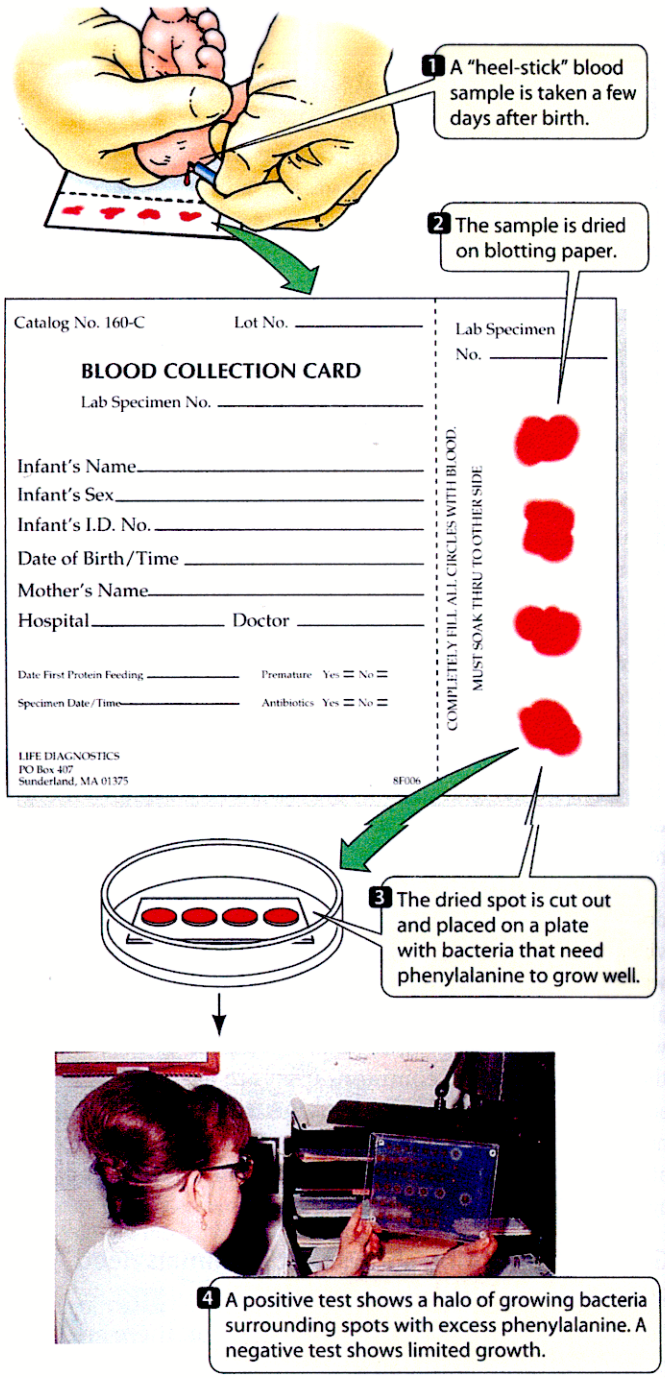


FIGURE 14.10 Metabolic pathway involving phenylalanine and tyrosine. Various metabolic blocks resulting from mutations lead to the disorders phenylketonuria, alkaptonuria, albinism, and tyrosinemia.

Newborn screens for compounds that accumulate before block in metabolic pathway

Screening Newborn Infants For Disease Genes

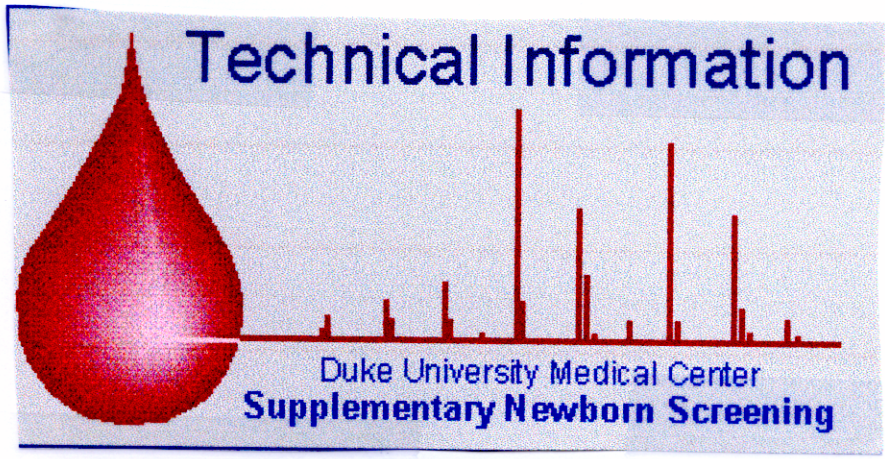


PKU

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.

Old way!

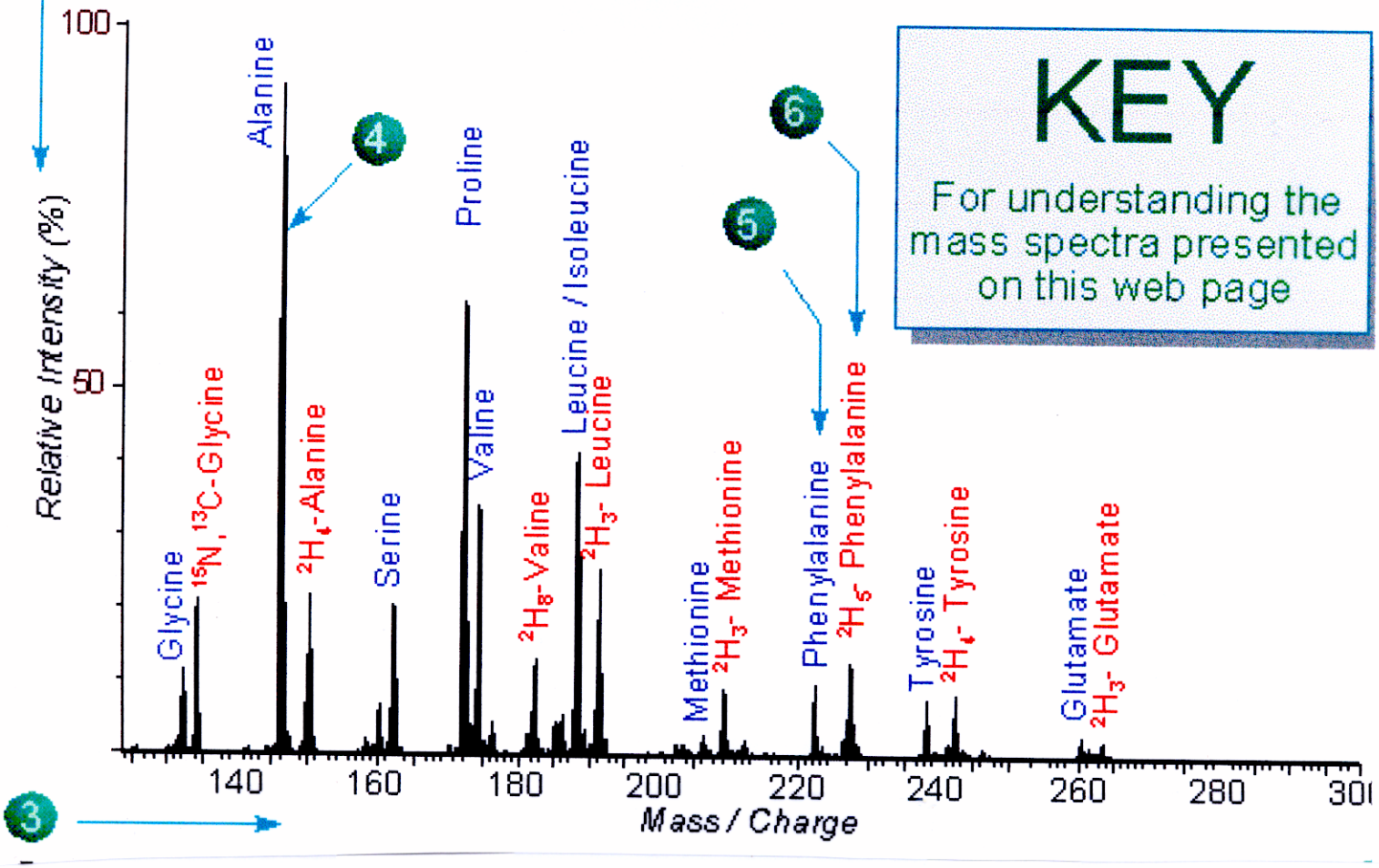
USING TANDEM MASS SPECTRUM TO SIMULTANEOUSLY SCREEN FOR Genes Affecting Metabolic Disorders



New way

1 Neutral Amino Acid Profile

(Tandem Mass Spectrum - Neutral Loss of 102 Daltons)



Detects Elevated Levels of toxic compounds Accumulating in Infancy

CALIFORNIA TANDEM MASS Spectrometry Screening Project

1/7/2001

GENETIC DISEASE BRANCH

SUPPLEMENTAL METABOLIC SCREENING

(Information For Parents)

DISORDERS POTENTIALLY DETECTED

All of the disorders below are **autosomal recessive**, which means that, although usually neither parent is affected, each parent must have passed a gene for the disorder to their baby in order for the baby to be affected. There is a one-in-four chance that this will happen each time the couple has a birth.

(+) Original 4 Tests
PKU
Sickle Cell
Galactosemia
Congenital Hypothyroidism

Amino Acid Disorders

Organic Acid Disorders

Fatty Acid Oxidation Disorders

Amino Acid Disorders

The terms "amino acidemia" and "amino aciduria" refer to disorders in amino acid metabolism (breakdown process to provide energy or heat for body functions). Amino acids are the chemical building blocks of human proteins. Proteins are responsible for the functioning of cells in the body. In order for amino acids to work, specific enzymes must be present. Aminoacidurias are disorders resulting from deficiencies (lack) of enzymes needed for amino acid metabolism or transport. This results in abnormal quantities of amino acids building up in the urine or blood. In large quantities, amino acids can be toxic to the body.

Symptoms in babies will vary by disorder and may include slow development, vomiting, diarrhea, abnormal ordor or color of urine and/or a buildup of acid in the body (acidosis) and can result in mental retardation.

Treatments may include replacement of the deficient enzyme, special diets and medication. Prompt treatment may prevent serious problems from developing.

- Maple syrup urine disease (MSUD)
- Homocystinuria/cystathionine beta-synthetase deficiency (CBS)
- Citrullinemia/argininosuccinic acid synthetase deficiency (ASAS)
- Argininosuccinyl-CoA lyase deficiency (ASAL)
- Phenylketonuria (PKU)
- Argininemia/arginase deficiency
- Tyrosinemia

Organic Acid Disorders

Organic acid disorders can be referred to as organic acidemias or organic acidurias. Organic acids are a group of chemicals that are used in critical metabolic processes of the body. Organic acid disorders usually result from a missing or malfunctioning step in amino acid catabolism (chemical breakdown) due to a lack of enzyme activity.

Symptoms will vary by disorder and may include poor feeding, vomiting, low blood sugar, drowsiness, seizures, brain disease and coma.

Treatment may include a special diet and/or medication to remedy the problems caused by the deficient enzyme activity.

- Propionic acidemia
- Methylmalonic acidemia
- Isobutyryl-CoA dehydrogenase deficiency
- Isovaleric acidemia
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA)
- Glutaric acidemia type-1 (GA-1)
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Beta-ketothiolase deficiency (BKD)

Fatty Acid Oxidation Disorders

Fatty acids are a component of fat in the food we eat and from fat in our tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one of these enzymes is missing.

Symptoms willl vary by disorder and may include drowsiness, poor tone, vomiting, low blood sugar, brain disease, liver failure, and muscle problems -- all of which, without treatment, can lead to severe outcomes such as coma and death.

Treatments include low-fat diets, avoiding fasting, and maintaining a regular intake of sugar, carnitines and other supplements.

- Short chain acyl-CoA dehydrogenase (SCAD) deficiency
- Medium chain acyl-CoA dehydrogenase (MCADD) deficiency
- Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
- 3-Hydroxy long chain acyl-CoA dehydrogenase (LCHAD) deficiency and tri-functional protein deficiency/trifunctional protein deficiency
- Carnitine palmitoyl transferase deficiency - type II (CPT-2)
- Carnitine-acylcarnitine translocase deficiency (CAT)
- Carnitine transporter deficiency
- Multiple acyl-CoA dehydrogenase deficiency (MADD)/glutaric acidemia type-2 (GA-2)
- Carnitine palmitoyl transferase deficiency - type 1 (CPT-1)

Other MS/MS topics:
Voluntary Supplemental Testing
Additional Information and Resources

NEWBORN SCREENING
NEWS

The California Newborn Screening Program Fall 2001

Supplemental Screening for Multiple Metabolic Disorders

MS/MS Research Project

Background

"The introduction of Tandem Mass Spectrometry (MS/MS) in the 1990's for population-based newborn screening has enabled healthcare providers to detect an increased number of metabolic disorders in a single process using dried blood spot specimens routinely collected for newborn screening."[1] MS/MS allows for screening of multiple metabolic disorders using a single analytical run. With this technology there is the potential to test for a wide array of metabolic disorders, including amino acid disorders, organic acidemias, and fatty acid oxidation disorders. Because the technology can detect these disorders (approximately 30 total) within 1 to 2 minutes, the system can handle the large numbers of specimens required in newborn screening. For some of the disorders identifiable via MS/MS, such as medium chain acyl-CoA dehydrogenase deficiency (MCADD), early detection and treatment can result in substantial improvements in health outcomes (i.e., prevention of mortality and improvement of quality of life). Several states have already expanded, or are in the process of expanding, their newborn screening program to add these disorders.

The California Newborn Screening Program, which has been in existence since 1980, currently tests for PKU, galactosemia, primary congenital hypothyroidism, sickle cell disease and other hemoglobinopathies. On September 28, 2000, Governor Gray Davis signed into law Assembly Bill 2427 (Kuehl) which provides for updating and expanding the newborn screening program in California. The law took effect on January 1, 2001. AB 2427 requires the Department of Health Services to investigate the feasibility of establishing a new and broader testing program, including development and evaluation of expanded genetic disease testing utilizing Tandem Mass Spectrometry. In response, the Department plans to expand screening as a part of a research (pilot) project.

The Genetic Disease Branch (GDB) of the DHS has been actively planning for implementation of the project for over a year. Meetings of metabolic and laboratory experts from across the state were held in October, 2000 and May, 2001 to develop recommendations regarding the specific disorders to be included in the initial phase and to discuss the implementation process. The research proposal for the project has been reviewed and approved by the State Health and Human Services Agency's Committee for The Protection of Human Subjects.

This study is being conducted in part to determine which of the disorders identifiable via MS/MS meet the criteria for inclusion in California's mandatory Newborn Screening Program, i.e., which of the unusual results have clinical significance and what warrants reporting. Initially all "interesting" or "unusual" results will be reported to the pediatric care provider, then to a metabolic specialist for evaluation. Treatment and outcome data will also be collected on all newborns referred to Metabolic Centers for follow-up.

The NBS MS/MS Research Project

The actual start date will be announced at least 1 month prior to implementation. The estimated duration of the supplemental testing is 12-18 months. Participation in the study will be voluntary and informed consent will be obtained for both the testing of specimens and for release of medical information for newborns referred to metabolic centers. There will be no additional fee charged

[1] Centers for Disease Control and Prevention. Using Tandem Mass Spectrometry for Metabolic Disease Screening Among Newborns: A Report of the Workgroup—Georgia, 2000. MMWR Morb Mortal Wkly Rep., Recommendations and Reports April 13, 2001:50:1

for participation in the supplemental screening and no additional blood will be needed. National experience to date in MS/MS screening using a full panel of acylcarnitines and amino acid analyses has resulted in a detection rate between 1:4,000 to 1:5,000[2] (Chace et al). Based on the annual California birth rate and the acceptance rate reported by other states that have offered MS/MS supplemental screening, we project about 400,000 newborns participating in the pilot project and detecting an additional 40-60 newborns with clinically significant metabolic disorders not included in the current California mandatory newborn screening program.

MS/MS Research Project Screening Process

Information on the mandatory newborn screening program as well as the supplemental screening via the pilot project will be provided to parents by prenatal care providers and hospital staff. Written verification of informed consent will be obtained by hospitals and birthing centers using a form provided by the State (included in the Newborn Screening Program booklet, entitled *Important Information for Parents About the Newborn Screening Test*).

Specimen collection, handling and transport will occur in the same manner as the current mandatory screening. Hospital staff will complete the demographic information on the newborn screening Test Request Form (TRF), also known as the Newborn Screening Specimen Collection Form. The blood specimen will be collected from the newborn's heel and dropped onto the five (5) blood spots on the filter paper attached to the TRF and allowed to dry. A separate collection form for this project will not be necessary. The hospital staff will indicate whether the newborn is to be enrolled in the supplemental study by affixing color-coded stickers (indicating "YES" or "NO") to both the demographic portion of the form and to the filter paper. They will then send the TRF with the dried blood spots to their assigned Newborn and Prenatal Screening (NAPS) Laboratories.

The NAPS Laboratories will conduct the mandatory testing as usual on all specimens deemed adequate. Data entry of demographic information will include the decision to participate in the voluntary supplemental screening. Data will be transmitted to GDB as usual. Upon completion of mandatory testing, all filter papers will be sent to the MS/MS testing laboratory, which is on site at the Genetic Disease Laboratory Section in Berkeley. The testing laboratory will run supplemental testing *only* on adequate specimens where informed consent has been obtained and the "YES" sticker is affixed to the form. The results of the MS/MS testing will be reviewed and released by the laboratory and then sent electronically to the Genetic Disease Branch.

Written results will be released only for specimens with unusual findings. These will be sent to the newborn's physician and the hospital/collection site as listed on the TRF. For all unusual results the primary care provider will be contacted immediately via telephone by the MS/MS Project Clinical Follow-up Coordinator and the newborn referred to one of the California Children's Services (CCS)-approved Metabolic Centers for confirmation of diagnosis and initiation of treatment, if warranted.

If there is a family history of one of the conditions or other special concerns the family should be offered information on the option of obtaining supplemental testing outside of, or in addition to, the research study e.g., optional supplemental screening is offered for a fee by Neo Gen Screening, Inc. (Bridgeville, Pennsylvania http://www.neogenscreening.com) and Baylor University Medical Center (Dallas,Texas, http://www.baylordallas.edu).

The evaluation component of the project will consist of: the development and maintenance of the supplemental screening database, ongoing monitoring of all aspects of the pilot project and outcome data, including analysis of laboratory data and results, collection and analysis of follow-up clinical data, collection and analysis of cost and treatment data, and assessment of which disorders would be appropriate for inclusion in the mandatory screening program. Feedback will be solicited from parents, primary care providers, CCS Centers, state staff and contractors.

Informed Consent

During the research project written documentation of informed consent will be required for the voluntary supplemental (MS/MS research/pilot project) testing. To help facilitate this process the information about the mandatory Newborn Screening Program and the voluntary supplemental testing have been combined into one booklet. The informed consent form, which needs to be signed at the hospital, is included in the booklet. Copies of these booklets will be distributed to hospitals and prenatal care providers one month prior to the project start date.

[2] Enhancement of Newborn Metabolic Disease Screening with the Implementation of Tandem Mass Spectrometry: Proceedings of a 2000 Workshop

Key Points About the Voluntary Supplemental Research Project:

- There is no additional cost for the voluntary supplemental screening test.
- No additional blood will be taken from the newborn.
- Knowledge gained from this project will be used to improve screening for newborns and families.
- There could be some benefit to families who participate e.g., early detection and treatment for newborns with one of the disorders.
- Because this is a research study, written results will only be provided on specimens with unusual results. **If a specimen is inadequate the supplemental testing will not be run and parents will not be notified or offered retesting through the program.**

Many current participants in the mandatory Newborn Screening Program will have the following new and/or expanded roles in this project:

Role of Prenatal Care Providers/Birth Attendants:
Prenatal care providers are required by law to distribute a copy of the informational material, *Important Information for Parents About the Newborn Screening Test,* which describes the mandatory newborn screening program.[3] Prenatal care providers will need to make sure that all women who are due to deliver during the pilot period receive a copy of the revised Newborn Screening Program booklet which contains information regarding the research project and have all of their questions regarding the MS/MS research project answered.

Birth attendants will be responsible for ensuring that women who did not obtain prenatal care receive information on both the mandatory Newborn Screening Program and the MS/MS research project prior to specimen collection. They will need to verify the mother's understanding of the project and offer the option of the supplemental screening.

Role of Hospitals/Birthing Centers
Written verification of informed consent will be obtained by hospitals and birthing centers using the form included in the Newborn Screening Program booklet. Hospital staff will indicate whether the newborn is to be enrolled in the MS/MS research study by affixing color-coded stickers (indicating "YES" or "NO") on the newborn screening Test Request Form (filter paper and demographic sheet). **The MS/MS research project testing will only be done on initial adequate specimens with a "YES" sticker on the filter paper.** Hospital staff should assure correct and accurate pediatric care provider information on the form and send the 5-blood-spot specimens on the Test Request Form via the usual Newborn Screening route to the NAPS Labs for processing.

Role of Pediatric Care Providers
Pediatric Care Providers should be knowledgeable about the program and available to answer questions and provide additional information to parents and hospital staff. They will need to refer patients with unusual screening results to approved CCS Metabolic Center specialists. As always, providers should not rule out metabolic disorders solely based on newborn screening results. Any signs and symptoms of potential disorders should be followed up and any diagnosed cases reported to the GDB. It is also essential that they assure that hospitals which are entering their names and addresses on the Test Request Form have accurate and current information.

Because this is a research study, written results of the MS/MS research project will only be provided on specimens with unusual findings. In these situations, the Pediatric Care Provider will be contacted via telephone by the MS/MS Follow-up Coordinator and the newborn referred to one of the California Children's Services (CCS)-approved Metabolic Centers for confirmation of diagnosis and initiation of treatment.

Role of Metabolic Centers
The Metabolic Medical Specialists will be available to answer questions about the program, the MS/MS technology and the disorders being tested. They will also be asked to consult and participate in development and evaluation of the project.

The Metabolic Centers will make the arrangements for confirmatory testing and develop the diagnostic and treatment plan, which will then be forwarded to the primary care provider and the Genetic Disease Branch. Based upon experience of the research project and input from metabolic specialists, follow-up guidelines will be developed.

Role of Local/County Health Departments
Health Departments may be asked to locate families in their area for screening or for follow up of unusual results.

Role of NBS Area Service Center Staff
Area Service Center Staff will contact hospitals in their regions to improve reporting of correct information on the TRF and to reinforce information provided by the State

[3] California Code of Regulations, Title 17, Subchapter 9 Heritable Diseases, Sections 6500-6508

regarding the project. They will follow up with hospitals not offering the MS/MS research project testing or who have only a small percent of parents agreeing to participate. They may be asked to assist the MS/MS Project Follow-up Coordinator in locating a family or in dealing with a provider in their region.

Providers or patients who have questions can call the California Department of Health Services, Newborn Screening Program MS/MS Research Project Staff at (866) 718-7915 toll free for additional information.

Changes in Billing for The Newborn Screening Test

In addition to authorizing the tandem mass spectrometry research project, AB 2427 requires the Genetic Disease Branch to dramatically change the manner in which newborn screening test panels are billed. Since 1980, GDB has billed hospitals and other newborn screening providers. The providers, in turn, would bill patients, their insurance companies, and Medi-Cal. AB 2427 requires that as of July 1, 2001 GDB stop billing hospitals and other newborn screening providers. GDB will initiate direct billing for newborn screening:

1. **Kaiser Permanente Health Plan** will be billed directly for their patients. Kaiser patients should not receive a bill for newborn screening from GDB.

2. **Medi-Cal** patients will be billed directly to Medi-Cal. GDB has added a field to the demographic portion of the Newborn Screening Test Request Form (NBS-TRF) for the mother's Medi-Cal number. GDB will use the hospital-reported Medi-Cal number to bill Medi-Cal. Those patients whose valid Medi-Cal number is reported by the hospital will not receive a bill for newborn screening from GDB.

3. The mothers of all other patients will receive a bill for newborn screening from GDB. Accompanying the bill will be an insurance information form. Mothers will have two choices. They can pay GDB directly and then submit a claim to their insurance company for reimbursement, or they can complete the insurance information form and return it to GDB. The Genetic Disease Branch will, in turn, bill their insurance company. Included with the bill, will be the telephone number that mothers can call with questions about their bill for newborn screening.

The Genetic Disease Branch anticipates sending out its first bills for newborn screening in mid-September. This means that patients whose babies were born and tested in July and August won't receive a bill for several months after the baby's birth. Newborns tested in July, August and September will be billed $42.00 for newborn screening. We anticipate that the cost of newborn screening will rise, for the first time since 1994, to $55.00 on or about October 1, 2001.

Newborn Screening Area Service Centers (NBS-ASCs)

CHO	
Children's Hospital Oakland	(510) 428-3127
VCH	
Valley Children's Hospital	(559) 353-6416
UCLA	
UCLA Medical Center	(310) 826-4458
Harbor/UCLA	
Harbor/UCLA Medical Center	(310) 222-3751
SDICDSI	
San Diego-Imperial Counties Developmental Services, Inc.	(858) 576-2975
Kaiser N	
Kaiser Permanente, Northern CA	(510) 752-6192
Kaiser S	
Kaiser Permanente, Southern CA	(626) 564-3322

Newborn Screening News is published by: California Department of Health Services, Genetic Disease Branch, Newborn Screening Program, 2151 Berkeley Way, Annex 4, Berkeley, CA 94704-9802, (510) 540-2534

Sickle - Cell Disease

The California Newborn Screening Program

Sickle cell disease (sickle cell anemia, sickle hemoglobin C disease, sickle hemoglobin D disease, sickle hemoglobin E disease, and sickle beta thalassemia) is a group of hereditary disorders that affect the red blood cells. Under certain conditions, the blood cells of infants with sickle cell disease become sickle shaped, causing obstruction in the blood vessels. This leads to pain and/or damage to the tissues. The most serious problem for infants is infections, which can prove fatal. Newborns diagnosed with sickle cell disease are placed on antibiotic therapy and parents are provided information and instruction about preventive health measures as well as identification of symptoms requiring prompt medical attention. Sickle cell disease and other hemoglobinopathies are present in all population groups but are more prevalent in persons of African, Mediterranean, Asian, Southeast Asian, Caribbean, and South and Central American origins. In California, the incidence of sickle cell disease is about 1 per 4,400. The Newborn Screening Program detects approximately 125 cases each year.

What is Sickle Cell Disease?

In sickle cell disease, there is no hemoglobin A. Instead, there is only sickle hemoglobin, called hemoglobin S, or there may be hemoglobin S and another type of hemoglobin (C, D, E, or beta thalassemia). These hemoglobins cause the red blood cells to be hard and sticky, and change to a banana ("sickle") shape. These sticky, sickled cells can clog up the small blood vessels so the blood can't bring oxygen to the tissues. That can cause pain and damage in the area. Eventually, the sickling can affect growth and cause organ damage. The most serious problem for babies with sickle cell disease is infections. These babies can easily develop high fevers or pneumonia which require prompt treatment.

There are several types of sickle cell disease. Hemoglobin SS (also called sickle cell anemia) is the most common. Other types of sickle cell disease include hemoglobin SC disease (sickle "C" disease), hemoglobin SD or SE, and hemoglobin S beta thalassemia disease (sickle beta thal disease). Some types of sickle cell disease can cause more problems than others. For example, hemoglobin SC is often less serious than hemoglobin SS. Sickle cell disease can also affect different people in different ways, so it may be hard to know how serious it will be for a particular person.

What is the Treatment for Sickle Cell Disease?

Babies with certain types of sickle cell disease are treated with penicillin every day and get special immunizations (shots) to help prevent infections. Parents work closely with their child's doctor, the children's blood specialist (hematologist) and the sickle cell clinic. They learn how to care for their baby and recognize when to take the baby to the doctor to treat problems early. Good nutrition and extra fluids are very important. Sometimes hospitalization is needed for treatment with IV (medicine given through a thin tube into a vein) antibiotics and fluids. When the child is older, (s)he may occasionally need to be given blood.

Medications to decrease or prevent sickling of the blood are being used with some patients; effectiveness and side effects are being carefully studied. For a few people with sickle cell disease, a bone marrow transplant can be done to "cure" the disease, but this is still a high-risk procedure. A new procedure called a related-donor cord blood transplant may be possible for some families with an affected child who are planning to have another child. A blood specialist can discuss all the options with the family.

What Other Hemoglobin Conditions are Detected by Newborn Screening?

There are other combinations of hemoglobin types that babies can inherit, in which there is little or no usual hemoglobin A. These conditions are uncommon, and do not cause the red blood cells to sickle. Examples of these include hemoglobin CC, hemoglobin C beta thalassemia, hemoglobin DD, hemoglobin CE, Hemoglobin DE, and hemoglobin DC. Some of these conditions cause very few problems, while others can cause health problems.

2

Human α & β Globin Genes

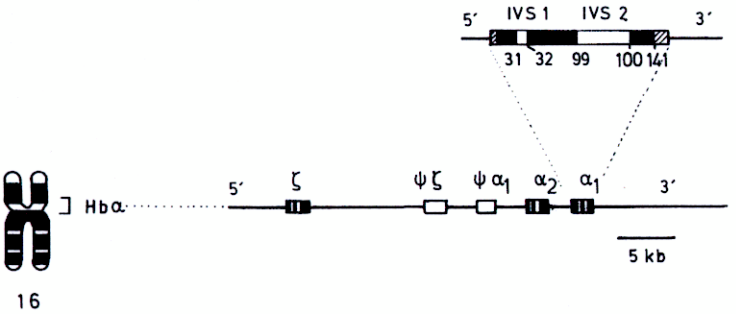


Fig. 7.32. Chromosomal location (16 p) and organization of the human α globin gene cluster. ψ, pseudogene; *IVS*, Introns (intervening sequences, *white boxes*). The numbers underneath the Hb α^1 gene 31, 32, 99, 100 ... refer to the codon numbers of the sequence at which a given intron interrupts the exon sequence. Intron 1 is interspersed between codon 31 and 32. (Only one pseudogene for Hb α shown; newly discovered pseudogene 3′ of Hb α_1 is not shown) (Updated Antonarakis et al., 1985 [12])

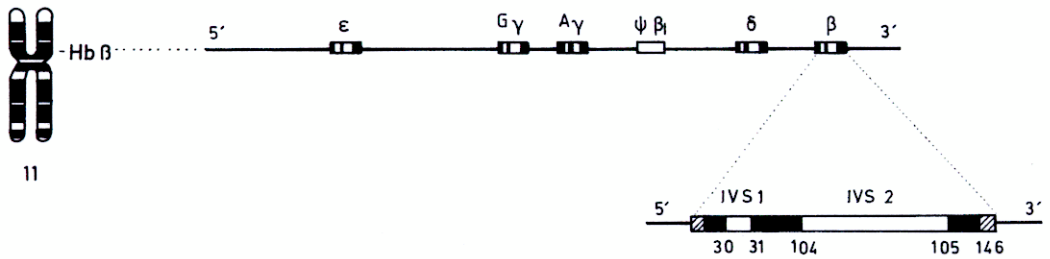


Fig. 7.33. Chromosomal location (11 p) and organization of the human β globin gene cluster. Symbols and explanation identical as for Fig. 7.32 [12]

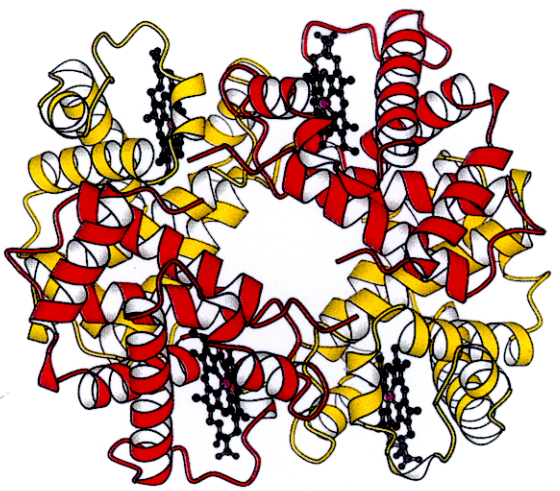


FIGURE 3.49 The $\alpha_2\beta_2$ tetramer of human hemoglobin. The structure of the two identical α subunits (red) is similar to but not identical with that of the two identical β subunits (yellow). The molecule contains four heme groups (black with the iron atom shown in purple).

Types of Alpha Thalassemia

- **Alpha thalassemia major (Hydrops Fetalis):** Deletion of all four alpha globin genes. No alpha chains, which are necessary for the formation of fetal hemoglobin, are produced. Death usually occurs in utero or early infancy. Treatment consists of ongoing transfusions.

- **Hemoglobin H (Hb H) disease:** Deletion of three alpha globin genes. The clinical complications associated with Hb H disease are variable. This generally results in mild to moderate anemia, and is often associated with microcytosis, hypochromia, and red cell fragmentation.

Hemoglobin H is an abnormal hemoglobin found in people with alpha thalassemia. When three or more alpha globin genes malfunction, there is an excess of beta globin chains. The excess chains create unstable tetramers called hemoglobin H. The tetramer of beta globin chains (4b) forms when there are insufficient alpha (a) chains to make normal adult hemoglobin (2a , 2b). The fetus manufactures gamma (g) chains rather than b chains, and the tetramer of g chains that forms is called hemoglobin Barts (4g). During the newborn period, when gamma globin production is still high and beta globin production is low, the gamma chains form the unstable tetramers identified as hemoglobin Barts. However, Hb Barts decreases with the normal decrease in gamma chain production and therefore, over time, it disappears and is replaced by Hb H. These unstable tetramers eventually precipitate in the red blood cells, causing membrane damage and premature destruction of the cells producing a chronic hemolytic anemia. It is the identification of large amounts of Hb Barts that leads us to presume the infant will have Hb H disease. DNA testing is necessary to make the final diagnosis.

- **Hemoglobin H (Hb H)-Constant Spring disease:** Deletion of two alpha globin genes and a point mutation of a third. This is generally a more severe form of Hb H disease, usually with a moderate to severe clinical course. Complications include the development of splenomegaly and cholelithiasis. Some individuals may require intermittent to chronic transfusions.

Clinical symptoms for both forms of Hb H disease that can begin at birth include pallor and jaundice. In addition, severe anemia may be caused by certain types of medications (including aspirin, sulfa drugs, some antibacterials) as well as fava beans and mothballs. Avoidance of these substances is recommended. Detailed list of substances to avoid

- **Alpha thalassemia trait (also called alpha thalassemia minor):** Deletion of two alpha globin genes. This condition is clinically benign. The clinical manifestations include microcytosis and mild, if any, anemia, which is often confused with iron deficiency anemia. However, unless the individual also has iron deficiency anemia, iron supplementation is usually not recommended. People with alpha thalassemia trait may be at risk for having a child with hemoglobin H disease or alpha thalassemia major.

- **Alpha thalassemia "silent carrier":** Deletion of one alpha globin gene. This condition is clinically benign, usually with no clinical manifestations.

What is the Treatment for Hemoglobin H Disease?

The child's doctor or blood specialist should be notified whenever the child becomes ill, so any infection can be promptly treated. If the anemia becomes severe, the child may need a blood transfusion. The doctor will discuss which medications to avoid. Extra amounts of a vitamin called folic acid may be given to the child. Parents should not have mothballs or fava beans in the home. The blood specialist will discuss how to care for the child, and what symptoms of severe anemia to watch for. Most people with hemoglobin H disease can lead relatively normal lives with proper treatment.

What is Beta Thalassemia Disease?

Beta thalassemia disease is also called beta thalassemia major, Mediterranean Anemia, or Cooley's Anemia (Dr. Thomas Cooley first described this disorder). In beta thalassemia disease, the child inherits a gene for beta thalassemia from each parent. There is an absent or decreased amount of one of the components of hemoglobin, the beta globin chains. This causes very little or no normal hemoglobin to be made. The red blood cells break down, and there is severe anemia. Without treatment, there is paleness, weakness, and poor growth. The liver and spleen can become enlarged, and changes in the bones can happen as they try to make more red blood cells. Without treatment, the heart fails, causing death.

Some types of beta thalassemia disease can be less severe, requiring less frequent treatment (these types may be called "beta thalassemia intermedia").

What is the Treatment for Beta Thalassemia Disease?

If the anemia is severe, the child will need regular blood transfusions, beginning as early as six weeks of age. Most transfusions are done once or twice a month. The child will also need medicine to remove the extra iron that builds up in the body as the red blood cells break down. There is more susceptibility to infections. Children with less severe anemia may receive less frequent transfusions, or may need them only occasionally.

For some children with beta thalassemia major, bone marrow transplants can be done if there is a well-matched donor. A successful transplant could cure the disease, however, it is still a high-risk procedure. A new procedure called related-donor cord blood transplant may be possible for families with an affected child who are planning to have another child. Some medications that could increase the amount of hemoglobin in the blood are being studied. The baby's blood specialist can discuss all the options with the family.

What is Hemoglobin E?

Hemoglobin E is a very common type of hemoglobin in Southeast Asians and in Californians of Southeast Asian origin. Newborn Screening in California detects hemoglobin E, without any of the usual hemoglobin A, in many babies every year. The test results for newborns with Hb EE and Hb E β thal look the same. Hb EE is not a disease, but Hb E β thal can be clinically significant. Repeat testing, which is part of the NBS Program, is required to distinguish between the two. Repeat testing most often will show that the baby has **hemoglobin EE**, which is not a disease, and does not require treatment. There is a mild anemia that is not helped by taking iron. The doctor should test for the amount of iron in the child's blood before giving the child extra iron.

Sometimes, further testing will show that the baby inherited a gene for hemoglobin E from one parent, and a gene for beta thalassemia from the other parent. In this case, the baby has a hemoglobin disease called **hemoglobin E beta thalassemia disease**. Effects of this disease range from mild to severe anemia and causes problems similar to beta thalassemia disease (see section above).

What is the Treatment for Hemoglobin E Beta Thalassemia Disease?

When the anemia is severe, the child will need regular blood transfusions, as in beta thalassemia disease.(See "What is the Treatment for Beta Thalassemia Disease?" above.)

Hemoglobin Diseases

Table 7.15. Clinically important hemoglobinopathies

	Disease	Genetics	Clinical severity
Sickle cell syndromes	Sickle cell anemia	Homozygote for HbS	+ + +
	Sickle β-thal disease	Compound heterozygote for HbS and β-thal	+ + to + + +
	Sickle Hb C disease	Compound heterozygote for HbS and HbC	+ to + +
	Sickle cell trait	Heterozygote for HbS	0
α Thalassemias	Hydrops fetalis	4 Hb α deletions	Lethal
	HbH disease	3 Hb α deletions (or 2 Hb α deletions and heterozygote for Hb CoSp) or point mutation	+ +
	α-thal-1 heterozygote	2 Hb α deletions or point mutation	+
	α-thal-2 heterozygote	1 Hb α deletion or point mutation	0
	Hb Constant Spring (CoSp) heterozygote	α Chain terminus mutant	+
β Thalassemia	β^0 (thalassemia major or Cooley anemia)	Homozygote	+ + + +
	β^+-thal major (Cooley anemia)	Homozygote	+ + + to + + + + [a]
	β^0/β^+ thalassemia	Compound heterozygote	+ + to + + +
	Hb Lepore heterozygote	δ-β fusion	+ (+ + + + for homozygotes)
	β^0, β^+, and $\delta\beta^0$-thal trait	Heterozygous	+
	HbE-β-thal	Compound heterozygotes	+ + + +
Unstable hemoglobin diseases	Congenital nonspherocytic hemolytic anemia of Heinz body type	Heterozygous – dominant (many different varieties)	+ +
Hemoglobins with abnormal oxygen affinity	Familial erythrocytosis (high affinity)	Heterozygote-dominant (many varieties)	+ +
M hemoglobin	Familial cyanosis (methemoglobinemia)	Heterozygote-dominant (5 varieties)	+ +

[a] Milder diseases in β-thal$^+$ homozygotes of African origin.

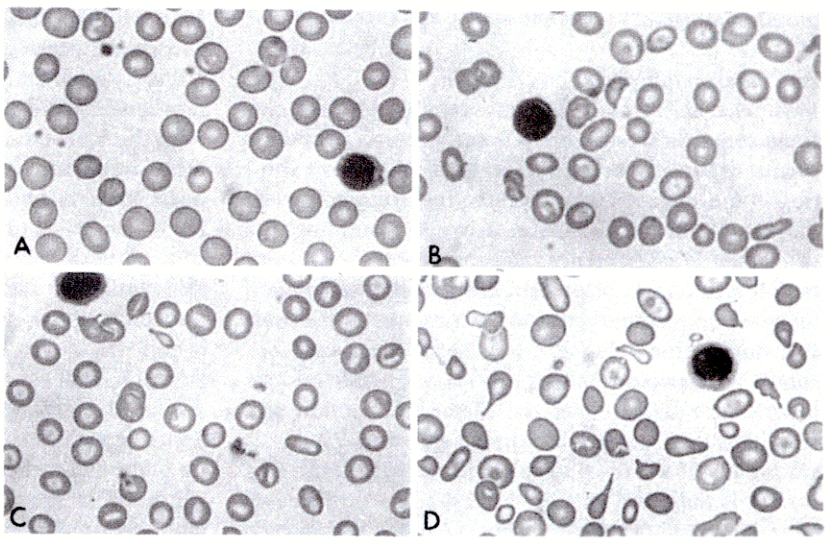


Fig. 7.48. Peripheral blood smears of a normal individual (**A**), of patients with heterozygous β thalassemia (**B**), of heterozygous α-thal-1 (**C**), and of β thalassemia major (**D**). (From Bunn et al. 1977 [42])

Thalassemia vs. Sickle cell

Thalassemia's eliminate or reduce a globin chain - α or β

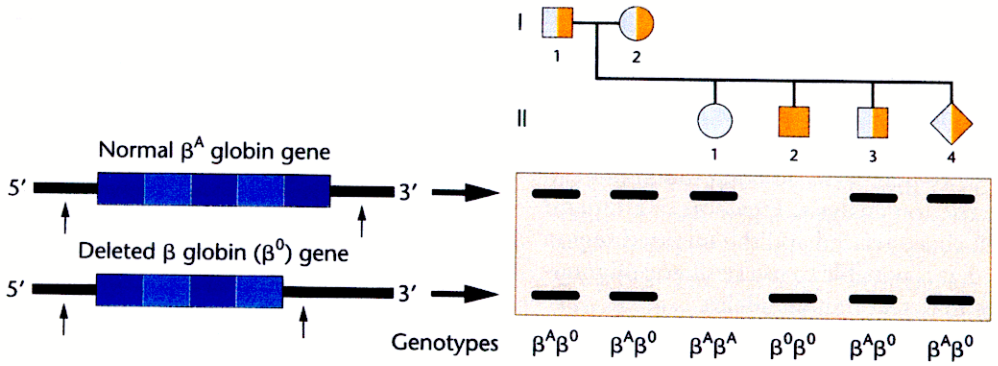


FIGURE 21.7 Diagnosis of β-thalassemia caused by a partial deletion of the β-globin gene. The family pedigree is shown positioned above each individual's genotype on a Southern blot. The normal β-globin gene (β^A) contains three exons and two introns. The deleted β-globin gene (β^0) has the third exon deleted. Arrows indicate the cutting sites for restriction enzymes used in this analysis. The normal gene produces a larger fragment (shown as the top row of fragments on the Southern blot); the smaller fragments produced by the deleted gene are represented at the bottom of the gel. The genotype of each individual in the pedigree can be determined from the pattern of bands on the blot, and these are shown below the blot.

Sickle Cell Changes the β-globin protein but does not decrease its amount

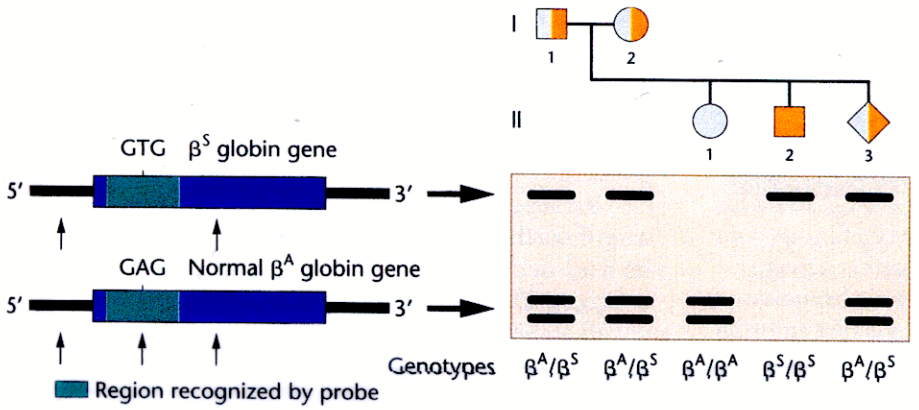


FIGURE 21.8 Southern blot diagnosis of sickle-cell anemia. Arrows represent the location of restriction enzyme cutting sites. In the mutant (β^S) globin gene, a point mutation (GAG → GTG) has destroyed a restriction enzyme cutting site, resulting in a single large fragment on a Southern blot. In the pedigree, the family has one unaffected homozygous normal daughter (II-1), an affected son (II-2), and an unaffected fetus (II-3). The genotype of each family member can be read directly from the blot, and these are shown below the blot.

MEDICAL CARE & HEALTH PROMOTION

CHILD/ADULT TESTING AND SCREENING

(1) Diagnostic process to determine whether a person has Late onset genetic disease, confirm genetic disease, or to determine if person susceptible to cancer, hypertension, high cholesterol, etc. in order to take preventive action –

(2) **Cannot Make Mandatory** – personal autonomy "Life, Liberty, ... without due process of Law" – 5th/14th Amendments to constitution. Canterbury vs. Spence (1972) informed consent. Can refuse necessary Medical care.

(3) Main issues – Access Voluntarily to all & Tort Liability

a. Legal Duty of Physicians to inform patients of availability of Test Results – If test reliable – then standard of care tort Law is applicable

b. Physician has – Liability if patients interest in needing test not considered!!

(4) Cost Barriers / Universal Access / Health Insurance pay for tests

CHILD TESTING CODE

COUNCIL ON ETHICAL AND JUDICIAL AFFAIRS, AMERICAN MEDICAL ASSOCIATION, CODE OF MEDICAL ETHICS: CURRENT OPINION 2.138

(1995).

Genetic testing of children implicates important concerns about individual autonomy and the interest of the patients. Before testing of children can be performed, there must be some potential benefit from the testing that can reasonably be viewed as outweighing the disadvantages of testing, particularly the harm from abrogating the children's future choice in knowing their genetic status. When there is such a potential benefit, parents should decide whether their children will undergo testing. If parents unreasonably request or refuse testing of their child, physician should take steps to change or, if necessary, use legal means to override the parents' choice. Applying these principles to specific circumstances yields the following conclusions:

e.g. newborn →

(1) When a child is a risk for a genetic condition for which preventive or other therapeutic measures are available, genetic testing should be offered or, in some cases, required.

(2) When a child is a risk for a genetic condition with pediatric onset for which preventive or other therapeutic measures are not available, parents generally should have discretion to decide about genetic testing.

(3) When a child is at risk for a genetic condition with adult onset for which preventive or other therapeutic measures are not

available, genetic testing of children generally should not be undertaken. Families should still be informed of the existence of tests and given the opportunity to discuss the reasons why the tests are generally not offered for children.

(4) Genetic testing for carrier status should be deferred until either the child reaches maturity, the child needs to make reproductive decisions or, in the case of children too immature to make their own reproductive decisions, reproductive decisions need to be made for the child.

(5) Genetic testing of children for the benefit of a family member should not be performed unless the testing is necessary to prevent substantial harm to the family member.

When a child's genetic status is determined incidentally, the information should be retained by the physician and entered into the patient record. Discussion of the existence of this finding should then be taken up when the child reaches maturity or needs to make reproductive decisions, so that the individual can decide whether to request disclosure of the information. It is important that physicians be consistent in disclosing both positive and negative results in the same way since if physicians raise the existence of the testing results only when the results are positive, individuals will know what the results must be. This information should not be disclosed to third parties. Genetic information should be maintained in a separate portion of the medical record to prevent mistaken disclosure.

When a child is being considered for adoption, the guidelines for genetic testing should be the same as for other children.

Genetic Testing Procedures Prior To Birth

Pre-Implantation Genetic Diagnosis (PGD)

Pre-natal DNA Testing

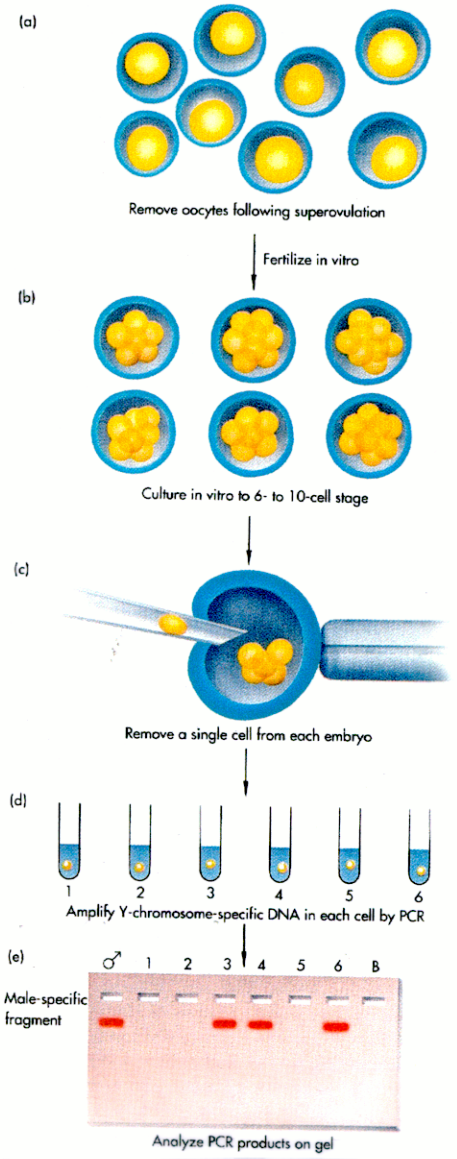


FIGURE 6-11
Determining sex of fetuses at risk for X-linked inherited disorders. (a) Oocytes are removed from the mother following superovulation and fertilized in vitro. (b) The oocytes that are fertilized successfully are cultured in vitro until there are 6 to 10 cells in each embryo. (c) A hole is made in the zona pellucida and a single cell removed from each embryo. (d) Amplification of the DYZ1 sequence is attempted. (e) Only in DNA from males is the male-specific DYZ1 sequence amplified by PCR, giving rise to a 149-bp, male-specific fragment. The lane marked with the male symbol is a positive control showing the expected fragment; the lane marked B (for "Blank") is from a PCR that included all the reagents but no DNA and is used to detect any contamination. Female embryos are negative (lanes 1, 2, and 5) and are implanted into the mothers.

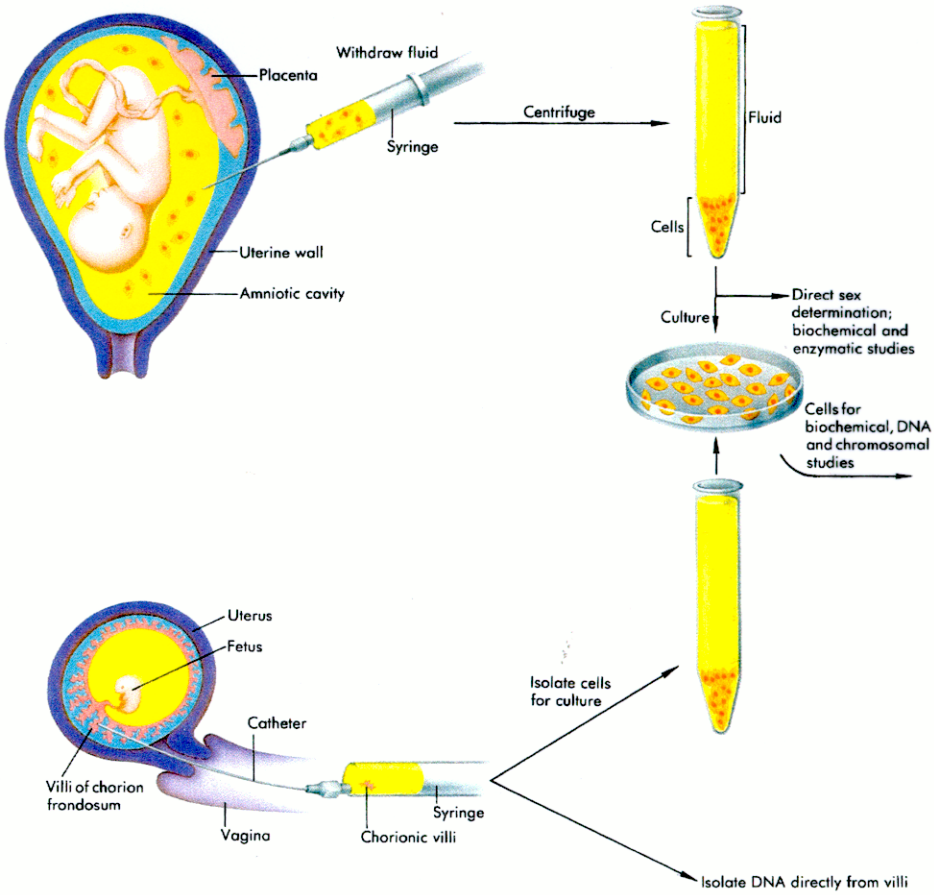
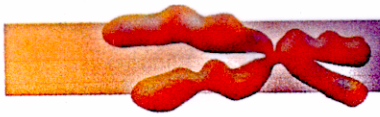


FIGURE 27-1
Amniocentesis and chorionic villus sampling. (a) A sample of amniotic fluid (mostly fetal urine and other secretions) is taken by inserting a needle into the amniotic cavity during or around the sixteenth week of gestation. The fetal cells are separated from the fluid by centrifugation. The cells can be used immediately, or more usually they are cultured so that a number of biochemical, enzymatic, and chromosomal analyses can be made. The cultured cells can also be a source of DNA. (b) Chorionic villus sampling is performed between the eighth and twelfth weeks of gestation. A catheter is introduced through the vagina or transabdominally, and a small sample of chorionic villi is drawn into the syringe. DNA can be isolated directly from the tissue, or cell cultures can be established. Note that the various elements of this figure are not drawn to scale.

DNA, Biochemical, Mass Spec, karyotype

DNA only!

Pre-Implantation & Pre-Natal Testing Issues



Social and Ethical Issues Surrounding Preimplantation Embryo Diagnosis

- In 1990, Germany passed a law that prohibits preimplantation embryo testing.
- A 1993 report from Canada's Commission on Reproductive Technologies warns against allowing market forces to determine the use of reproductive technologies. It also calls for creation of a permanent regulatory and licensing body to govern all aspects of the new reproductive practices, including sperm banks and *in vitro* fertilization.
- In 1994, France and Norway passed legislation that limits genetic testing to situations in which the results are medically therapeutic, and authorizes governmental bodies to establish the criteria for defining "therapeutic" in this context. These laws prohibit the use of genetic testing for sex selection and normal trait enhancement.
- In 1994, a U.S. National Institutes of Health advisory panel issued guidelines for federally funded research on embryos. These guidelines allow the use of preimplantation embryo testing for disease diagnosis and accept the practice of determining an embryo's gender to diagnose a sex-linked disease, such as hemophilia A. The guidelines do not accept sex selection for any other reason. An oversight committee would monitor compliance with the guidelines to ensure the scientific qualifications of federally funded researchers as well as the likelihood that their studies will produce "significant scientific or clinical benefit" that cannot be "otherwise accomplished by using animals or unfertilized gametes."

At the same time, the United States had more than 300 privately run, unregulated *in vitro* fertilization clinics, commonly referred to as IVF Centers. Most of these centers were willing to do whatever a paying client requested, including sex selection and analysis of the genetic susceptibility for complex traits whose inheritance is not yet well understood.

This range of responses to the issues generated by the new reproductive technologies shows a diversity of approaches based on national culture and history. It also reflects international apprehension about the potential for misuse and abuse of the new technologies. Here are some of the main concerns.

1 When Should the Tests Be Used?

The couple in our opening story whose firstborn suffered from cystic fibrosis faced a medical problem. Preimplantation diagnosis could help them have a second child unaffected by the disease. With no cure at present for CF and no therapy that allows CF-affected people to look forward to a life of normal length, this is an example of medically therapeutic testing. Most governmental committees and bodies argue against testing for any other reason, but commercial clinics do not. Moreover, if postnatal therapies for cystic fibrosis, such as nasal sprays that introduce a normal CFTR protein into the respiratory tissues or protocols that insert normal *CF* genes in the cells of the lungs and nasal passages, become available, some medical practitioners may no longer consider preimplantation diagnosis a preferred therapy.

2 How Should the Tests Be Carried Out?

The couple in our opening story began by consulting a genetic counselor and then worked with medical practitioners associated with a university laboratory. Most geneticists agree that counseling before a procedure should foster an open discussion of all the issues (including the possibility that the tests might give false negatives); and that long-term follow-up should be part of the process. The preimplantation testing itself, like other forms of genetic testing, should be carried out by highly trained personnel in licensed laboratories. These accredited laboratories operate according to professional standards and have scientific and ethical review boards that monitor all work.

3 Who Should Have Access to the Technology?

The combination of *in vitro* fertilization and preimplantation testing cost $6000 to $10,000 in 1994. Should the government provide tests for people who cannot afford them? How should society decide this issue? (A related discussion of access to medical technology appears in the Genetics and Society box in Chapter 1.)

4 Should Parents Have the Right to Make Any Genetic Decision?

If, for instance, they decide to have a child affected by a genetic disease, should they bear all financial responsibility for its care, or should some form of universal health insurance provide help?

5 Who Should Have Access to Test Results?

Just the parents? The parents and eventually the child? The parents, the child, and certain community institutions, such as schools? Some combination of these plus commercial enterprises such as insurance companies and places of employment? (We discuss this same question of privacy in relation to other types of genetic testing in the Genetics and Society boxes in Chapters 1 and 2.) Family?

6 What Constitutes a Human Individual?

Cultural and religious beliefs, rather than scientific knowledge and social customs, are the basis for answers to this question. Some people see preimplantation diagnosis as an alternative to abortion that allows a couple to make a decision before pregnancy, and thus a life, begins. Others argue that even at the eight-cell stage, a preimplantation embryo is the equivalent of a human being, and rejection of an embryo is the equivalent of killing a human being.

Although there are no simple solutions to these complex is , geneticists around the world agree on the need for continuous discussion and tight oversight of the development of the nev - productive technologies.

Roe vs. Wade

FAMILY PLANNING AND REPRODUCTIVE ISSUES

(1) Major Impact - Hundreds of genetic disease genes can be screened for using PGD, amniocentesis, or chorionic villi testing + parental/family testing — with Human Genome Sequence - ALL disease genes can be tested for!!!

Can determine whether parents are carriers &/or embryo, fetus, child has genetic disease!

(2) Remember - ~ 1.2% of all live births have genetic defect due to a mutation in a disease gene! And we all carry a few deleterious gene alleles!

(3) Legal Issues Controversial

a. abortion debate/embryo rights
b. women's rights/reproductive choice
c. eugenic concerns
d. genetic engineering of human cells

(4) Ethical & Legal Presumption of Voluntary Choice for Reproductive Matters - Voluntary testing & Woman's Right to Reproductive Choice -

a. VOLUNTARY TESTING
b. Griswold vs. Connecticut + Roe vs. Wade (contraception/abortion) right to privacy is conceptualize as a substantive 5th/14th amendment liberty - Procreative choice is hedged by a number of

tradition embedded into Living constitution — right to privacy + subsidiary right to determine pregnancy — up to 1st Trimester —

(5) Main Legal Issue — Tort Liability

a. Gain access to knowledge + act on it by contraception, embryo testing, pre-natal testing, &/or abortion

b. Tort Law states that couples have the right to avoid birth of handicapped children if tests or procedures to avoid birth are available

c. Wrongful birth/Life Cases

(1) Curlender vs. Bio-Science Laboratories (CA) Child/parents can bring tort suit against a lab that failed to carry out Tay-Sachs test properly giving birth to child — Legal Tort Liability

(2) Grodin vs. Grodin (MI)

Court held that boy can sue mother for causing his teeth to be brown because she took tetracycline during pregnancy!

Several States have now enacted statutes prohibiting wrongful Life suits against parents by children — CA — Turpin vs. Sortini — "purpose to eliminate any Liability or other economic pressure which might induce parents to abort or conceive a potentially defective child"

"Wrongful-Birth" Lawsuits Abolished in Georgia and In Michigan

By Liz Townsend

Courts in Michigan and Georgia have rejected attempts by parents of disabled children to sue doctors who, the parents claimed, failed to discover their babies' birth defects in time for an abortion.

The Michigan Court of Appeals and the Georgia Supreme Court ruled that these "wrongful-birth" lawsuits are invalid under state law. The Michigan appeals court warned that such suits "could quickly slide into applied eugenics and the elimination of supposedly unfit lives," while Georgia's high court held that state law "does not recognize a cause of action for wrongful birth."

Wrongful-birth lawsuits remain legal in 27 states. Doctors such as James Delahunty of New Jersey, founder of the Association of Pro-Life Obstetricians and Gynecologists, have lost suits that claimed they refused or neglected to offer amniocentesis or other diagnostic tests that could have identified babies' disabilities to pregnant women.

Last March Delahunty was ordered to pay $1.85 million to the parents of Michael Imber-gamo, a four-year-old little boy with Down syndrome. Michael's parents testified that they would have aborted him if they had discovered his condition before birth, the *Washington Times* reported.

"Some women want to kill their children because they are handicapped," said Delahunty, according to the *Times* . "If genetic tests give them wrong results, they blame the doctor. I was blamed."

Delahunty's lawyer said that wrongful-birth lawsuits are a product of technology that can more easily identify disabilities in unborn children. "Patients who had disabled children in the past didn't think of suing the doctor," Tom Chamsky told the *Times* . "But as technology has grown, some women think that their child's disability is someone else's fault."

Both the Georgia and Michigan cases concerned babies whose disabilities were not identified by doctors from ultrasound tests.

The Georgia case involved the son of Andrew and Jennifer Etkind, who was born with Down syndrome in September 1995. According to the Georgia Supreme Court's July 8 decision, Dr. Ramon Suarez told Jennifer Etkind (who is also a doctor) that her baby "was developing normally and that she was not at risk for birth defects" after two ultrasounds and a blood test, and advised against the more invasive amniocentesis procedure. Dr. Etkind did not have an amniocentesis.

After their son was born with Down syndrome and a malformed heart, the Etkinds sued Suarez. According to the court decision, the Etkinds asserted that "but for the treatment or advice provided by the defendant, [they] would have aborted the fetus, thereby preventing the birth." The Etkinds sought to have Suarez pay for the costs of raising their son, the *Atlanta Journal-Constitution* reported.

The Georgia Supreme Court had previously abolished wrongful-birth lawsuits in the 1990 *Atlanta Obstetrics & Gynecology Group v. Abelson* decision. The Etkinds asked the court to overturn *Abelson* on several grounds, including constitutional and due process concerns. However, the court, by a 6-1 majority, rejected all their arguments, ruling that "Georgia tort law does not recognize a cause of action for wrongful birth."

The Etkinds' main contention was that Dr. Suarez's failure to identify the baby's Down syndrome "interfered with their choice of whether to have an abortion" and that the ban on wrongful-birth suits also stands in the way of the abortion "right," according to the court decision.

However, the court insisted, "refusal to recognize wrongful birth, absent authorizing legislation, does not interfere with Dr. Etkind's constitutional right to an abortion."

In a strongly worded decision, the Michigan Court of Appeals rejected the lawsuit brought by the parents of four-year-old Shelby Taylor, who sued Dr. Surender Kurapati for finding "no visible abnormalities" in a December 4, 1993, ultrasound.

According to the June 25 appeals court decision, Shelby was born on April 19, 1994, with a "missing right shoulder, fusion of left elbow, missing digits on left hand, missing femur on left leg and short femur on right," according to the court. Her parents contended that "the failure to reveal the disabilities deprived the Taylors of their right to make a reproductive decision regarding the pregnancy," according to the court decision. They also alleged that Kurapati was liable for the "emotional distress" they suffered when their little girl was born.

Overturning prior decisions that had allowed such lawsuits, the Court of Appeals rejected the Taylors' arguments and ruled that wrongful-birth suits are not valid under state law. The court saw much danger in the theory behind these suits, that parents should be compensated if they were not able to abort a disabled child.

"The very phrase 'wrongful birth' suggests that the birth of the disabled child was wrong and should have been prevented," Judge J. Whitbeck wrote for the 2-1 majority. "If one accepts the premise that the birth of one 'defective' child should have been prevented, then it is but a short step to accepting the premise that the births of classes of 'defective' children should be similarly prevented, not just for the benefit of the parents but also for the benefit of society as a whole through the protection of 'public welfare.' This is the operating principle of eugenics."

The court also rejected the argument that wrongful-birth lawsuits are required to ensure the "right" to abortion that was legalized in *Roe v. Wade* . Whitbeck wrote that *Roe* allows the "state to make a value judgment favoring childbirth over abortion." For example, previous courts have found that the Michigan Consti-tution does not require the state to fund abortions, but Michigan does provide financial support for childbirth.

"As the state has no obligation to affirmatively aid a woman in obtaining an elective abortion by paying for it," Whitbeck wrote, "the state similarly has no obligation to take the affirmative step of imposing a civil liability on a party for failing to provide a pregnant woman with information that would make her more likely to have an elective, and eugenic, abortion."

The Michigan decision called attention to the "slippery slope" that is evident in wrongful-birth lawsuits, a slope that pro-lifers have been warning about for years. "[I]t is but another short half step from the concept of preventing the birth of an 'unfit' or 'defective' child to proposing, for the benefit of the child's overburdened parents and of the society as a whole, that the existence of the child should not be allowed to continue," Whitbeck wrote.

"After all, if that child never should have been born, then that child has no real right to go on living, thereby imposing the costs of the child's continued existence upon the parents and society. This, we conclude, is the logical end of the slippery slope inherent in the application of the benefits rule through the wrongful birth tort."

(5) Mandatory testing

a. would violate right to bodily integrity!

b. Tort Law requirement that persons be informed that tests are availble to screen for carrier status! But tests

c. Key questions?

(1) Do people have an obligation to reproduce "reasonably" & not impose undue burdens on offspring or society? Who pays?

(2) Is it possible/desirable to reduce births of children with genetic diseases? IF CAN BE PREVENTED By Testing (e.g., BABI?) Eugenics!

Ethical Scenarios in Genetic Testing

GENETIC TESTING
- DISCUSSION
- LAWS, CONVENTIONS & GUIDELINES
- INDUSTRY ASSOCIATIONS IN ACTION
- SCIENCE CENTRE

OTHER DISCUSSION SCENARIOS
- CLONING
- GENE THERAPY
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WHOSE VALUES? WHO DECIDES?

Genetic Testing Discussion Scenario

Introduction

Welcome to the *Discussion Scenarios*. The five scenarios in this section present many of the ethical issues that come up in connection with some uses of biotechnology. You've entered a discussion about: **genetic testing**.

You are about to read a series of short stories. The stories are fictitious, but we hope the situations we describe and the questions we raise will help you consider different points of view on the ethical issues associated with **genetic testing**. The questions aren't necessarily intended to lead you to a set of answers. The purpose is to encourage you to think about the issues from a variety of perspectives.

These *Discussion Scenarios* may not address all the ethical issues or concerns related to **genetic testing**. We also recognize that we may not have asked all the 'appropriate' questions to help bring the issues to light or that are of importance to you, and realize that no choice of questions can be truly 'ethically neutral.'

This is why the questions are intended as a starting point for a broader look at the issues associated with **genetic testing**. We'll be revising the questions over time, so we encourage you to get back to us with new issues that you consider important.

Elsewhere in this web site, in the section called Whose Values? Who Decides?, we talk about the difference between individual and societal ethics. There are many situations where what we want as individuals may not be the same as what we expect our government to do. As you read the *Discussion Scenarios*, think about which questions in the stories should be left to the individuals or companies to decide, and which ones should be answered by society as a whole. We also look at two different philosophical viewpoints that underlie ethical decisions. In one of these traditions, decisions are evaluated based on their *consequences*. In the other, choices are based on

a set of *principles* , regardless of the consequences. As you read each story, think about how these philosophical approaches and other kinds of information can help you reach your own conclusions on the ethics of **genetic testing**.

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Scenario A : Having Children; Exploring the Options

Faye and Michael want to start a family. But they know that both of their families have a history of *Tay-Sachs Disease*, an incurable condition that leads to deterioration in a person's brain. Children born with Tay-Sachs usually lose their eyesight after about a year, and rarely live beyond the age of five. [1]

Knowing that a person can carry Tay-Sachs without getting it, Michael and Faye asked their doctor for **genetic testing** first to determine if they are carriers and second to find out whether their future children might be at risk. Based on blood sampling, they found out that they were both carriers, meaning that a child they conceived naturally would have a one in four chance of being born with the disease.

Faye and Michael must now decide whether to conceive a child naturally, adopt a child, not have children at all or request pre-implantation genetic diagnosis (PGD). PGD is a relatively new technology where a number of Faye's eggs are fertilized by Michael's sperm in a laboratory. Genetic testing identifies the embryos that are most likely to be Tay-Sachs carriers, or to acquire the disease, and those embryos are not reimplanted in Faye's womb.

Discussion Questions

1. If Faye and Michael decide to have a child, they want to do everything they can to make sure the child is not born with Tay-Sachs, since they believe this would be a very painful experience for the child, and for themselves. Is this a reasonable decision to make? Why or why not?

   Here are some of the alternatives available to Faye and Michael if they decide they want a child:

   - They could conceive the child naturally, but terminate the pregnancy if a prenatal genetic test shows that the fetus has Tay-Sachs. The couple would have a choice of two tests chorionic villus sampling, which takes place after 10 to 12 weeks of development, or amniocentesis, which is carried out after 16 weeks of development. Both tests carry a risk of miscarriage, in the range of 1 in 500.[2] The risk may be slightly higher for chorionic villus sampling.

   - Another option is to use the relatively new technology of pre-implantation genetic diagnosis (PGD), where a number of Faye's eggs are fertilized by Michael's sperm in a laboratory. Genetic testing identifies the embryos that are most likely to be Tay-Sachs carriers, or to acquire the disease, and those embryos are not reimplanted in Faye's womb.

This type of genetic testing takes place at a much earlier stage, and avoids the risk of miscarriage or harm to the fetus that can occur with amniocentesis or chorionic villus sampling. But the *in vitro* fertilization technique that accompanies the test has other drawbacks. Only one in four implanted embryos results in a pregnancy, and some women experience side-effects from the fertility drugs they have to take during *in vitro* fertilization. The process is very expensive, is usually paid for by the couple, and is not currently available in all Canadian cities.

- The couple could adopt, knowing that their child will not likely have Tay-Sachs. However this may not be a realistic option if Faye and Michael are determined to raise a child "of their own flesh and blood."

- Can you think of any other alternatives available to Faye and Michael?

Of the options we've listed, is one more or less acceptable than the others? To what extent is Faye and Michael's decision theirs alone? Are there social norms or values that would make any of the options more or less acceptable?

2. Should the public health care system ensure that genetic testing is available to any Canadian who wants it? Should the health system cover the cost? Should the system cover some tests, but not others? If some tests are not covered, to what extent should they be available to people who are willing to pay for them? Who should make these decisions, and on what basis?

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Scenario B. Prenatal Genetic Testing

Adapted from a Scenario Composed by Ted Schrecker

Instead of dealing with a specific condition, this scenario refers to ***Condition X***, to highlight the element of the genetic testing debate that has to do with the nature of the conditions to be detected.

Susan and her husband Jean-Claude know that there is a history of Condition X in both of their families. When Susan finds out she is pregnant, she asks her doctor whether there is a test that can determine whether the fetus:

- will be affected by the disorder; or
- will be a carrier of the disorder who can pass it on to future generations.

The test is available, so Susan and Jean-Claude decide to have it performed as early as possible in the pregnancy. Prior to having the test performed, Susan and Jean-Claude hear a radio interview with a medical geneticist, who says it would be truly unfortunate for a child to be born with Condition X when genetic testing can diagnose the disorder prenatally.

Discussion Questions -- Prenatal Genetic Testing

1. Let's say Condition X is Huntington's disease, and tests show that the fetus

will develop the disease. Huntington's symptoms do not appear until a person reaches middle age, so that he or she could make constructive, informed life decisions with the information available through genetic testing. On the other hand, advance knowledge of what the future holds could be devastating for the person and his or her family, even before the disorder develops. Should Susan terminate the pregnancy, or carry it to term?

2. What if Condition X is familial hypercholesterolemia, a condition that increases the likelihood of dying of heart disease by middle age, but can be treated through diet and other choices?

3. What if Condition X is WAGR syndrome, a rare hereditary disorder that can involve mental retardation, several kinds of cancer, and genito-urinary abnormalities?

4. What if people with Condition X could live almost as long as anyone else, but only if they had access to full-time care, either at home or in an institution? If Susan and Jean-Claude continue the pregnancy, who should pay for that care?

5. Susan and Jean-Claude did not plan their pregnancy and therefore did not seek counselling to discuss their options before Susan became pregnant? What form of counselling would be appropriate now? Who should provide the counselling? How can individual choices be respected?

6. Are there genetic tests that should or should not be funded by the public health care system? Who should decide which tests are funded? What criteria should be used to determine which tests are funded? If some tests are not covered, should they be available to people who are willing to pay for them, and to what extent?

7. Do you agree with the view expressed in the radio interview with the medical geneticist? Does your answer depend on what Condition X is? How could the geneticist's point of view affect people who are already living with Condition X, and their families? How could our answers affect social attitudes, and even legal attitudes, toward people with genetic disorders?

8. Many genetic tests are now being developed and marketed by private companies. What should these companies, and industry as a whole, be doing to inform consumers and health professionals about the possibilities and limitations of genetic testing?

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Scenario C. Selecting for Genetic Traits

(Adapted from GenEthics Consortium Case Literature NHGRI at NIH)[3]

Harry and Martha are worried about having a second child with Severe Combined Immune Deficiency (SCID). Children born with SCID have seriously impaired immune systems, as a result patients may succumb to any number of infections. As recently as 20 years ago, children with SCID died early in life, but the use of bone

marrow transplants has greatly extended survival and, in some cases, led to better quality of life. In general, results are best when a transplant is done early, and when the marrow donor and recipient have similar genes that code for *Human Leukocyte Antigens (HLAs)*. HLAs are a family of cell surface proteins that are critical for the activation of immune responses. The HLA genes are the most variable set of human genes known[4] and a close match is most likely if the donor is a brother or sister.

Harry and Martha have signed up with a new private clinic that offers pre-implantation genetic diagnosis (PGD). With this technique, a number of the woman's eggs are fertilized by her partner's sperm in a laboratory, and each of the embryos is tested before being reimplanted in her womb. This makes it possible to select embryos that are free of genetic disease.

Harry and Martha tell the medical geneticist they want to undergo PGD so they can begin their pregnancy knowing that the baby won't have the disorder. A few weeks later, they give a second reason: Their six-year-old daughter with SCID is getting sicker with the disease, and they hope to use bone marrow from a second child to save their daughter. Is it possible, they ask, to test the healthy embryos for HLA genetic compatibility and transfer only those that most closely match their daughter's type?

The geneticist knows that the technology can be used in this way, but wonders whether agreeing to the couple's request would be ethical.

Discussion Questions

Genetic Traits vs. Genetic Disorders

1. PGD can be used to identify embryos that are less likely to develop specific disorders, like muscular dystrophy or Down Syndrome. Harry and Martha asked the geneticist to select embryos that were free of the SCID mutation and had genes that were compatible with their daughter's. But a person's HLA status is not a disorder it's a genetic trait, just like his or her gender, or the colour of his or her eyes or hair.

    a. Is it ever appropriate to select an embryo based on genetic *traits*, rather than *disorders*?
    b. Should the decision be up to the individuals involved?
    c. Are there social norms or values that make it acceptable or unacceptable to select embryos for their genetic traits in certain situations?

Fate of the Unselected Embryos

2. The PGD procedure involves fertilizing a number of eggs in a laboratory *(in vitro* fertilization). In Harry and Martha's case, if the geneticist agreed to their request, only those embryos that were free of the SCID mutation and compatible with their daughter's HLA genes would be implanted.

    a. What should be done with the embryos that have been screened out?
    b. Should the couple donate them for medical research?.. store them for later use?... donate them to other couples for *in vitro* fertilization?...or have them destroyed?
    c. Is the couple's decision completely up to them, or are there social norms or values that would argue for or against any of these options?

PRIVACY AND CONFIDENTIALITY ISSUES OF TEST RESULTS

(1) State laws now exist requiring confidentiality of genetic testing –

(2) Privacy = Control over access to others!
vs.
Confidentiality = Control <u>after</u> access given to someone

(3) <u>Generally</u> – a person controls or "owns" whether his/her DNA given for testing (not property rights once given, however – Moore vs. Regents UC!!) or if given for testing whether results are disclosed.

(4) <u>Legal Issues</u>

a. Relatives duty to provide DNA for linkage studies – If could benefit from treatment or serious illness & if bodily intrusion minimal (e.g.; cheek swab), state could override privacy interests & require DNA sample to be given!!

b. Physician's Duty to inform at risk relatives – e.g.; cancer – health protection overrides privacy issue! Must inform relatives if prevent harm to others!

Tarasoff vs. Regents UC – a psychiatrist <u>must</u> inform – person who is at risk as a result of information obtained from patient!

State Genetic Privacy Laws

Last updated: 4/15/02

Medical information is presumed confidential, but increasing capabilities to store and rapidly transfer data escalate the challenge of protecting privacy. Laws in all states restrict access to medical records. At issue is whether genetic information should be protected generally, as another component of health data, or by special genetic privacy laws.

The case against "genetic exceptionalism" asserts that genetic information is fundamentally no different than other health data and special protections for one type of information could deny safeguards that should be established more generally. Proponents argue that the stability of genetic information and unique predictive - rather than merely historic - qualities warrant special consideration.

Laws in 16 states require informed consent for a third party either perform or require a genetic test or to obtain genetic information. Twenty-three states require informed consent to disclose genetic information. In addition, Rhode Island and Washington require written authorization to disclose genetic information. Colorado, Florida, Georgia, and Louisiana explicitly define genetic information as personal property. In 2001 Oregon repealed its property right to DNA samples and genetic information. Four states mandate individual access to personal genetic information, and 17 states have established specific penalties - civil or criminal - for violating genetic privacy laws.

State and Statute	Personal Access to Genetic Information Required	Informed Consent Required to				Define as Personal Property		Specific Penalties for Genetic Privacy Violations
		Perform/ Require Genetic Test	Obtain/ Access Genetic Information	Retain Genetic Information	Disclose Genetic Information	Genetic Information	DNA Samples	
California Insurance €10149.1					Ã			Ã
Total	4	11	5	6	25	4	0	17

1 Limits disclosures of and access to genetic information by employers and insurers.
2 Requires written authorization only

WORKPLACE ISSUES

STATE LAWS

(1) Several States now have Laws preventing genetic testing & discrimination in the workplace! Including CA

(2) Many States prohibit discrimination on the basis of specific genetic traits — e.g., sickle-cell

(3) But— American Employment Law is based on "At will" rule — employers free to hire or fire people for no reason unless protected by collective bargaining agreements — & now protect against race, gender, handicap, & genetic discrimination —!!

(4) State Laws vary from state to state —

WORK PLACE ISSUES

FEDERAL LAWS

(1) ADA - Americans with Disability Act (1990) enforced by Equal Employment opportunity Comission (EEOC). EEOC in 1995 interpreted the ADA to include genetic predisposition/diseases - not including carriers!

(2) Title VII of 1964 Civil Rights Act - prohibits discrimination on basis of race, sex, religion, or national origin. could include genes if present at high level in a particular group (e.g. sickle cell).

(3) OSHA - Occupational & Safety Health Act

Employers required to furnish employees a place of employment free of hazards!

What if employee has genetic condition making him/her sensitive to workplace environment? Could require testing - Carpal tunnel syndrome case -

(4) Clinton Executive Order - 2000

Prevents/Prohibits Federal Employees from being discriminated against on basis of genetics/genes!

NCSL Genetics Tables

State Genetics Employment Laws

Last updated: 2/3/03

Several states acted against employer use of genetic information in the 1970s and '80s to prohibit employer discrimination against applicants with the sickle cell trait. Wisconsin was the first state to ban genetic testing and discrimination in the workplace in 1991. With Hawaii, Utah and Virginia enacting measures in 2002, genetic nondiscrimination in employment laws are in place in 31 states. The scope and functions of these laws vary widely. All laws prohibit discrimination based on the results of genetic tests; many extend the protections to inherited characteristics, and some include test result of family members, family history and information about genetic testing, such as the receipt of genetic services. Most states also restrict employer access to genetic information, with some prohibiting employers from requesting, requiring and obtaining genetic information or genetic test results, or directly or indirectly performing or administering genetic tests.

On the federal level, the Equal Employment Opportunity Commission in 1995 interpreted "disability" in the Americans with Disabilities Act to include genetic predisposition to disease, but conflicting rulings raise questions whether the Supreme Court would accept the EEOC interpretation. President Clinton in February 2000 banned genetic discrimination in the federal workplace and called on Congress to pass a federal genetic information nondiscrimination law for private sector employment. The U.S. Senate debated the matter during the summer of 2000, but took no action.

State and Statute	Genetic Nondiscrimination Covers					Genetic discrimination prohibited in hiring, firing, and/or terms, conditions or privileges of employment	Prohibits Employer From				Specific Penalties for Genetic Discrimination in Employment
	Predictive Genetic Information Only	Genetic Test Results	Information About Genetic Testing	Family History	Inherited Characteristics		Requesting Genetic Information/Genetic Test	Requiring Genetic Information/Genetic Test	Performing Genetic Test	Obtaining Genetic Information/Genetic Test Result	
Total	9	31	9	12	16	31	18	24	16	10	12
California Gov't. §12926, Gov't. §12940	√	√		√	√	√			√		

WORK PLACE ISSUES

U.S. News 2/19/01

The dark side of genetic testing

Railroad workers allege secret sampling

By Dana Hawkins

John Wiebelhaus, a fourth-generation railroad man, makes his living with his hands, laying miles of track, repairing heavy steel rails, and picking ice from the track's switches. Tough work–but Wiebelhaus loves it. "I like the idea that tracks I lay could be there 100 years," he says. "It's in my blood."

That may not be all that's in his blood, which is why, the track-maintenance foreman claims, his employer, the Burlington Northern Santa Fe railroad, has been secretly testing the blood of workers with carpal tunnel syndrome. "The railroad wants to be able to say: 'You were a time bomb. Because you are genetically predisposed to the disease, you would've gotten it whether you were a soda jerk or running a jackhammer,' " says Harry Zanville, an attorney for the railworkers' union that last week filed a lawsuit, along with Wiebelhaus, to force the company to stop the alleged covert testing. He claims that 125 workers recently gave blood samples and that at least 18 were subjected to gene tests without the employees' consent. The reason: Money, says Zanville, insisting the company hopes to avoid paying out millions in medical bills and disability to workers who develop the painful musculoskeletal disorder on the job.

The federal court lawsuit, the first of its kind against a private company, charges that the furtive testing violates the Americans with Disabilities Act and several state laws barring DNA testing by employers. The U.S. Equal Employment Opportunity Commission filed a separate petition, also in a federal court in the Northern District of Iowa. The EEOC alleges that the Fort Worth-based railroad required blood samples from workers who had submitted claims arising from carpal tunnel injuries. The blood was then allegedly tested for a genetic defect that may predispose a person to some forms of the ailment. Athena Diagnostics, the lab that allegedly conducted the tests, is also a defendant in the union's case.

Hidden reason. Gary Avary, a BNSF employee, says he discovered the alleged covert screening last month after he received a letter from his employer directing him to get his blood tested. The Nebraska track laborer had recently returned to his job after successful carpal tunnel surgery. The lawsuit alleges that when his wife, a registered nurse, inquired about the test "the secret intentions of the BNSF were inadvertently revealed." After Avary refused to take the test, the company informed him that he would be investigated for failing to cooperate. A railroad spokesperson says BNSF doesn't require workers to submit to genetic testing but that "some employees were asked to take a test."

The railroad employees are encouraged by a federal court's approval last December of a settlement in a case involving the genetic privacy rights of workers at Lawrence Berkeley Laboratory. As first reported by *U.S. News*, LBL workers for decades were tested without their knowledge for syphilis, pregnancy, and the genetic trait for sickle cell disease. President Clinton last year banned genetic discrimination against federal employees, but Congress has not extended the rule to the private sector. "It's important for the public to have confidence that genetic tests will be used for their benefit," says Paul Billings, cofounder of GeneSage, a company that promotes responsible DNA screening. "Unfortunately, this case suggests that we're still in the dark ages of employment-based testing."

Geneticists in particular question the legitimacy of the carpal tunnel test. They point out that the disease is a common workplace disability, and mutations of it are extremely rare. "I'm a humanist physician. I try hard to make the world a better place," says Phillip Chance, a geneticist at the University of Washington in Seattle, who discovered one of the mutations. "This would be the last thing I'd want to see happen with my work."

What is Carpal Tunnel Syndrome?
Carpal tunnel syndrome occurs when tendons or ligaments in the wrist become enlarged, often from inflammation, after being aggravated. The narrowed tunnel of bones and ligaments in the wrist pinches the nerves that reach the fingers and the muscles at the base of the thumb. The first symptoms usually appear at night. Symptoms range from a burning, tingling numbness in the fingers, especially the thumb and the index and middle fingers, to difficulty gripping or making a fist, to dropping things. Some cases of carpal tunnel syndrome are due to work-related cumulative trauma of the wrist. Diseases or conditions that predispose to the development of carpal tunnel syndrome include pregnancy, diabetes, and obesity.

Is there any treatment?
Carpal tunnel syndrome is treated by immobilizing the wrist in a splint to minimize or prevent pressure on the nerves. If that fails, patients are sometimes given anti-inflammatory drugs or injections of cortisone in the wrist to reduce the swelling. There is also a surgical procedure in which doctors can open the wrist and cut the ligament at the bottom of the wrist to relieve the pressure. However, only a small percentage of patients require surgery.

What is the prognosis?

INSURANCE ISSUES

(1) Health Insurance

a. Risk Assessments or underwriting generally does not occur in providing health insurance to Large Groups / HMOs / Employer-based Plans — Genetic testing won't apply here —

b. State & federal Laws now Exist to prevent Discrimination using genetics in health insurance —

(1) US Health Insurance portability & Accountability Act of 1996 (HIPAA). Cannot use genetic information for employer-based & commercially-issued group health plans — Does not apply to private/individual health plans — individuals

(2) California Genetic Nondiscrimination Law — group health plans!

c. Risk Assessment/Genetic Testing can be required for Individual health insurance plan ∴ People could be denied coverage — need universal Health Insurance!

State Genetic Nondiscrimination in Health Insurance Laws

Last updated: 8/7/02

A patchwork of federal and state laws govern discrimination based on genetic information for health insurance. The 1945 McCarran-Ferguson Act explicitly endorses the primacy of state insurance regulation. The Employees Retirement Income Security Act of 1974 preempts state laws pertaining to self-funded employee benefits plans. The Health Insurance Portability and Accountability Act of 1996 became the first federal law to directly address genetic information. The law prohibits health insurance discrimination based on any "health status-related factor," including genetic information, for group health plans, usually those with more than 50 individuals.

States have acted to fill in the gaps left by HIPAA. Laws in 34 states strictly prohibit the use of genetic information for risk selection and risk classification purposes. Additionally, Arizona, Vermont, and West Virginia require actuarial justification for the use of genetic information. Texas bans use of genetic information in group health plans, and Alabama prohibits discrimination based upon predisposition to cancer.

State	Citation	Type of Insurance Policy	May not Establish Rules for Eligibility based on Genetic Information	May not Require Genetic Tests/Genetic Information	May not Use Genetic Information for Risk Selection or Risk Classification Purposes	May not Disclose Information Without Informed Consent	Components of Definition for Protected Genetic Information
California	Insurance Code: §§742.405, 7, 10140, 3, 6 to 9, 9.1	Individual and Group	√	√	√	√	GT, GF, IC

NOTES:
"GT" indicates individual genetic test results
"GF" indicates genetic test results of family members
"AC" indicates practices commonly accepted in scientific and medical communities
'FH" indicates family history
"IC" indicates inherited characteristics
"RP" indicates routine physical measurements
"CA" indicates standard chemical, blood, and urine analyses
"IM" indicates indirect manifestations of genetic disorders

INSURANCE ISSUES

(2) Life / Disability Insurance

a. State Laws Exist Dealing with genetic testing / genetic discrimination in Life insurance.

b. However — Risk Assessment ok / genetic tests ok in determining risk — right of insurers to obtain that information well established & can refuse &/or charge high rates, if risk high!

Genetics and Life, Disability and Long-term Care Insurance

Updated 10/14/02

While a majority of states have enacted laws that strictly prohibit the use of genetic information for risk selection and risk classification in health insurance, fewer states restrict the use of genetic information in life, disability and long-term care insurance. Six states prohibit genetic discrimination in life and disability insurance without actuarial justification. Of these six, Arizona, Maine and New Jersey also prohibit genetic discrimination in disability insurance without actuarial justification, and Montana and New Mexico extend their prohibitions to disability and long-term care insurance. Seventeen states restrict insurer use of genetic information in life, disability or long-term care insurance in some manner. Other states mention life, disability or long-term care as exclusions to their genetic nondiscrimination legislation.

State and Statutes	Restricts Discrimination Based on Genetic Information in Life Insurance	Restricts Discrimination Based on Genetic Information in Disability Insurance	Restricts Discrimination Based on Genetic Information in Long-term Care Insurance	Requires Actuarial Justification to Use Genetic Information in Life Insurance	Requires Informed Consent to Use Genetic Information
California Insurance €€10146 to 10149.1	Ã	Ã			Ã[1]

NOTES:

***** Arizona, Maine and New Jersey also prohibit genetic discrimination in disability insurance without actuarial justification. Montana and New Mexico extend their prohibitions to include disability and long-term care insurance.
1 Can only require a person to undergo a genetic test unless the cost of the test is paid by the insurer.
2 Applies only to group disability and long-term care insurance.
3 Applies only to "sickle-cell trait, thalassemia-minor trait, hemoglobin C trait, Tay-Sachs trait, or a genetic trait that is harmless in itself."
4 No life insurance company shall refuse to issue or deliver life insurance or charge a higher rate solely because of possession of sickle cell trait or hemoglobin C trait.
5 Must notify individual that genetic test may be used.
6 No insurer shall refuse to issue, fail to deliver, or charge a higher rate solely because a person has the sickle-cell trait.

California, AB2797, approved: Provides that a person or entity that underwritesor sells annuity contracts or contracts insuring, guaranteeing, orindemnifying against loss, harm, damage, illness, disability, or death, and any affiliate of that person or entity, shall not disclose individually identifiable information concerning the health of, or the medical or genetic history of, a customer, as specified, for use with regard to the granting of credit. Because a violation of the bill's provisions would be a crime, this bill imposes a state-mandated local program by creating a new crime. The California Constitution requires the state to reimburse local agencies and school districts for certain costs mandated by the state. Statutory provisions establish procedures for making that reimbursement. This bill provides that no reimbursement is required by this act for a specified reason.

CALIFORNIA Genetics Life Insurance Law

INSURANCE CODE SECTION 10146-10149.1

10146. The purposes of this article are to establish standards regarding unfair discrimination among individuals of the same class in the underwriting of life or disability income insurance on the basis of tests of a person's genetic characteristics; to establish minimum standards for determining insurability which are sufficiently reliable to be used for life and disability income insurance risk classification and underwriting purposes; to require the maintenance of strict confidentiality of personal information obtained through a test of a person's genetic characteristics; and to require informed consent before insurers underwrite on the basis of a test of a person's genetic characteristics. This article and Sections 10140 and 10143 shall constitute the exclusive requirements for insurers' practices relating to genetic characteristics or to tests thereof.

10147. As used in this article:

(a) "Disability income insurance" means insurance against loss of occupational earning capacity arising from injury, sickness, or disablement, and includes insurance which provides benefits for overhead expenses of a business or profession when the insured becomes disabled.

(b) "Genetic characteristics" means any scientifically or medically identifiable gene or chromosome, or alteration thereof, that is known to be a cause of a disease or disorder, or that is determined to be associated with a statistically increased risk of development of a disease or disorder, and that is presently not associated with any symptoms of any disease or disorder.

(c) "Life or disability income insurer" means an insurer licensed to transact life insurance or disability income insurance in this state or a fraternal benefit society licensed in this state.

(d) "Policy" means (1) a life insurance policy or a disability income insurance policy delivered in this state, or (2) a certificate of life insurance benefits or disability income insurance benefits, issued under a group life or disability income insurance policy and delivered in this state by a life or disability income insurer or a fraternal benefits society, regardless of the location of the group master policy.

(e) "Test of a person's genetic characteristics" means a laboratory test which is generally accepted in the scientific and medical communities for the determination of the presence or absence of genetic characteristics.

10148. No insurer shall require a test for the presence of a genetic characteristic for the purpose of determining insurability other than for those policies that are contingent on review or testing for other diseases or medical conditions. In those cases, the test shall be done in accordance with the informed consent and privacy protection provisions of this article and Article 6.6 (commencing with Section 791) of Chapter 1 of Part 2 of Division 1. Notwithstanding any other provision of law, this constitutes the exclusive requirements for informed consent and privacy protection for that testing.

(a) An insurer that requests an applicant to take a genetic characteristic test shall obtain the applicant's written informed consent for the test. Written informed consent shall include a description of the test to be performed, including its purpose, potential uses, and limitations, the meaning of its results, procedures for notifying the applicant of the results, and the right to confidential treatment of the results.

(b) The insurer shall notify an applicant of a test result by notifying the applicant or the applicant's designated physician. If the applicant tested has not given written consent authorizing a physician to receive the test results, the applicant shall be urged, at the time the applicant is informed of the test results, to contact a health care professional.

(c) The commissioner shall develop and adopt standardized language for the informed consent disclosure form required by this section to be given to any applicant for life or disability income insurance who takes a test for a genetic characteristic.

(d) A life or disability income insurer shall not require a person to undergo a test of the person's genetic characteristics unless the cost of the test is paid by the insurer.

(e) No policy shall limit benefits otherwise payable if loss is caused or contributed to by the presence or absence of genetic characteristics, except to the extent and in the same fashion as the insurer limits coverage for loss caused or contributed to by other medical conditions presenting an increased degree of risk.

(f) Nothing in this chapter shall limit an insurer's right to decline an application or enrollment request for a life or disability income insurance policy, charge a higher rate or premium for such a policy, or place a limitation on coverage under such a policy, on the basis of manifestations of any disease or disorder.

(g) No discrimination shall be made in the fees or commissions of agents or brokers writing or renewing a life or disability income policy on the basis of a test of that person's genetic characteristics.

10149. (a) All underwriting activities undertaken by insurers pursuant to this article shall be subject to all applicable provisions of Article 6.6 (commencing with Section 791) of Chapter 1 of Part 2 of Division 1.

(b) No life or disability income insurer shall require a genetic characteristic test if the results of the test would be used exclusively or nonexclusively for the purpose of determining eligibility for hospital, medical, or surgical insurance coverage or eligibility for coverage under a nonprofit hospital service plan or health care service plan.

10149.1. (a) This section shall apply to the disclosure of the results of a test for a genetic characteristic requested by an insurer pursuant to this article.

(b) Any person who negligently discloses results of a test for a genetic characteristic to any third party, in a manner which identifies or provides identifying characteristics of the person to whom the test results apply, except pursuant to a written authorization, as described in subdivision (g), or except as provided in this article or in Sections 1603.1 and 1603.3 of the Health and Safety Code, shall be assessed a civil penalty in an amount not to exceed one thousand dollars ($1,000) plus court costs, as determined by the court, which penalty and costs shall be paid to the subject of the test.

(c) Any person who willfully discloses the results of a test for a genetic characteristic to any third party, in a manner which identifies or provides identifying characteristics of the person to whom the test results apply, except pursuant to a written authorization, as described in subdivision (g), or except as provided in this article or in Sections 1603.1 and 1603.3 of the Health and Safety Code, shall be assessed a civil penalty in an amount not less than one thousand dollars ($1,000) and no more than five thousand dollars ($5,000) plus court costs, as determined by the court, which penalty and costs shall be paid to the subject of the test.

(d) Any person who willfully or negligently discloses the results of a test for a genetic characteristic to a third party, in a manner which identifies or provides identifying characteristics of the person to whom the test results apply, except pursuant to a written authorization, as described in subdivision (g), or except as provided in this article or in Sections 1603.1 and 1603.3 of the Health and Safety Code, which results in economic, bodily, or emotional harm to the subject of the test, is guilty of a misdemeanor punishable by imprisonment in a county jail for a period not to exceed one year, by a fine of not to exceed ten thousand dollars ($10,000), or by both that fine and imprisonment.

(e) Any person who commits any act described in subdivision (b) or (c) shall be liable to the subject for all actual damages, including damages for economic, bodily, or emotional harm which is proximately caused by the act.

(f) Each disclosure made in violation of this section is a separate and actionable offense.

(g) The applicant's "written authorization," as used in this section, applies only to the disclosure of test results by a person responsible for the care and treatment of the person subject to the test. Written authorization is required for each separate disclosure of the test results, and shall include to whom the disclosure would be made.

NOTE - POLICY CAN BE TURNED DOWN - NOT AUTOMATIC!

LEGAL ISSUES in LAW Enforcement

(1) DNA Fingerprinting

a. Case Law / Frye v Daubert → Use Scientific Method in testing & Witness Expertise

b. Constitution –

No obstacles to suspects in criminal case

(1) Blood or voice samples are not "testimonials" & not protected by 5th Amendment Protection Against Self-incrimination.

(2) If probable cause – Search warrant can compel person to give DNA sample consistent with 4th Amendment!

(2) Data BANKS

a. Convicted felons can be compelled to give DNA for FBI & State data banks! And data banks exist!

b. What about suspects?

c. What about everyone?!

The *Daubert* Worldview

"To summarize: 'general acceptance' is not a necessary precondition to the admissibility of scientific evidence under the Federal Rules of Evidence, but the Rules of Evidence -- especially Rule 702 -- do assign to the trial judge the task of ensuring that an expert's testimony both rests on a reliable foundation and is relevant to the task at hand."

-- Blackmun, J., in Daubert v. Merrell Dow Pharmaceuticals, Inc., 509 U.S. 579 (1993)

Chapter 2: *Daubert* in a Nutshell

The Supreme Court's decision in *Daubert* lends itself to brisk summary.

For many years, the admissibility of expert scientific evidence was governed by a common law rule of thumb known as the *Frye* test, after a 1923 decision by the District of Columbia Court of Appeals in which it was first articulated. Under the *Frye* test, expert scientific evidence was admissible only if the principles on which it was based had gained "general acceptance" in the scientific community.

Despite its widespread adoption by the courts, this "general acceptance" standard was viewed by many as unduly restrictive, because it sometimes operated to bar testimony based on intellectually credible but somewhat novel scientific approaches.

In *Daubert*, the Supreme Court was asked to decide whether the *Frye* test had been superceded by the adoption, in 1973, of the *Federal Rules of Evidence*. After all, Fed. R. Evid. 702, the rule broadly governing the admissibility of expert testimony, did not even *mention* "general acceptance," but simply provided: "If scientific, technical, or other specialized knowledge will assist the trier of fact to understand the evidence or to determine a fact in issue, a witness qualified as an expert by knowledge, skill, experience, training, or education, may testify thereto in the form of an opinion or otherwise."

The majority opinion in *Daubert*, authored by Justice Blackmun, held that Rule 702 did indeed supplant *Frye*. This did not mean, however, that all expert testimony purporting to be scientific was now to be admissible without further ado. Rule 702 did require, after all, that the testimony actually *be* founded on "scientific knowledge." This implied, according to the Court, that the testimony must be grounded in the *methods and procedures* of science -- a.k.a. "the scientific method." Evidence thus grounded, said the Court, would possess the requisite *scientific validity* to establish *evidentiary reliability*.

The Court also noted Rule 702's requirement that expert testimony *assist the trier of fact*. This, according to *Daubert*, was primarily a question of *relevance* or "fit." The testimony must be sufficiently tied to the facts of the case, the Court held, to aid in the resolution of an issue in dispute.

The Court explicitly refused to adopt any "definitive checklist or test" for determining the reliability of expert scientific testimony, and emphasized the need for flexibility. The Court did list several factors, however, that it thought would commonly be pertinent:

- whether the theories and techniques employed by the scientific expert have been *tested*;
- whether they have been subjected to *peer review* and *publication*;
- whether the techniques employed by the expert have a *known error rate*;
- whether they are subject to *standards* governing their application; and
- whether the theories and techniques employed by the expert enjoy widespread acceptance.

By way of offering further guidance, the Court emphasized that the admissibility inquiry must focus "solely" on the expert's "principles and methodology," and "not on the conclusions that they generate."

To assuage fears that its ruling would result in a "free for all" in which juries would be confounded by "absurd and irrational pseudoscientific assertions," the Court emphasized the continued availability of traditional tools under the adversary system, including vigorous cross-examination, presentation of contrary evidence, and careful instructions to jurors on burdens of proof. The Court also noted the availability of other mechanisms of judicial control, including summary judgment and the ability to exclude confusing or prejudicial evidence under Fed. R. Evid. 403.

In response to the fear that its new evidentiary standards would sometimes stifle courtroom debate, the Court acknowledged that those standards would occasionally prevent juries from "learning of authentic insights and innovations," but concluded that such was the inevitable consequence of evidentiary rules "designed not for the exhaustive search for cosmic understanding but for the particularized resolution of legal disputes."

All of that is straightforward enough. Right?

The FBI's national DNA database

Russ Hoyle

The US Federal Bureau of Investigation's (FBI; Washington, DC) new national DNA database was introduced amid much fanfare by FBI Director Louis Freeh in Washington on October 13. The new database, declared Freeh, should provide "signifcant crime prevention benefit as this new DNA program identifies serial offenders who might otherwise escape detection for their repeat crimes." The new DNA program, added FBI lab director Dr. Donald Kerr, will "allow this exciting technology to reach its full potential in solving violent crimes through nationwide information sharing. . . ."

Though the FBI has operated a state and local DNA database in 41 states and the District of Columbia since 1991, the new National DNA Index System (NDIS) will serve for the first time as a repository for hundreds of thousands of DNA profiles of convicted criminals in all 50 states. NDIS profiles will be accessible by police and law enforcement laboratories across the country, allowing speedy tracking of individuals convicted of felony sex offenses and other violent crimes, as well as of crime-scene evidence such as blood, semen stains, or hair.

So far, the seven-year old US system, and a more advanced version in Great Britain, have helped solve an impressive number of crimes by linking crime scenes and identifying criminals, even in cases in which no suspects had been identified. In the US, state and local FBI databases have already produced more than 400 such matches. To date, the states have collected some 600,000 DNA samples and analyzed, or profiled, more than 250,000.

For now, the FBI is not saying what constitutes the "full potential" of the new database—or even how long it will take to become fully operational. One reason is simple enough: The enabling federal legislation, the DNA Identification Act of 1994, sharply circumscribes its lawful uses. To accomodate strict constitutional guidelines for privacy, confidentiality, and lawful search and seizure, the FBI index can only collect genetic information on convicted criminals, crime scenes, and unidentified human remains.

Theoretically, at least, that means the FBI cannot keep a DNA sample or profile from this columnist, or you, or President Clinton, unless we are convicted of crimes. It also means that police or federal agents cannot collect DNA samples from suspects nor even from indicted, not-yet-convicted felons—including terrorists—for investigative purposes. In addition, DNA law also sharply limits DNA identification technology to 13 basic probes that can isolate genetic characteristics, but are unable to provide fuller details of identity, such as hair, eye or skin color.

Though FBI officials will not say it outright, it is likely that as US law enforcement officials gain experience with DNA fingerprinting, monitor the arrest records of less constitutionally constrained police abroad, and track the inevitable advance of DNA identification technology, the 1994 law will rapidly become outdated and need modification. The betting here is that any future amendment of the law will significantly expand the segment of the population from whom DNA samples may be collected and will bring to bear increasingly sophisticated DNA identification technology.

All of this is good news for the biotechnology community. Even now, technical advances, such as phenotypic analysis, in which DNA markers may be used to provide identifying physical, psychological or medical characteristics, are being hotly debated as the wave of the future in criminological circles. In the meantime, because of staffing shortfalls in state and local labs, the massive backlog of offender samples that must be analyzed and profiled, not to mention new biological evidence coming in the door, the development of a full-blown national DNA database is likely to take several years. In that time, the system may well be overtaken by procedural and technological advances.

The natural inclination of law enforcement officials to press for wider latitude in applying DNA identification technology, however, has alerted legal watchdogs and ethicists to possible problems presented by the gathering, storing and utilization of genetic data on criminals. In response, the FBI formed a DNA Advisory Group to oversee establishment of the new database.

Legal challenges have been mounted in 13 of the 50 states aimed at the laws establishing DNA databases, mostly on Fourth Amendment grounds—and defeated in all but one. In Massachusetts, a lower court held that the state database law would allow effectively allow unconstitutional search and seizure of bodily substances. "A bodily intrusion with or without the use of force," wrote the presiding judge, "can only be considered reasonable if probable cause exists to believe the person in question participated in the criminal act for which a. . .sample is relevant evidence."

The decision is being appealed. Nonetheless ethicists have seized on the issue at the heart of the case: Where is the line between coercing citizens to give up bodily tissues that may incriminate them and somehow compelling them to volunteer those tissues legally?

Already, FBI guidelines on the scope of genetic testing have been broadened to include a separate category for juvenile offenders—along with violent felons, burglars, and convicted criminals on parole or probation. Why are juveniles singled out? According to FBI officials, because juvenile crime is increasingly violent, genetic testing might nip criminal careers in the bud, and experience so far has shown that DNA testing has worked well to curb youth crime.

Knotty issues of privacy and confidentiality are likely to continue to plague the stockpiling of genetic data and tissue samples. In fairness, the FBI new national database will store only limited genetic profiles, not samples. But before the system is up and running, simple genetic analysis of that huge backlog of biological samples—which will only increase as time passes—will require storehousing samples in labs across the country.

Though the use of tissue samples for other purposes is forbidden in most states, ethicists point out that pressure not to destroy samples may be considerable, especially from scientific researchers. Indeed, they say exceptions that allow scientific and medical research are common in current state genetic privacy laws. Moreover, from a researcher's perspective, destruction of such a well-defined body of biological samples is "a tremendous waste," as one ethicist concedes.

Still, what happens to samples or data in cases where juveniles records are erased, as happens in many states? What happens when genetic material from a deceased person becomes is requested for an unanticipated purpose, such as genetic research? How do researchers isolate genes indicating a predisposition, say, to criminal behavior without the best available data?

By law, of course, none of this is supposed to happen. For now at least, the federal law enforcement community has fashioned a judiciously circumscribed first step toward a national DNA database that will identify criminals and match them to their crimes. The question of course is whether such a national network will be able to deflect pressures in the future to abuse this powerful new tool in the name of expanding DNA-based law-enforcement strategies. ///

The promise and perils of criminal DNA databanking

Jonathan Kimmelman

Embryonic stem cells, GM foods, microbe patents, and cloning have all been greeted with some measure of public opprobrium. Not so with DNA profiling technologies. Save philandering presidents and descendants of Louis XVII, nearly everyone has something good to say about them. They've vindicated the Left's assaults on capital punishment and freed over 60 death row inmates. They've fortified the cause of law and order by solving countless criminal investigations and arming prosecutors with irrefutable evidence, and they've provided many rape victims the small solace of knowing their assailants have been (or may be soon) brought to justice. But behind this nearly universal mist of euphoria dwell important ethical deficiencies in how DNA samples are collected and processed in the operation of criminal DNA databanks. With New York State, which operates one of the largest criminal justice systems in the US, proposing massive renovations in its DNA databanking policy, these concerns are especially pertinent.

DNA databanks provide a function similar to fingerprint indices, in that they allow investigators to compare genetic profiles recovered from crime scenes with those taken from convicted individuals. But facile analogies to fingerprints obscure critical differences that warrant ethical solicitude[1]. First, DNA is far more information-rich than fingerprints; it contains the cipher of a person's hereditary propensities and susceptibilities, parentage, and racial origins. Second, DNA is a more dependable informant, in that it is durable, amplifiable from minute quantities, and recoverable from the skin particles, hair roots, and finger smudges we leave unwittingly in our wake. Third, genetic information is, unlike fingerprints, shared among biological relatives. As a result, the decision to place in a databank one person's DNA profile is an indirect decision to do the same with the half-profiles of that person's biological siblings and parents. Collectively, these differences raise concerns with respect to criminal DNA databanking's expanding ambit, irresolute storage policies, and questionable authorizations for how banked DNA samples can be used. A recent survey of US and foreign criminal DNA databanking laws indicates that whereas databanks have been extending themselves into uncharted ethical areas, they have also failed to repair deficiencies present since their inception (J. Kimmelman, unpublished data).

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Mergers and acquisitions

The earliest DNA databanking statutes enacted in the US restricted themselves to persons convicted of sexual offenses[2,3]; not long after, databanking laws were extended to other violent offenses, including murder and crimes against children. Because these crimes are severe, prone to high rates of recidivism, and likely to involve incriminating deposition of DNA evidence, such laws make for sound public policy. Within a relatively short period, however, databanks have since waded into more turbid waters, at least from a social standpoint. According to Table 1, 17 states (34%) presently cover certain categories of property offenses (up from 17% of states with databanking laws in 1994), 23 (or 46%) states cover certain misdemeanors (up from 39% in 1994), 7 states cover consensual sodomy, and 6 states cover all felonies, which include crimes like perjury, forgery, larceny, tampering, and credit card fraud, where DNA evidence is unlikely to have a high degree of probity. The latter policy appears to be the leading edge of a trend in US databanking policy: four states are considering bills in their current legislative sessions to extend their DNA databanks to all felons, and US Federal Bureau of Investigation's (Washington, DC) criminal DNA indexing program director, Steve Niezgoda, has predicted that felony databanking will be a fait accompli within a decade[4].

Table 1. US states criminal offender DNA databanking policies

Policy	Number of states
Collect DNA from individuals convicted of	
Sex crimes (e.g. rape, sexual assault)	50
Crimes against persons (e.g. murder)	38
Crimes against property (e.g. burglary)	17
Any felony	6
Certain misdemeanors	23
Consensual, nonspousal sodomy	7
Collect DNA from categories of juvenile offenders	26
Expungement when convictions are overturned	
Requires petitioning	29
Automatic	5
Authorize use of	
Records for forensic research	18
Samples for forensic research	5
Policy on structural gene information	
Inclusion in records prohibited	6
Use of samples for such analysis prohibited	2
Use of samples for such analysis clearly authorized	1
Require or authorize that DNA samples be	
Stored in a repository	29
Destroyed after profiling	1

Others are calling for yet more aggressive measures. New York City police chief Howard Safir has energetically advocated a policy of felony arrestee sampling[5]—similar to the UK's (see below)—and North Carolina has actually proposed such legislation. To date, however, only one state (Louisiana) has made the leap from convicted persons to arrestees, but only for individuals suspected of violent sexual felonies[6].

A parallel trend is the penetration of DNA databanking into the juvenile justice system. Where 9 out of 23 states collected DNA profiles from certain categories of juvenile offenders in 1994, today the number stands at 26. These policies reverse a tradition in US law enforcement of not collecting permanent criminal records from juveniles, a practice founded on the goal of rehabilitation. Moreover, this policy further risks inadvertently magnifying racial disparities in the juvenile justice system; since minority youth are disproportionately convicted on a per crime basis[7,8], databanks would be more effective at solving crimes committed by minorities.

Liquid assets

While databanks have expanded into larger populations, they have meanwhile failed to clarify their policies and vision concerning what law enforcement agencies can do with the DNA samples. On the positive side of the balance sheet, federal laws mandate penalties for disclosures of genetic information to unauthorized parties like employers or insurers[9], and 28 states have also added penalties as well to ensure that such sensitive information doesn't fall into the wrong hands. Additionally, two states prohibit any testing of databanked samples for structural genes.

On the negative side, however, 29 states either authorize or require that samples be retained in a repository, whereas 23 states (directly or indirectly) authorize release of samples or records for research uses that would assist law enforcement; one state actually authorizes using anonymous profiles outside the law enforcement context, in medical research[10]. These provisions are particularly troubling. Retaining samples sustains the possibility that they will find ethically problematic uses in the future; authorizing research on samples, even if they are stripped of individual identifiers (as mandated by most laws) nearly delivers them to this unseemly fate. One needn't be a behavioral geneticist to appreciate the promise offered by offender DNA repositories for those interested in the genetics of violence, sexual deviance, or recidivism (one Massachusetts lawmaker, in fact, has endorsed these aims[11]).

Although many potential benefits of such research can be envisioned (e.g., determining suitable treatment regimens for particular prisoners), transferring databanked DNA for research protocols would violate the right of research subjects to opt out of participating in potentially controversial medical research; it would also run counter to the guidelines on handling genetic materials proposed by several commentators, including the American College of Medical Genetics (Bethesda, MD)[1,12].

If states are retaining samples because of concerns that evolving profiling technologies will outmode current profiles, they should articulate criteria for determining when technologies are sufficiently stable to allow for sample destruction. In the meantime, states should specify the fate of samples from deceased convicts and require that consent be obtained from prisoners before their samples are made anonymous or released for research.

Foreign currencies

Foreign DNA databanking practices offer contrasts that highlight the strengths and weaknesses of US policies. In addition to the US, Austria, Australia[13], Canada[14], UK[15], France[16], Germany, the Netherlands, and Switzerland[17] all operate similar databanks. By far, the UK's is the most comprehensive program. Authorized in 1994, UK laws allow police to collect samples from any individuals suspected of (but not necessarily convicted of or detained for) any "recordable offenses"[18]. Authorities in the UK project that the profiles and samples of one-third of the UK male population will eventually be databanked[19]. Additionally, UK police are authorized to collect samples from the general population provided consent is given—a practice often called "sweeping." The possibilities for abuse in such a system are extensive, and the fact that failure to consent could invoke police suspicion challenges the notion that consent is truly voluntary. Although genetic sweeps have been conducted in Australia[20], Canada[21], and Germany[17], none has been conducted in the US, where Fourth Amendment protections would probably prevent these searches without reasonable suspicion.

At the opposite extreme, France appears to be embarking on the world's most cautious venture into databanking. According to newspaper accounts[16], French police are authorized only to collect samples from convicted sexual offenders and are required to destroy all samples after 40 years or whenever an offender reaches the age of 80.

Positioned somewhere in between the two, Canada's policy offers a system that better protects the privacy and social interests threatened by US policy. Canada collects samples from all individuals convicted of serious violent offenses and leaves sample collection to the discretion of magistrates for serious nonviolent offenses such as robbery. In addition, Canada expunges all DNA and profiles of individuals whose convictions are overturned (29 US states place the onus of protecting privacy on the wrongly convicted, who must petition to have their records expunged), and forbids uses for collected DNA other than for forensic profiling. Finally, profiles of juveniles are eliminated after a specified period, thus preserving for youths the possibility of wiping their sullied reputations clean and beginning anew.

Auditing the databanks

The net result of US policies, as they stand now, is that many people are providing DNA to law enforcement agencies that are not required to apply stringent protections against their misuse. Because genetic profiles are shared with biological relatives, law enforcement agencies are furthermore collecting information by proxy from offenders' relatives. Some commentators, including New York City's mayor Rudolph Giuliani, have anticipated this trend's logical conclusion by entertaining the notion of databanking the DNA of all newborns[22].

Why should this concern anyone? The claim is frequently made, for example, that "only the guilty have something to fear." Although the negative perceptions of police forces by many members of racial minorities undermine the force of this argument, one needn't invoke nefarious motives of police forces to find such broad-based databanking socially questionable. Storing information on otherwise unsuspected individuals that would be primarily used for criminal investigations in effect expresses an ethos of suspicion. Although such defensive policing might deter some crimes and solve others, it nevertheless creates a chilling dynamic between the government and its citizens, and undermines the long-standing legal tradition in the US of presumptive innocence. Moreover, such information gathering by proxy is nearly unprecedented: police do not collect fingerprints from persons who are not suspected of particular crimes.

If the diminishing costs and increasing speed of gene sequencing are any prelude, the most formidable barriers constraining DNA databanking practices will soon be swept away. The refrigerated vaults of law enforcement agencies will then be awash in bloodstains and buccal swabs. But the obvious law enforcement benefits of DNA databanking shouldn't diminish our sensitivity to the values at stake: privacy and presumptive innocence. When the technological and legislative levees break, let's hope the policymakers and law enforcement agencies have found moral high ground.

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Nation

DNA Tests for Inmates Debated

A St. Louis prosecutor wants to keep the guilty from reopening old wounds. Others want to 'cut to the truth.'

By STEPHANIE SIMON
Times Staff Writer

ST. LOUIS — Prosecutor Jennifer Joyce knows the horror of imprisoning an innocent man.

She freed a convicted rapist last summer when genetic analysis of old evidence exonerated him after he spent nearly 18 years in prison. She understands the power of such testing to right wrongs.

But Joyce also is convinced that DNA analysis can be abused.

In the last decade, more than 100 inmates across the nation — including at least 13 on death row — have been freed after DNA tests cleared them. Sen. Patrick J. Leahy (D-Vt.) plans to introduce a bill in Congress this session that would expand access to such tests.

He wants the federal government to recognize that inmates have a constitutional right to access the biological evidence against them.

That proposition has Joyce up in arms. From her post as circuit attorney of St. Louis, she is waging a passionate — and for the most part lonely — battle to restrict rather than broaden inmates' rights to DNA tests. A few fellow prosecutors back her. Others whisper that she's crazy to spend so much energy on an issue they view as a nuisance at most.

To Joyce, it is no mere nuisance.

Twice last month, DNA tests at the police crime lab in St. Louis confirmed the guilt of convicted rapists. Two other tests, last year and in 2001, also showed the right men were behind bars for brutal rapes committed a decade or more earlier.

Joyce's staff spent scores of hours and thousands of dollars on those tests. She personally counseled shaking, sobbing victims who were distraught to learn that their traumas were being aired again.

One victim, she said, became suicidal and then vanished; her family has not heard from her for months. Another, a deaf elderly woman, grew so despondent that her son has not been able to tell her the results of the DNA test. Every time he raises the issue, she squeezes her eyes shut so she will not be able to read his lips.

"She finally seemed to have some peace about the rape, and now she's gone back to being angry," the woman's son said.

Associated Press

'Maybe we need to go through all this to find the one innocent person out there. But when these guys know good and well that they committed the crime, they're just being sadistic in requesting the tests. We have to have some provision that will make them think twice.'

Jennifer Joyce, circuit attorney of St. Louis

DNA tests confirmed that she was raped by Kenneth Charron in 1985, when she was 59. To get that confirmation, however, investigators had to collect a swab of saliva from her so that they could analyze her DNA. They also had to inquire about her sexual past, so they could be sure the semen found in her home was not that of a consensual partner.

The questioning sent the woman into such depression that she's now on medication. "None of this needed to happen," her son said.

Joyce agrees. So she is drafting a bill — apparently the first in the nation — to deter "frivolous" DNA tests.

She wants inmates to pay for the analysis, which can run up to $2,500, unless they are exonerated. She wants probation and parole boards to consider an inmate's request for a test as a black mark.

And she wants to use an existing statute barring frivolous lawsuits to add 60 days to an inmate's sentence if the requested DNA test ends up confirming guilt.

"Maybe we need to go through all this to find the one innocent person out there," Joyce said. "But when these guys know good and well that they committed the crime, they're just being sadistic in requesting the tests. We have to have some provision that will make them think twice."

In the meantime, Joyce is refusing to test DNA from two other old sexual assaults, insisting that the analysis would not prove guilt or innocence, given the circumstances of the cases. Lawyers for the inmates involved are seeking to charge Joyce with contempt of court for withholding the biological evidence. A hearing is scheduled for this month. The circuit attorney refuses to yield.

Associated Press

GUILTY: *DNA tests recently confirmed Kenneth Charron raped a woman in 1985.*

"I don't want to set the precedent " of testing DNA in every case, she said. "We don't have the resources for that."

Under Missouri law, a convict can get biological evidence tested at state expense if there is a "reasonable probability" that the results would have affected the case had they been available at trial. California has a similar law. In all, 28 states permit convicts to petition a judge for DNA analysis, although some restrict that right to death row inmates and others require that the prisoner pay for the test.

Even in the states with laws that make it easier for inmates to get tests, there is little evidence that they are demanding DNA tests in droves. Public defenders in California, Illinois, New York and several other states report just a handful of such cases over the last several years.

Many of the inmates who would like to request testing find that the evidence from their case file has long since been destroyed. Judges reject other pleas because DNA analysis would shed little light on a case. And some convicts withdraw their requests when they realize that their DNA will be entered into databanks and compared against biological evidence left at thousands of unsolved crime scenes.

"It's not a flood of people requesting this," said Vanessa Potkin, an attorney with the Innocence Project, a nonprofit legal clinic in New York that has led the drive for post-conviction DNA tests.

The Innocence Project screens inmate petitions, selecting only the cases that seem to offer the best shot at exoneration. Still, Potkin said, 60% of the inmates represented by the clinic prove to be guilty when the results come in.

Why they demand DNA tests when they know they committed the crime "would be the subject for a great psychological study," Potkin said. "Maybe after 15 years of telling everyone you're innocent, you start to believe yourself."

Whatever the reason, Joyce and a few other prosecutors argue that such frivolous requests — even if there are just a dozen a year — present a major burden.

"If we had unlimited resources, you might say, 'So, there are a couple hundred more people who want DNA testing. What's the harm?' " said Joshua Marquis, the district attorney in Astoria, Ore.

"But there are 500,000 rape kits [containing evidence] sitting on the shelves of police stations across the country right now, untested because we don't have the resources."

DNA labs everywhere are strained to the breaking point; a survey released last month by the U.S. Justice Department found that 81% of crime labs have fallen well behind in their work. The backlog included more than 16,000 criminal cases, which would take about eight months to work through if not a single additional test request came in.

Still, Potkin argues, it is a moral imperative to test DNA if it could prove that an inmate was wrongly convicted. She calls Joyce's resistance a "violation of due process," adding that the cases she's working in St. Louis have taken longer than just about anywhere else.

"We're not fortunetellers, and neither is she," Potkin said.

"We don't know if these guys are innocent. But there's a scientific test out there that can cut to the truth of the situation. Until we do the test, there's no way to know."

The New York Times
nytimes.com

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Police Dragnets for DNA Tests Draw Criticism

By DAVID M. HALBFINGER

BATON ROUGE, La., Jan. 3 Ñ Recently, the police asked Shannon F. Kohler if they could swab the inside of his mouth to analyze his DNA. It was a request they made of 800 men in southern Louisiana as they searched for the serial killer who has slain four young women, leaving behind genetic material in each case.

It was his choice, Mr. Kohler said the officers told him, but if he refused, they would get a court order and that would get in the newspapers and then everyone would know he was not cooperating. The approach was heavy-handed and foolish, he said, especially since he has feet much bigger than the prints left by the killer and had phone bills that show he was at home when the murders took place.

The questions Mr. Kohler is raising about DNA testing are also being asked by lawyers and other experts around the country who say the growing use of DNA dragnets like the one here, already one of the largest in American history, is troubling.

The tests, supposedly voluntary, can still be coercive, critics say, not only harassing innocent people but also potentially violating suspects' constitutional protections against compelled self-incrimination and unreasonable search and seizure. Future prosecutions could be undermined, some legal scholars, defense lawyers and even some prosecutors say. Some question whether the dragnets' limited success justifies the effort and expense. And even those who endorse the idea of DNA sweeps argue over whether Ñ and why Ñ the government should keep on file the genetic profiles of those who are proved to be innocent.

The tests trouble some for the very reason that police find them attractive: they offer the most incontrovertible proof of identity.

The idea for a DNA dragnet Ñ sampling people who are not suspects but merely live or work near a crime scene Ñ emerged in Britain. In 1987, the police tested 4,000 men in Leicestershire before the rapist and killer of two girls was caught after he got another man to take the DNA test for him. One of the first dragnets in which DNA actually identified a killer was in Wales; a neighbor of a slain rape victim was caught in a DNA sweep of 2,000 men.

By 1998, dragnets had taken hold in northern Germany, where 16,400 people were tested Ñ believed to be the most yet Ñ before a mechanic was matched to a rape-murder.

In the United States, mass screenings have had less success and stirred up far more controversy. In 1994 and 1995, the Metro-Dade police in the Miami suburbs took more than 2,000 DNA samples in search of the strangler of six prostitutes, and initially focused on three possible matches before each man was ruled out. Still, the killer was caught only after neighbors found a prostitute bound and gagged in his apartment while he appeared in court on an unrelated robbery charge.

In 1998, the police in Prince George's County, Md., sought DNA samples from 400 male workers at a county hospital where an administrator had been raped and strangled. Union members complained that the police were bullying employees into agreeing and were singling out maintenance workers. No match was made, and the killing remains unsolved.

The chief of the county's police force at the time, John Farrell, defended the DNA tests to USA Today in 1998 as analogous to fingerprinting everyone who worked or shopped in a store that was burglarized, to eliminate potential suspects as well as to catch the criminal.

But mass fingerprint gathering is all but unheard of in criminal cases, said James Alan Fox, a professor of criminal justice at Northeastern University, precisely because of the probability that a print obtained from a crime scene will turn out to be someone's other than the criminal's.

DNA is different, Professor Fox said, which accounts for its allure: "If you have a rape and murder, and there's semen recovered, it's highly unlikely that it was innocently left there."

Not surprisingly, DNA screenings have been much more successful, if no less provocative, when the police have narrowed their focus to smaller groups Ñ generally those with opportunity, if not motive.

In Lawrence, Mass., in 1999, the police drew blood from 32 men at a nursing home where a resident had been raped and impregnated. A nurse's aide was linked to the crime and pleaded guilty. In Los Angeles that year, detectives who reopened the case of a 1985 killing of a sheriff's deputy set about sampling 165 potential suspects. They had finished 12 when a former colleague of the victim refused to comply; detectives won a court order, matched his DNA to the crime, and were about to arrest him when he killed himself.

Professor Fox, an expert on serial killers who wrote a book on the murders of five University of Florida students in Gainesville in 1990, said investigators in that case, with whom he worked as a consultant, checked the DNA of hundreds of people identified as possible suspects, often surreptitiously.

"We'd follow people as they went through Burger King, and pick up a straw they used, for saliva," he said. "We'd go through their trash on the sidewalk. Not everybody we got DNA on even knew it."

The police were far less quiet about their DNA testing in Ann Arbor, Mich., in 1994, after 13 women in a predominantly white community were raped by a black man. Investigators identified more than 700 suspects and took 160 DNA samples from black men, relying on tips that often proved specious.

The strategy caused a racial furor, with blacks saying they were being randomly singled out, and the rapist was caught only after a cab driver spotted him with blood on his clothes.

Some legal experts are now calling for an even more controversial use of genetic forensics: a national databank of DNA taken from every American at birth, solely for the purpose of criminal identifications.

Michael E. Smith, a University of Wisconsin law professor who led a working group for the National Commission on the Future of DNA Evidence, said such a databank would remove the danger of racial discrimination in DNA testing, as well as the risk that law enforcement agents seeking genetic information would turn to hospitals and medical laboratories, eroding medical privacy rights.

Even better, Professor Smith said, it would make DNA a true deterrent to crime, which it cannot be so long as the DNA databanks contain only information on known criminals and suspects.

The federal government's existing DNA database, by law, includes only material taken from convicted criminals and crime scenes. Increasingly, states including Louisiana and Virginia have authorized the collection of DNA from people arrested for rape, murder and other violent crimes, and in some states even for burglary and lesser charges. The law in most states is much less clear when it comes to the DNA of people merely suspected of a crime but not charged. Yet it is being tested.

In New York City, for example, the medical examiner's office maintains a citywide database of DNA obtained from crime scenes and from suspects in major crimes, either

ith their consent or with a warrant, said Dr. Howard J. Baum, deputy director of forensic biology.

But in November, a defendant in a Brooklyn rape case who was compelled to give a DNA blood sample won a court order barring the medical examiner from placing it in the citywide DNA database, known to medical examiners as Linkage. The defendant, Carlos Rodriguez, argued that a 1994 state law preventing DNA test results from being disclosed without the subject's consent also barred officials from entering those results into the city database. Justice John M. Leventhal of the State Supreme Court even wrote that the mere existence of the database might constitute a felony under the 1994 law. The medical examiner's office is appealing the ruling.

Mr. Kohler, the Baton Rouge man who demanded a court order before giving a DNA sample, says he, too, plans to sue to get it, and his genetic information, back from the police.

Mr. Kohler, a 44-year-old welder, said he resented the way the police relied on a pair of sketchy tips and seemingly irrelevant evidence as their probable cause, though it was enough to persuade a local judge to issue a warrant. Mr. Kohler said the police cited his 20-year-old burglary conviction, but not his full pardon and restitution in 1996.

Mr. Kohler said he felt that the police violated the Constitution by leaning on him for the DNA sample.

"These rights are what makes America America, to me," he said, adding that he felt he could afford to protest while many others could not.

"My friends know me, and I know me, and other people really don't matter," he said. "I'm not running a business, and I don't have any kids. So I had the freedom to take a stand and not hurt the people around me."

In the end, Mr. Kohler, alone among 15 people who refused the DNA test, was indeed identified in public court documents, and hours later a local television reporter appeared at his front door. The police called the court filing a good-faith clerical mistake. The DNA test later cleared Mr. Kohler. And the killer is still at large.

HUMAN GENETICS

Iceland OKs Private Health Databank

Ending months of furious and, at times, bitter debate, the Icelandic parliament has given a private company permission to build a database containing the health records of the entire nation. But critics of the legislation, passed 16 December by a sizable majority, immediately pledged to find ways to block its implementation.

The new law grants one company, deCODE Genetics from Reykjavik, the right to establish and commercially exploit a nationwide database created through agreements with hospitals, clinics, and individual physicians to submit their patients' medical records. The company expects this information to greatly speed up its search for disease-causing genes, on which diagnostic tests and therapies could be based. Icelanders belong to a very homogeneous gene pool, making disease genes much easier to spot here than in other populations.

The Icelandic government hopes the database, which will also be available to health officials, will improve the country's health care system. It also sees genetics as a promising way to generate high-tech jobs for the country's small, fish-based economy. "We have quite a few people abroad who have educated themselves in this field. Now, they can come home and work on this," says Siv Fridleifsdottir, vice-chair of the Committee on Health in the Althingi, the Icelandic parliament. But the deCODE bill, introduced last spring and then revised over the summer, has touched off a sulfurous battle within the research community (*Science*, 14 August 1998, p. 890, and 30 October 1998, p. 859). "This has totally destroyed the scientific atmosphere," says Eirikur Steingrimsson, a geneticist at the University of Iceland.

Solid majority. Iceland's parliament says yes to deCODE's databank.

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Critics of the bill say it violates basic ethical principles because patients will not be asked for their consent before their records are deposited in the database. They argue that there should be more safeguards to secure privacy, and that one company should not have the commercial rights to a whole nation's gene pool. Over the past few months, dozens of medical, scientific, and patients' organizations testified against the bill in committee hearings. "We look at this as a black day in the medical and scientific community," says psychiatrist Tomas Zoega, chair of the Ethics Committee of the Icelandic Medical Association. "But the battle will keep on going."

deCODE's founder and president, Kari Stefansson, says that many opponents have acted out of professional envy rather than ethical concerns. "A subpopulation of people working in biomedicine in Iceland feels that we have disrupted their lives simply by our size," says Stefansson, a former Harvard University geneticist. "They have great difficulty recruiting people in their labs and competing with us." Now that the bill is passed, he adds, "I expect that there will be a lot of reconciliation." Adds University Hospital gastroenterologist Bjarni Thjodleifsson, who is working with deCODE on a genetic study of inflammatory bowel disease, "This is a revolutionary bill, and people are unduly paranoid about their position. As the dust settles, matters will clear up, and trust can be obtained."

With only two defections from the ruling coalition, the bill passed parliament by a vote of 37 to 20. Still, the debate opened many wounds in the body politic. Critics claim that deCODE had too much influence in drafting the bill. In particular, they point to a last-minute addition that allows deCODE to link the database's medical information to existing genealogical records and to genetic information that the company collects in its own studies—an arrangement that critics say will make it relatively easy to identify individual patients and learn sensitive details about them. "I have never witnessed such a stronghold [on the parliament] by one company that has interests in a law," says Social-Democrat Össur Skarphedinsson, chair of the health panel.

But Stefansson says the company was not trying to hide anything. "This [database link] had been the idea that was discussed from day one," he says. "If the politicians say they didn't know about it, they are being very disingenuous." He also denies that the company has received any special favors. "You can have a stronghold simply by the power of your idea."

Despite their defeat, deCODE's critics haven't given up. One recourse, says Zoega, is to ask the Icelandic and European courts to overturn the law on the grounds that it violates an individual's right to privacy. In addition, the bill allows individuals to notify the surgeon-general if they oppose use of their data, and the medical association may place ads and provide patients with the necessary forms, he adds. Already, 44 general practitioners and 109 hospital specialists have pledged not to send information to the database unless a patient specifically requests them to do so. "We will certainly be dragging our feet," Zoega says about participating in the data collection.

–MARTIN ENSERINK

Martin Enserink is a science writer in Amsterdam.

Paternity Testing Issues

(1) CLAIM MAN/not husband is father of child for support –

(2) Divorce – husband disputes being father of child. (Texas allows husband to challenge paternity at anytime).

(3) PRE-Natal testing – Rapist or Spouse vs. Sexual partner as father?

(4) Adoption/IMMIGration to see if persons related. e.g., Argentina → grandparents find children of killed sons/daughters.

(5) Widely Accepted Requiring Testing. Courts generally do not recognize parental rights or duties if genetic tie missing & vice versa!!

e.g. – husbands who have parented a long time – Social father-can escape obligations by using DNA tests to show no biological connection & vice versa!

Other Laws Involving Genetics

1. Human Cloning
2. Embryo / Fetal Research
3. Genetic Privacy
4. Storage / Disposal of Human Embryos

State Human Cloning Laws

Updated: 1/22/03

Six states have laws pertaining to the use of human cloning, which was first addressed by the state of California with a ban on human cloning in 1997. Since then, five other states-Louisiana, Michigan, Rhode Island, Virginia and most recently Iowa-have enacted measures to prohibit human cloning. In addition to prohibiting the creation of human embryos for the purpose of initiating a pregnancy, Michigan and Iowa extend their restrictions to the creation of human embryos via cloning techniques regardless of the intended use. Virginia's law also was intended to prohibit human cloning for any purpose, but the law does not define human being, which could be interpreted as from the moment of fertilization onward, from the fetal stage onward or beginning at birth. Finally, Missouri forbids the use of public funds for human cloning research.

State	Citation	Summary	Expiration
California	Business And Professions §16004, §16105, Health & Safety §24185, §24187, §24189, §12115-7	Provides for the revocation of licenses issued to businesses for violations relating to human cloning; prohibits cloning of human beings for the purpose of initiating a pregnancy and the purchase or sale of ovum, zygote, embryo, or fetus for the purpose of cloning human beings; establishes civil penalties	
Iowa	2002 SB 2118	Prohibits human cloning for any purpose; prohibits transfer or receipt of a cloned human embryo for any purpose, or of any oocyte, human embryo, fetus, or human somatic cell, for the purpose of human cloning; establishes civil penalties and grounds for revoking licensure	
Louisiana	40 §1299.36.1 to 6	Prohibits human cloning for the purpose of initiating a pregnancy; establishes civil penalties	July 1, 2003
Michigan	§§333.26401 to 06; §333.16274, §16275, §20197, §750.430a	Prohibits human cloning for any purpose and prohibits the use of state funds for human cloning; establishes civil and criminal penalties	
Missouri	§1.217	Bans use of state funds for human cloning research which seeks to develop embryos into newborn child	
Rhode Island	§23-16.4-1 to 4-4	Prohibits human cloning for the purpose of initiating a pregnancy; establishes civil penalties for corporations/hospitals and individuals	July 7, 2010
Virginia	§32.1-162.32-2	Prohibits human cloning, or the creation of or attempt to create a human being by transferring the nucleus from a human cell from whatever source into an oocyte from which the nucleus has been removed (human being is undefined); also prohibits the implantation or attempted implantation of the product of somatic cell nuclear transfer into an uterine environment so as to initiate a pregnancy; the possession of the product of human cloning; and the shipping or receiving of the product of a somatic cell nuclear transfer in commerce for the purpose of implantation of such product into an uterine environment so as to initiate a pregnancy. The law establishes civil penalty not to exceed $50,000 for each incident.	

Source: NCSL

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State Embryonic and Fetal Research Laws

Updated 1/22/03

There are four primary sources for embryonic stem cells: existing stem cell lines, aborted fetuses/embryos, unused in vitro fertilized embryos, and cloned embryos. Current federal policy limits federally funded research to research conducted on embryonic stem cell lines created before August 2001. Federal funding of research involving cloning for the purpose of reproduction or research is prohibited. However, there is no federal law banning human cloning altogether. The Food and Drug Administration has claimed authority over the regulation of human cloning technology as an investigational new drug (IND) and stated that at this time, they would not approve any projects involving human cloning for safety reasons, but Congress has not passed legislation confirming the FDA's authority to prohibit cloning.

State laws may restrict some or all sources for embryonic stem cells or specifically permit certain activities. State laws on the issue vary widely. Approaches to stem cell research policy range from California's law enacted in 2002 that encourages embryonic and adult stem cell research to South Dakota's law, which strictly forbids research on embryos regardless of the source. If, however, a fetus is aborted for the health of the mother in South Dakota, the fetus may be used for research purposes with maternal consent. Many states restrict research on aborted fetuses or embryos although research is often permitted with consent of the patient. Almost half of the states also restrict the sale of fetuses or embryos. Louisiana is the only state that specifically prohibits research on IVF embryos. Illinois and Michigan also prohibit research on live embryos. Finally, Iowa and Michigan prohibit research on cloned embryos. Virginia's law also may ban research on cloned embryos, but the statute may leave room for interpretation because human being is not defined. Therefore, there may be disagreement about whether human being includes blastocysts, embryos or fetuses. California, Louisiana and Rhode Island also have human cloning laws, but these laws prohibit cloning for the purpose of initiating a pregnancy only, or reproductive cloning. These laws do not prohibit embryonic stem cell research. Missouri also forbids the use of state funds for reproductive cloning but not for cloning for the purpose of stem cell research.

State	Specifically permits research on embryos	Specifically prohibits research on aborted fetus/ embryo	Requires maternal consent to conduct research on nonliving fetus	Prohibits research on fetus or embryo resulting from sources other than abortion	Prohibits sale of fetus or fetal tissue	Prohibits sale of embryo
California Health & Safety € 123440, 24185, 12115-7	Ã	Live fetus			Ã[2]	Ã[2]

[1] Permitted on aborted fetuses born dead with consent

[2] Prohibited for the purpose of cloning a human being or for stem cell research on cloned embryos

[3] Not permitted on aborted fetus

[4] Prohibits the sale of fetus, embryo or neonate for illegal purposes

[5] Minnesota law protects live embryos for 265 days after fertilization; research on embryos kept alive through cryopreservation past 265 days is permitted

[6] Permits the buying and selling of a cell culture line or lines taken from a nonliving human conceptus

[7] Prohibits sale, distribution or donation of live or viable aborted child

[8] Prohibits abortion for the purpose of selling the fetus to researchers

[9] Requires consent to conduct research on a nonliving fetus or embryo resulting from an occurrence other than abortion

[10] May not sell fetus to be used for illegal purposes

[11] May not sell fetus or fetal remains resulting from an abortion

[12] No consideration may be given to mother consenting to research or others in connection with transfer of fetal tissue.

[13] In cases involving abortion, consent must be provided after decision to abort has been made.

[14] Except for expenses occasioned by the actual retrieval, storage, preparation and transportation of the tissues is permitted

[15] Permits research on fetus aborted for the health of the mother

[16] Consent required to conduct research on an aborted fetus

[17] Prohibits sale of aborted fetus only

[18] Statute refers to "live unborn children". The term is not defined, but appears not cover in vitro fertilized embryos. The abortion chapter where it is located refers to abortion as a procedure undertaken to terminate a human pregnancy after implantation of a fertilized ovum or kill a live unborn child.

[19] Virginia law does not expressly prohibit research on cloned embryos, but it is forbidden to possess the product of human cloning. Under the state human cloning statute human cloning is defined as the creation of or attempt to create a human being by transferring the nucleus from a human cell from whatever source into an oocyte from which the nucleus has been removed. Human being is not defined as to whether it includes neonates, embryos or fetuses only.

[20] Virginia law prohibits shipping or receiving of the product of human cloning for commerce. Under the state human cloning statute human cloning is defined as the creation of or attempt to create a human being by transferring the nucleus from a human cell from whatever source into an oocyte from which the nucleus has been removed. Human being is not defined as to whether it includes neonates, embryos or fetuses only.

[21] Prohibits sale, distribution or donation of live or viable aborted child, defined to include embryos, for the purpose of experimentation

State Laws and Legislation:
Use, Storage and Disposal of Frozen Embryos

Health Programs

Updated August 2002

Selected Case Law
Legislation

State	Statutes
Florida	**Fla. Stat. Ann.** § 742.17 requires written agreement that provides for the disposition of a couple's eggs, sperm, and pre-embryos in the event of a divorce, the death of a spouse, or any other unforeseen circumstance.
Louisiana	**La. Rev. Stat. Ann.** § 9:391.1 declares that any child conceived after the death of a decedent, who specifically authorized in writing his surviving spouse to use his gametes, shall be deemed the legitimate child of such decedent, provided the child was born to the surviving spouse, using the gametes of the decedent, within two years of the death of the decedent. Any heir of the decedent whose interest in the succession of the decedent will be reduced by the birth of a child conceived shall have one year from the birth of such child within which to bring an action to disavow paternity.
North Dakota	**N.D. Cent. Code** § 14-18-03; 14-18-07 clarifies legal parentage of a child conceived after invalidity or annulment of marriage or death of spouse.
Texas	**Tex. Family Code Ann.** § 160.001, et seq. creates the Uniform Parentage Act and describes various aspects of determination of maternity and paternity as well as parentage. The law requires a man and woman to sign consent to assisted conception. If the father does not sign, however, it does not necessarily mean that he is not the legal father.
Virginia	**Va. Code** § 20-158(3)(B) clarifies legal parentage of a child conceived after death of or divorce from a spouse.
Washington	**Wash.** House Bill 2346 / Senate Bill 5207 (2002) creates the Uniform Parentage Act and clarifies legal interpretation of parentage of a child of assisted reproduction, including in the event of divorce or death.