

# GENETICS: ETHICAL AND LEGAL CONSIDERATIONS IN GENETIC TESTING

## **Goal:**

Identify the ethical and legal issues that may arise during the genetic counseling process and strategies to effectively address those issues.

## **After completing this activity participants will be able to:**

- Describe the characteristics of genetic information that may complicate the associated ethical, legal, and psychosocial issues beyond those of routine medicine
- Recognize applications of the 4 main biomedical ethical principles (autonomy, beneficence, nonmaleficence, and justice) in medical genetics
- Demonstrate understanding of the role of federal, state, and tort law in managing the genetic counseling and testing process for patients
- Identify potential common ethical and/or legal conflicts in managing the genetic testing process for patients and the strategies to avoid such conflicts

## **Professional Practice Gaps**

In an effort to define what healthcare providers need to know about medical genetics, several organizations developed core competencies (NCHPEG, 2000; ASHG, 2001). However, because clinical genetics is a relatively young and evolving field of medicine, many practitioners received insufficient formal genetics education. As a result, they express a lack of confidence in their clinical genetics knowledge and a lack of confidence in their ability to provide genetic counseling.

## INTRODUCTION

### **Complexities of medical genetics**

The science of genetics has been integrated into everyday life. Information appears in all forms of the media each day. It is no surprise, then, that a patient brings up health issues that involve genetics when visiting your office.

Besides the ever-growing amount of scientific knowledge that has to be assimilated, medical genetics raises ethical and legal issues that may not attach to general medical practice. While medical genetics shares many of the ethical and legal considerations as other areas of medicine, there are factors that may make clinical application of genetic knowledge more complex at times (Harris, Winship, & Spriggs 2005). These factors must be appreciated by healthcare providers when assisting patients in making genetic decisions and in handling genetic information.

### **Factors That May Complicate Medical Genetics Beyond Routine Healthcare Issues**

- Knowledge is advancing at a more rapid rate than in most other medical fields.
- Clinical application of this knowledge often occurs in the face of significant limitations in our understanding of the clinical implications.
- Genetic information has implications for the extended family versus a single individual.

- Genetic information may be viewed more personally than other health information and be associated with significant psychological burden.
- The predictive nature of some forms of genetic testing may be particularly challenging, especially when there are no effective preventive measures.
- A history of misuse of genetic information (coercive eugenics) fuels public fear and misconceptions.
- While privacy is of central importance in all aspects of medicine, patients may have a greater desire to protect the privacy of their genetic information.

Throughout this course, some of the more common questions that may confront you when a patient asks questions about or is a candidate for genetic testing will be reviewed.

## ETHICS

### Biomedical Ethics

A society generally has a code of conduct or agreement about what is and is not acceptable behavior. Ethics provides a theoretical framework for evaluating and explaining these societal codes and defining what is acceptable conduct. This allows rules to be developed. There are times in our personal and professional lives when the best course of action is unclear or there may be motivation to pursue a path that is **not** generally considered "moral." A systematic application of an ethical framework may help to resolve these conflicts.

Biomedical ethics is the study of what is morally acceptable in medical practice and defines the code of conduct followed by the medical profession. The ethical principles that apply in medical genetics are those of general medicine.

Ethical principles are the source of the following guidelines for behavior:

- **Values:** qualities or priorities that are thought to be important and desirable
- **Rules:** specific statements about what should or should not be done that must be observed at all times
- **Duties:** behaviors that are defined by an individual's role in society
- **Rights:** justified claims that individuals or groups can make on others or on society

(Adapted from Muthuswamy 2011)

## AUTONOMY

### Autonomy

Autonomy refers to the potential of the individual to independently make decisions about his or her healthcare and life choices. This is not only good in itself but is also a means for identifying an individual's best interests as assessed on the basis of her own values, preferences, and goals.

Respecting a patient's autonomy requires that we:

- Tell the truth
- Obtain informed consent for treatment or for sharing information
- Maintain confidentiality

This principle is also called ***respect for personal autonomy***.

### **Clinical Example**

Physicians may take care of various members of the same family. Suppose Audrey is affected with, or is a carrier of, a genetic disorder (such as cystic fibrosis) or she carries a chromosome variation (such as a translocation). Audrey's brother (Bill) and sister (Carol) or even her cousins may be at risk for related complications. Even though both Audrey and Carol are in your practice, respect for autonomy requires that Audrey's health status may not be shared without her express permission. She may not want her sister or brother informed of her results at that time. The principle of autonomy also prohibits doing tests on individuals without their knowledge or consent.

## **BENEFICENCE**

### **Beneficence**

Beneficence imposes the obligation to seek the good for the patient under all circumstances. It gives rise to the obligation to maximize possible benefits for, while minimizing the possible harms to, the patient. Healthcare providers act as the patient's advocate.

Beneficence requires

- Honesty
- Candor
- Loyalty
- Fidelity

When beneficence is overemphasized and autonomy minimized, we have a paternalistic system.

### **Clinical Example**

In general, parents are the healthcare decision-makers for their minor children. Situations can arise that require the physician to put the best interests of the child above his respect for the parents' autonomy and make decisions about the care of the child (with permission from the court). For example, not every family will accept blood transfusions as part of a treatment plan. If this belief puts a child's life in jeopardy, beneficence would require that the physician exercise his best judgment in the interest of the child and seek legal permission to treat with transfusion.

## **NONMALEFICENCE**

### **Nonmaleficence**

Nonmaleficence imposes an obligation to avoid or refrain from intentionally harming the patient. A provider who cannot bring about good for the patient is obliged to at least avoid harm. Harm can be caused by ***omission***, failing to do what you should, or by ***commission***, doing what you should not have done. The obligations (moral responsibilities or duties imposed legally or socially) derived from nonmaleficence are more stringent than those from beneficence.

## Clinical Example

Parents of infants with anencephaly may desire to help others through organ donation. The dead donor rule, which is derived from the principle of nonmaleficence, requires that an organ donor be dead before life-sustaining organs are removed. The medical definition of death precludes the procurement of organs from these infants. The principle of nonmaleficence also forms the basis of the guidelines for using human subjects in research.

# JUSTICE

## Justice

Justice exists on 2 levels: individual justice and societal (or distribution) justice.

On the **individual** level, we treat patients in a fair, equitable, and appropriate manner in light of what benefits are due or owed them -- what they have a right to.

When we consider **societal** justice, we have to think of the equitable distribution of burdens and benefits. That entails weighing the common good versus the good of the individual. Problems arise when there is scarcity of and competition for resources. This imposes the role of stewardship on the healthcare provider. These questions are not appropriate for the bedside and belong in the public forum.

## Clinical Example

Baby K was born with trisomy 18 and lived in a nursing home. She frequently developed pneumonia and was brought to the hospital for treatment. The treatment she required was not for her birth defect but for the infection. The principle of justice prohibits discrimination and requires fair treatment. The baby was treated for each new infection and lived for over 2 years.

# TEST YOURSELF

You are Laura's obstetrician. She is currently 14 weeks pregnant and is known to have a hereditary breast and ovarian cancer (HBOC) predisposing gene mutation. Her mother died from breast cancer when Laura was a child, and several other relatives have had breast and/or ovarian cancer. In considering HBOC genetic testing for the current pregnancy, Laura had thorough genetic counseling and informed consent with the same genetic counselor who worked with her when she was deciding about her own personal testing. Laura says she would not intentionally bring a child into the world with her "family's curse." She wants to pursue prenatal diagnosis for fetal HBOC gene mutation analysis. You disagree with her decision. There will likely be many positive advances in prevention by the time an at-risk child would manifest HBOC. You also believe that Laura is likely to experience serious feelings of guilt if she does terminate a pregnancy because the fetus has the same mutation as Laura.

In deciding how to proceed with this case, what ethical principle bears the greatest weight in this scenario?

Choose one:

1. Respect for autonomy

- Feedback:

This is the best choice. While all of these ethical principles will play a role in deciding how to proceed, respect for autonomy will likely bear the greatest weight. Respect for autonomy requires that we recognize Laura as a competent adult who understands and can evaluate the benefits and harms of the test as well as the implications of positive and negative fetal BRCA test results. At this time, she is the decision-maker for her fetus. The future autonomy of the fetus with respect to BRCA testing should be included in the consent process but does not outweigh Laura's autonomy.

Physician autonomy is also a consideration. Your professional judgment and standard of practice should guide you when you do not think a test demanded by a patient is appropriate. While you do not have to personally provide testing, it is your responsibility to make an appropriate referral.

## 2. Beneficence

- Feedback:

This is NOT the best choice. Beneficence requires that we do what is in the best interest of the patient. While this is usually Laura, as an obstetrician you will likely consider the interests of the fetus as well. Benefits to the fetus may include being born without the HBOC mutation and being a chosen child if negative. Harms might include the physical harms from the procedure itself or not being born at all if positive for the mutation. Laura has made what appears to be an informed decision about the best course of action, in her view. However, there is a disagreement about what is in Laura's best interest that is largely based on personal, not medical, factors. Testing is available but there is no medical standard of care. Therefore, respect for autonomy must take precedence.

## 3. Nonmaleficence

- Feedback:

This is NOT the best choice. Nonmaleficence imposes an obligation to avoid or refrain from intentionally harming the patient. While this is usually Laura, as an obstetrician you will likely consider the interests of the fetus as well. Do the harms of prenatal testing outweigh the benefits? One source of harm is the risk to Laura and the fetus from the procedure itself. Another consideration is the harm to the fetus of not being born at all, if positive for the mutation, and the long-term implications for Laura of terminating an otherwise normal pregnancy if the fetus has inherited her HBOC mutation. While these are potential harms, Laura appears to have carefully considered the options. She made an informed decision that the medical and psychological risks are acceptable to avoid the outcome of having an affected child. Thus, respect for autonomy must take precedence.

## 4. Individual Justice

- Feedback:

This is NOT the best choice. Individual justice requires that treatment be appropriate, equitable, and fair. Individual justice does not obligate you to offer all available testing to every patient. Your professional judgment and standard of practice should guide you

when you consider a patient's request for testing. Because testing is available, is technically appropriate, and there exists no medical standard of care to guide decision making, respect for autonomy must take precedence.

## LEGAL PARAMETERS

### Legal Parameters

Individuals have rights that they may claim from others.

**Moral rights** come from a community's values and beliefs and encompass, for example, the right to make one's own choices and act on them.

A **legal right** is one that is allowed by law. These rights are not absolute and are not unchanging. Society has codified much of our cultural values and beliefs into the legal system, although there is a lag between the evolving ethical attitudes of society and the law. Laws exist on many levels and in several forms in something of a hierarchy.

The United States Constitution is the supreme law of the land in the United States. On the federal level, there are several sources of laws that govern different aspects of healthcare. View the links in the Related Resources section to learn more.

States also have constitutions as well as statutes that apply to that state. Many states have enacted laws that pertain specifically to genetic privacy. Cases that are decided in federal and state courts establish precedents for future decisions by those courts. Areas of law that are left solely to the states include public health, insurance, and professional licensing.

### Case Examples

- The Americans With Disabilities Act forbids discrimination in the workplace. Lawrence Berkeley Laboratory was found to be testing some of its employees for sickle trait and others for pregnancy without their knowledge [ *Norman-Bloodsaw v. Lawrence Berkeley Laboratory*, 135 F. 3d 1260 (9th Cir. 1998)].
- More recently (in 2001), the Burlington Northern Santa Fe Railroad was stopped by the Equal Employment Opportunity Commission from obtaining blood for DNA testing from employees who sought disability compensation as a result of carpal tunnel syndrome that occurred on the job. Testing employees after they were disabled without their informed consent violates antidiscrimination laws.

## TORT LAW

### Tort Law

A **tort** is a civil wrong committed against a person or property. The basic purpose of tort law is to keep peace between individuals by supplying a substitute for vengeance and to find fault for wrongdoing, deter the wrongdoer, and encourage adherence to the law. An intentional tort is a willful act that violates the rights of someone else, such as assault or invasion of privacy. Negligent torts are the performance of an act or failure to perform an act that a reasonably prudent person would or would not do. Malpractice is a negligent tort.

## Torts of Particular Interest in Medical Genetics

**Wrongful birth actions** are brought by the parents of an unplanned, unwanted, or defective child, claiming that the negligence of the healthcare provider was a failure to act appropriately. For example, in the classic case of *Becker v. Schwartz* [386 N.E. 2d 807 (N.Y. 1978)], the parents of a child with Down syndrome claimed the physician should have counseled the 37-year-old mother about the increased risks and available testing.

**Wrongful life suits** are brought by the defective child claiming it would have been better not to be born than to be born with defects and to suffer pain. The healthcare provider is accused of breaching a duty to the parents to inform them of the probability that the child would be defective. There is almost uniform rejection by states of this type of suit. When the suit *Berman v. Allen* [80 N.J. 404 A.2d 8 (1979)] was brought by a child with Down syndrome whose mother was 37 years old at delivery, the New Jersey courts denied recovery, stating that the valuation of nonlife was impossible and there is no injury recognizable at law by being brought into existence.

# NEGLIGENCE

## Types of Negligence

There are 3 major types of negligence encountered in malpractice torts as shown below, with some specific examples relevant to medical genetics.

- Misuse of information
  - ordering the wrong test or misinterpreting a test result
- Failure to act
  - not taking a family history or not recognizing a high-risk situation
  - not ordering appropriate tests
- Failure to provide complete and accurate counseling
  - provision of incorrect recurrence risks

# PROFESSIONAL STANDARDS

## Professional Standards

There are 4 major components to a negligence tort: 1) duty, 2) breach of duty, 3) causation, and 4) injury (Schmerler, 1998). In medical negligence torts, the professional's duty to the patient, and therefore what defines a breach of duty, is commonly the central issue. The duty is typically defined by the standard of care. A **standard of care** describes what is expected of an individual in a given situation. And what is expected is that degree of care ordinarily exercised by health professionals of similar training and experience or what a reasonably prudent person acting under similar circumstances would do.

Possibly the greatest tool healthcare providers have in protecting themselves from malpractice liability is being familiar with and adhering to his or her professional standards. The policy and position statements of such organizations as those shown below can help define those standards.

## Policy and Position Statement Resources

The following resources are available on policy and position statements. View the Related Resources section for links to the following resources:

- American Academy of Family Physicians: Policy and Advocacy
- American Academy of Pediatrics, Committee on Genetics: Current AAP Policy Statements Authored by the COG
- US Department of Health and Human Services: National Guideline Clearinghouse

Ethical and legal issues. In: Baker DL, Schuette JL, Uhlmann WR, eds. *A Guide to Genetic Counseling*. 1998.

## INITIAL HISTORY

Your patient, Connie, is a 26-year-old woman with no significant personal medical history. She is seeing you today for the first time in about a year for a routine physical. She mentions during your history review that a paternal uncle was diagnosed with colon cancer about 6 months ago, when he was 44 years old.



Does Connie's family history warrant any further medical consideration?

Choose one:

1. Yes

- Feedback:

This is the best choice Connie's uncle had colorectal cancer diagnosed at a relatively young age. Colorectal cancer diagnosed prior to age 50 is considered to be an indicator that there may be a hereditary component. This may have implications for Connie's recommended medical management plan and warrants further investigation.

2. No

- Feedback:

This is not the best choice Connie's uncle had colorectal cancer diagnosed at a relatively young age. Colorectal cancer diagnosed prior to age 50 is considered to be an indicator that there may be a hereditary component. This may have implications for Connie's recommended medical management plan and warrants further investigation.

## ETHICAL PERSPECTIVE

### Standard of Care

Connie brought up the subject of her uncle's diagnosis, suggesting that it has importance for her. Therefore it needs to be acknowledged and addressed. In order to best serve Connie within the standard of care defined by your profession, you have a duty to recognize that colon cancer may be inherited and know what questions to ask. This requires a basic level of competence and

preparedness. The value of preparedness can be derived from the principles of autonomy and beneficence. It also requires that you know the limits of your own knowledge and that you can seek assistance from appropriate experts.

## LEGAL PERSPECTIVE

### Standard of Care

The physician/patient relationship requires that you conform to a standard of care that is usually defined by your profession. If you take a family history, it should be done according to published guidelines. Does the standard of practice **require** you to know that colon cancer can be inherited? This can be argued in some situations. For this case, there are court decisions that may apply.

- *Donadio v. Crouse-Irving Memorial Hosp., Inc.* [75 A.D.2d 715, 427 N.Y.S. 2d 118 (1980)]: The physician failed to properly evaluate the history and refer to a genetic counselor and neonatologist.
- *Phillips v. United States* [575 F. Supp. 1309 (D.S.C. 1983)]: Suit was brought for failure to consider positive family history of a 23-year-old patient with Down syndrome.

According to *The Core Competencies* of the National Coalition of Health Professional Education in Genetics (NCHPEG 2007), all healthcare providers should be able to "identify clients who would benefit from genetic services" (NCHPEG 2007, skill 2.2). This indicates that a healthcare provider who fails to recognize that Connie has an increased risk for an inherited form of colorectal cancer could be liable.

### Provider-Patient Privilege

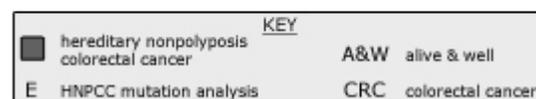
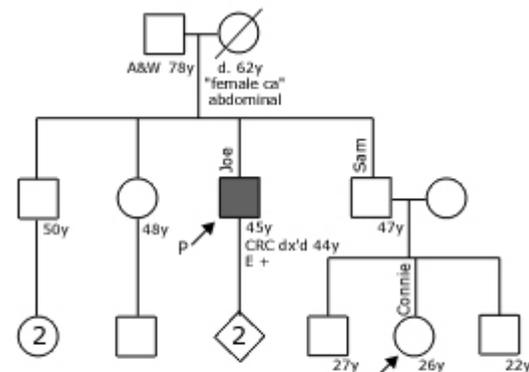
The information Connie shares with you is privileged. This privilege was established when you agreed to treat her. The information that Connie shares with you (including family history) can be documented, provided it has the same privacy and security protections as any other medical chart contents.

You will likely desire additional information about the family history (that Connie may not know) before making important management decisions. You may not pursue obtaining this information from family members or mention her visit with you to any third party without her consent. When additional information is required, ideally the patient should personally inform relatives of the family history concerns and seek their consent for obtaining additional necessary information and documentation.

## FAMILY HISTORY

### Clinical History

After probing further into the family history, you learn that Connie's uncle, Joe, has had molecular genetic testing for hereditary nonpolyposis colorectal cancer



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(HNPCC). He was found to have a cancer predisposing gene mutation. No one else in the family has had colorectal cancer, although Connie's paternal grandmother is reported to have had an abdominal "female cancer." HNPCC is inherited as an autosomal dominant disorder, which means that Joe's first-degree relatives have up to a 50% chance for having also inherited the HNPCC mutation. Connie's father has declined genetic testing. He is not your patient.

## ETHICAL PERSPECTIVE

### **Unwilling Disclosure**

Connie's father does not want to know his own mutation status. He has exercised that right by not pursuing testing. What is the relationship of his rights to Connie's autonomy and her right to be tested? When does one right trump another? The principle of nonmaleficence says you do no harm, but to whom does this apply first and foremost? Connie is your patient. Your duty is first to her. Her father has no specific claim on you as a professional.

The implications of a positive test for Connie's father and his rights should be a part, but not the focus, of your discussion with Connie. However, Connie's results do not need to be disclosed to her father or anyone else. A plan to minimize potential harms to Connie's father can be discussed and developed prior to initiating testing.

## LEGAL PERSPECTIVE

### **Privacy and Confidentiality**

The need for privacy of all parties is important. Connie's uncle made the decision to share his own test results with his family members. Connie may not feel the same way. Her decision to be or not to be tested and any results of her tests are privileged. You may not share them without consent. HIPAA regulations regarding confidentiality of patient information will apply here.

If Connie decided to pursue genetic testing, you will likely need to obtain a copy of her uncle's test results so that she can have targeted testing for the known family-specific mutation (this significantly increases test sensitivity). To protect her uncle's privacy, you might consider creating a separate chart for him and his results.

### **Professional Competence**

Failure to offer testing has been the basis of a number of suits.

- As early as 1977, not offering an amniocentesis to a woman over 35 years of age was the cause of action in *Karlsons v. Guerinot* [57 A.D.2d 73, 394 N.Y.S.2d 933 (1977)].
- In *Smith v. Cote* [513 A.2d 341, 355 (N.H. 1986)], a mother claimed that the physician failed to advise her of the risk of birth defects in a fetus exposed to rubella (failure to inform of risks and alternatives).

# GENETIC TESTING DECISION

## Clinical History

After you discuss the implications of a known familial HNPCC mutation with Connie, she indicates that she is interested in learning more about genetic testing. She has considered her father's decision not to be tested but believes that she might make a different decision for herself.

## Points to Consider

Before initiating any form of genetic testing, Connie will need to give informed consent. Informed consent is a **process** that involves several elements [see Related Resources section for more information]. In order for Connie to give informed consent, she must be competent to do so. She must also receive and understand adequate and accurate information as described in the table above. This includes a discussion of the benefits, risks, and limitations of both pursuing and declining genetic testing. This should include such issues as psychosocial ramifications and the possibility of genetic discrimination. Consent for any course of action must be voluntary and well documented. You are expected to **adequately address** all of these elements of informed consent or to refer to an appropriate expert.

- Arranging for genetic testing may involve **coordination of testing** as well as **collaboration with other professionals**.
- Consider also that genetic testing may yield unexpected findings such as the following:
  - Ambiguous results that cannot be interpreted with current technology
  - Results that can only be interpreted with the cooperation of other family members
  - Results that suggest a diagnosis or risk for a condition other than the one specifically tested
  - Discovery of misattribution of paternity

These possibilities should be addressed in the consent process and prepared for as adequately as possible.

# ETHICAL PERSPECTIVE

## Informed Consent

Respect for autonomy allows people to make medical decisions for themselves. The informed consent process ensures that Connie has the basis needed for making the best decision for herself. Informed consent requires candor and honesty. All the goals listed on the previous page need to be included in the discussion, as well as costs of testing and the possibility of misuse of the information by third parties (e.g., employment or insurance discrimination). Your opinion about the best course should not unduly influence Connie's decision. Making the decision for her or coercing a choice is an example of paternalism. A consent form that the patient signs is important documentation that the informed consent process has occurred, but it does not substitute for the communication process.

## Access to Testing

You will need to understand the caveats of available tests and be able to coordinate or collaborate with other professionals and laboratories or refer Connie to an appropriate specialist. On an individual basis, justice requires fairness in offering available testing to patients, regardless of your estimate of

their ability to pay or difficulty of testing. The patient will take these factors into account when she decides about testing. Decisions about who should have access to scarce resources, such as expensive tests and who should pay for them, are social justice issues to be discussed in a public forum.

## LEGAL PERSPECTIVE

### **Discrimination**

There is a growing fear about the misuse of genetics. The risk of discrimination based on one's genetic status has not been substantiated although there is anecdotal evidence of genetic discrimination in insurance, employment, and other settings. There currently are no federal protections against life, disability, and long-term care insurance discrimination. However, many states have enacted laws that prohibit some forms of discrimination. You should review your state's protections (summarized in an easy-to-interpret table) at the National Conference of State Legislatures' *Genetics and Life, Disability and Long-Term Care Insurance* article. Some patients may wish to obtain these forms of insurance prior to undertaking genetic testing. However, the results of genetic testing should not be omitted from an application for insurance.

Health insurance discrimination is prohibited for government programs and employer-sponsored plans for medium to large employers. The *Health Insurance Portability and Accountability Act* sets a floor for the protection of patient information. Some states have laws that place specific restrictions on such insurer activities as requiring genetic testing or using genetic information for eligibility or risk classification purposes for individual and/or group policies. You should review your state's protections at the National Conference of State Legislatures' *Genetics and Life, Disability and Long-Term Care Insurance* article

The Privacy Rule permits a covered entity to use and disclose protected health information without authorization, and with certain limits, for its own treatment, payment, and healthcare operations activities. HIPAA prevents disclosure of genetic information by insurers to employers in most cases. Patients should be certain that they have not signed an authorization form that allows sharing of information. HIPAA also clearly states that genetic information should not be treated as a preexisting condition.

The National Society of Genetic Counselors provides a brochure for consumers, *Genetic Information, Privacy & Discrimination...What You Need to Know*. A full-color PDF version is freely available to print and disseminate to your patients.

### **Professional Competence – Choose Appropriate Tests**

You have a duty to offer Connie appropriate testing options or refer her to another professional for testing. In *Howard v. Lecher*

<sup>1</sup>, the family claimed their ethnic background was not evaluated and they were not offered carrier testing for Tay-Sachs disease. A woman with a family history of hemophilia was tested for factor VIII deficiency but not for factor IX deficiency<sup>2</sup> and subsequently told she had a low risk of being a hemophilia carrier. Improper testing for Tay-Sachs disease also resulted in a number of lawsuits naming both institutions<sup>3</sup> and laboratories<sup>4</sup>. The California State Department of Health was named in

a suit when it failed to follow-up on newborn screening results that indicated an infant had congenital hypothyroidism<sup>5</sup>.

Any testing offered to Connie must be performed and interpreted correctly. Several different options, including full gene sequence analysis, single mutation testing, or mutation scanning, are available. Choosing the correct test for the clinical circumstances may require a sophisticated understanding of the test applications.

**References:**

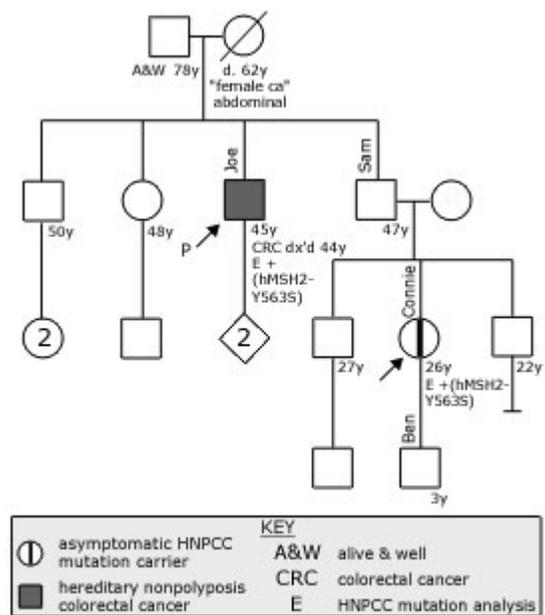
- <sup>1</sup>Howard v. Lecher, 397 N.Y.S.2d 363 (1977).
- <sup>2</sup>Siemieniec v. Lutheran General Hospital, 480 A.2d 1227 (Ill. App. 1 Dist. 1985).
- <sup>3</sup>Gildiner v. Thomas Jefferson University Hospital, 451 F. Supp. 692 (E.D. Pa. 1978).
- <sup>4</sup>Curlender v. Bio-Science Laboratories, 106 Cal. App.3d 811, 165 Cal. Rptr. 477 (1980).
- <sup>5</sup>Creason V. State Dept. of Health Services, 64 Cal. Rptr.2d 534 (1997)

## LOCATE A GENETICS PROFESSIONAL

### Locate A Genetics Professional

To easily locate local genetics professionals, the following organizations maintain searchable databases of genetic counselors, physician geneticists, and other genetics professionals:

- National Society of Genetic Counselors: ResourceLink -- The National Society of Genetic Counselors is the professional membership association for the genetic counseling profession. The website provides a searchable database of genetic counselors by location, name, or specialty.
- American College of Medical Genetics: Membership Directory -- The American College of Medical Genetics consists primarily of doctoral- (MD, PhD, DO) and master's-level (genetic counselors) medical genetics professionals. The organization provides a searchable database of all members by name or location.
- GeneTests – The GeneTests website hosts searchable directories of both genetic testing laboratories and clinics . GeneTests is searchable by disease, location, services offered, lab name, or lab director's name.
- National Cancer Institute: Cancer Genetics Services Directory – A database of professionals who provide cancer genetics services, including counseling and testing. The database is searchable by type of cancer, family cancer syndrome, location, or name.



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# POSITIVE RESULTS

## **Clinical History**

After thorough informed consent, Connie does decide to pursue genetic testing. Her uncle released his HNPCC test results to you that provide his specific mutation. After about 2 weeks, you receive a report indicating that Connie does have the same HNPCC gene mutation as her uncle. She is scheduled to see you for result disclosure in 3 days.

# ETHICAL PERSPECTIVE

## **Result Disclosure**

Respect for autonomy requires truth and candor in result disclosure. Now that Connie has been found to have an HNPCC predisposing mutation, we know that she has an approximate 80% lifetime risk for colorectal cancer and an increased risk for several other cancers. In order for Connie to make medical care and life decisions for herself and her family, she must have sufficient and relevant information, such as management options, therapies, and prognosis.

Connie could also decide any time prior to result disclosure that she no longer wants to receive her results. If this should happen, her reasons for changing her mind should be explored. Psychological support may be useful. The results should be maintained in the chart. Connie should be reassured that result disclosure can happen at any time in the future if she ultimately decides that she would like to have them.

## **Duty to Warn**

The question of your responsibility to third parties is a problem only if Connie decides not to share her test results with at-risk relatives. Her claim of confidentiality has to be weighed against possible harm to the other family members. Confidentiality is a very important value, so any anticipated harms should be highly likely to occur, imminent, and serious. There are guidelines to help make the decision when to breach the patient's confidentiality. If you decide to breach an ethical principle, you must be able to apply your reasoning to all patients in the same situation, have sound justification, and do so openly (rather than in secret). Working with the patient on this issue may take time and patience. Discussions about sharing positive results are best held prior to testing.

# LEGAL PERSPECTIVE

## **Duty to Warn**

Your duty to warn third parties is determined in part by the standard set by your professional code of ethics and practice guidelines and in part by the laws of your state. While there may be a compelling ethical argument for warning at-risk family members, the HIPAA Privacy Rule prohibits the disclosure of protected health information without an authorization, except in specific circumstances. There is currently no exception for the notification of at-risk relatives. Breach of confidentiality and intentional infliction of emotional distress could be the basis of a lawsuit in this situation. Consequently, patients should be urged to inform their at-risk relatives; but, from a legal perspective, the choice of whether to do so currently appears to rest with the patient.

## Privacy of Results and Records

HIPAA regulations will apply to the electronic transmission and storage of the patient's results. Many states have genetic privacy acts that define what constitutes a genetic test, what protections need to be in place for the patient's privacy, and what points need to be included in the consent for genetic testing. You should be familiar with the regulations in your state.

## TESTING OFFSPRING

Now that Connie knows that she has an HNPCC mutation, she is understandably concerned about her 4-year-old son, Ben. Connie is asking about her options for testing Ben.



Should HNPCC gene mutation testing be offered for Ben?

Choose one

1. Yes

- Feedback:

This is the best choice In most cases, testing Ben for HNPCC in childhood would not be the best choice. However, this is a complex decision, and several factors must be weighed. When a gene mutation is identified in a family, very often parents question the gene mutation status of their children and request testing for the children. There are reasons to perform presymptomatic genetic testing for children that include psychological and social issues as well as medical benefits. However, there are also potential harms to the child from such testing that must be considered.

2. No

- Feedback:

This is not the best choice In most cases, testing Ben for HNPCC in childhood would not be the best choice. However, this is a complex decision, and several factors must be weighed. When a gene mutation is identified in a family, very often parents question the gene mutation status of their children and request testing for the children. There are reasons to perform presymptomatic genetic testing for children that include psychological and social issues as well as medical benefits. However, there are also potential harms to the child from such testing that must be considered.

## ETHICAL PERSPECTIVE

### Genetic Testing in Minors

The right of a parent to decide how to raise his or her children (parental autonomy) has to be balanced with the child's future right to autonomy. This becomes most important when the testing will not change the medical care of the child (when the disorder has an adult onset or the genetic test is one of susceptibility or predisposition). The benefits of knowing the test results need to be compared

with possible burdens. It is important to consider the child's future right not to know and to self-determination. Parents may also elect not to reveal test results to children.

## **Screening**

Screening for HNPCC-related cancers is generally recommended to begin at 20 years of age (except in rare families where very early onset is documented and screening begins 10 years earlier than the youngest age of onset). For Ben, this means that there will be no clear medical benefit to testing in childhood. Thus, the balancing of benefits and burdens is largely based on psychosocial factors. Connie may desire Ben to be screened, hoping that he will be negative and a burden of guilt will be lifted or her anxiety reduced. Has she really considered, however, what she will feel if he is confirmed to have a familial HNPCC mutation? Will his family, peers, and/or society treat him differently? What would Connie tell him now? If she tells him nothing, when and how will she prepare him in the future? Will such information negatively alter his self-image or unduly impact his life decisions? Will Ben's parents make different financial arrangements (trust fund, life or disability insurance, etc.) for him in preparation for the future if he has an HNPCC mutation? Each family presents a unique constellation of circumstances. These and many more issues need to be carefully discussed with a parent who requests predictive testing on his or her child.

The American Society of Human Genetics and American College of Medical Genetics (2013) issued a joint statement on genetic testing in minors in 1995. Applicable to Ben's case, it states, "If the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset diseases, genetic testing generally should be deferred . . . Further consultation with other genetic-service providers, pediatricians, psychologists, and ethics committees may be appropriate to evaluate these conditions."

Ben's age is also a factor. He is quite young now and unable to adequately assent or consent to testing. However, as a child matures, so does the ability to take part in the healthcare decision-making process. Decision making involving the healthcare of older children and adolescents should include, to the extent feasible, the assent of the child to testing. If Ben was 16 years old and desired testing after informed consent, the benefits and burdens analysis might be quite different.

If a parent insists that you do a test that in your professional judgment is inappropriate, you do not have to comply. Balancing your own autonomy as a professional with that of the patient, you need to maintain your integrity, dignity, and self-respect. You may suggest that the patient seek opinions from other valued sources.

# **LEGAL PERSPECTIVE**

## **Genetic Testing in Minors**

Parents have the legal authority to make decisions for their minor children. This right, however, is not absolute. A court can override that authority when asked to do so in the best interest of the child. Court-ordered blood transfusions for the children of parents who are Jehovah's Witnesses are a common example of the court's use of its *parens patriae* authority.

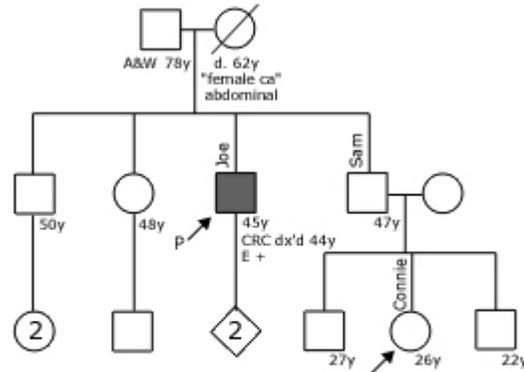
Children age 18 and older are considered competent to make healthcare decisions for themselves. Younger children may be given that right by their state for various reasons. For example, minors who

are married or pregnant may have the authority to make decisions for themselves. The "best interest of the child" doctrine should inform decisions for younger children.

## PRENATAL DIAGNOSIS

### Clinical History

Connie ultimately decided not to have Ben tested for HNPCC. But now, several months later, she has learned that she is pregnant. While she understands the reasons for delaying testing of her son, it has been such a source of anxiety for her that she feels that she may not want to bring another child into the world who has HNPCC.



## ETHICAL PERSPECTIVE

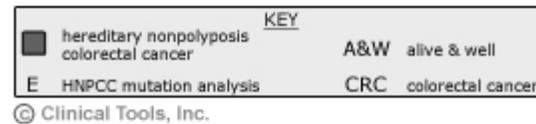
### Healthcare Gate-keeping

The same issues raised regarding testing Ben should also be considered for this pregnancy. In addition, the request for prenatal diagnosis of an adult-onset disorder raises the issue of healthcare gatekeeping. Professionals may have policies or protocols that could limit access to testing for "medical reasons only." Prenatal diagnosis for sex selection, for example, is not available at all testing centers. When cultural or psychological influences are important for the patient, however, we have to consider whether those protocols are a substitute for paternalism.

Recognizing that Connie and other at-risk parents may have compelling reasons for choosing to avoid having an affected child, some centers have attempted to offer prenatal diagnosis for adult-onset conditions only if the parent(s) have decided they will terminate a pregnancy if the results are positive. This obviously cannot be enforced. If a parents decide after learning the results that they do not wish to terminate, they have in effect presymptomatically diagnosed a minor. However, if therapeutic termination is not an option for an affected fetus, would parents with a 25% or 50% risk to have an affected fetus choose not to conceive out of fear? Would other approaches such as in vitro fertilization with preimplantation genetic diagnosis be preferable? Assisted reproduction is quite expensive and invasive. These are some of the most challenging issues faced in clinical genetics.

Physicians do not have to agree or comply with a patient's medical choice. Physician autonomy also requires respect. The physician-patient relationship does, however, require that the patient not be coerced or abandoned when there is a difference of opinion, and proper notification and referrals are necessary. In this case, the preferred approach may be to refer Connie to a prenatal diagnostic center with experience in managing prenatal testing for adult-onset disorders.

Does social justice require you accept the role of steward for limited resources? The decision about who benefits from and who has access to limited resources is a social issue and should be discussed in open forum, as was done in Oregon for the ranking of health problems requiring public funding.



# LEGAL PERSPECTIVE

## Reproductive Rights and Wrongful Birth

Since the decision in *Roe v. Wade*<sup>1</sup>, abortion has been a legal option for women. Wrongful birth lawsuits claim that the parent has been harmed by having been deprived of the right to make a timely and informed decision regarding her pregnancy. Parents can claim a right to information about the diagnostic tests available to assess the health of a fetus.

Wrongful birth suits are prompted by the birth of a child with significant disabilities. The list of genetic conditions includes cystic fibrosis<sup>2</sup>, spina bifida<sup>3</sup>, anhidrotic ectodermal dysplasia<sup>4</sup>, and albinism<sup>5</sup>, among others. Failure to inform of abnormalities led parents of a child born with multiple congenital anomalies to sue for failure to notify them of abnormalities detected by prenatal tests<sup>6</sup>. Failure to warn of the risk for problems from rubella exposure has been the basis of many successful lawsuits<sup>7</sup>. The question of whether a healthy infant that carries an adult-onset gene, such as for Huntington disease, is a harm to the parents has not been addressed.

When prenatal testing is undertaken, the guidelines for testing and interpretation of results are the same as for all genetic tests.

### References:

<sup>1</sup>*Roe v. Wade*, 410 U.S. 113 (1973).

<sup>2</sup>*Schroeder v. Perkel*, 432 A.2d 834 (N.J. 1981).

<sup>3</sup>*Reed v. Campagnolo*, 630 A.2d 1145 (Md. 1993).

<sup>4</sup>*Viccaro v. Milunsky*, 551 N.E.2d 8 (Mass. 1990).

<sup>5</sup>*Pitre v. Opelousas General Hospital*, 519 So.2d 105 (La. 1987).

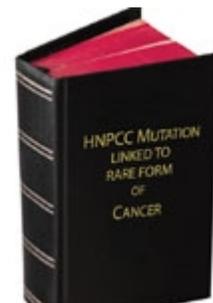
<sup>6</sup>*Bader v. Johnson*, 675 N.E.2d 1119 (Ind. App. 1997).

<sup>7</sup>*Smith v. Cote*, 513 A.2d 341 (N.H. 1986).

# FUTURE CONSIDERATIONS

## Clinical History

A few years later, you read an article in a medical journal indicating that the specific mutation identified in Connie, her uncle Joe, and several other family members appears to also significantly increase the risk for a rare form of cancer that was not previously felt to be associated with HNPCC. Effective screening is available but is only performed when there is an identified increased risk. However, even with screening and early detection, prognosis is poor once diagnosed. Prophylactic options are not available.



## **Points to Consider**

- When new information that may alter management becomes available in medicine, we must consider our duty to recontact patients who may benefit from the knowledge.
- The technical challenges of simply identifying and recontacting patients who may benefit are daunting. But also consider these equally challenging questions: Is this information that Connie gave informed consent to learn? If not, when do the benefits outweigh the potential harms of sharing the new information? How is this duty different if Connie continues to actively be in your care versus being cared for by another?
- This also raises the question: at what point do such findings move from research to the clinical realm of care?

# **ETHICAL PERSPECTIVE**

## **Duty to recontact**

Patients need and expect sufficient and relevant information when medical decisions have to be made. When considering what to do with this new information, one factor may be whether or not you are still Connie's primary care provider at the time you learn about this new association (a few years later). If you have an ongoing physician-patient relationship with Connie, respecting her autonomy by keeping her informed of new or more accurate information about the interpretation of her results is part of her care and specific consent is not required.

## **Former patient records and practicality**

Do former patients, however, have the same need for or right to current, appropriate information? Or suppose you saw Connie only as a consultant and have no plans to see her on a recurring basis. If Connie is no longer actively in your care, we have to compare the benefits to her of receiving such information (increased surveillance for a different form of cancer, psychosocial preparation) against possible harms (anxiety over learning about the risk for another form of cancer that will require more resources for screening and that has a poor prognosis even when detected early). Improved medical care and reduction in diagnostic uncertainty are presumably desirable outcomes for Connie. However, in this case, it is unclear if there would be significant medical gain for her. It would be important to determine if early detection might improve the disease course even minimally. The possible adverse emotional impact on Connie appears significant. She may not have recently thought about the situation or considered other ramifications of her HNPCC results, which has to be considered (nonmaleficence).

The burdens to the professional must also be taken into consideration. In a large practice, it may be practically difficult to manage patient records, necessitating diverting funds for case review and management from other patient services. The principle of justice needs to be considered in this situation.

## **Balance**

To balance these responsibilities, consider establishing reciprocity in the professional-patient relationship from the outset. Inform the patient of his or her obligation to seek periodic updates either from the primary care provider or specialist as well as keeping the professional(s) informed about

changes in contact information. Document this agreement in chart notes, consent forms, and/or patient letters.

## LEGAL PERSPECTIVE

### Duty to Recontact

A physician's general duty of care is considered to include an obligation to advise a patient of any developments in management and treatment. This duty has not yet been extended to former patients. Although under debate, a duty to recall former patients is not part of the standard of care in genetics.

## SUMMARY AND KEY POINTS

### Summary and Key Points

#### Background

Although a significant amount of attention has been paid to the ethical, legal, and social issues that surround human genetic information, much remains to be done in defining policy. However, the application of this genetic information is already being rapidly integrated into clinical practice.

While medical genetics shares many of the ethical and legal considerations as other areas of medicine, factors that may complicate clinical application of genetic knowledge include the following:

- Rapidly advancing knowledge base that is often applied before the clinical implications are well studied
- Information has direct implications for extended family, not simply the individual
- Genetic information may be viewed more personally by the public than other forms of medical information, resulting in potentially significant psychological burden and heightened concerns about privacy and discrimination
- The predictive nature of some forms of genetic testing may be particularly burdensome, especially when there are no effective preventive measures and may lead to an inappropriate view of genetic determinism

#### Ethics

The 4 main biomedical ethical principles that are called upon to identify, evaluate, and resolve ethical dilemmas are as follows:

- **Autonomy:** the potential of the individual to be self-determining
- **Beneficence:** the obligation to seek the good for the patient under all circumstances
- **Nonmaleficence:** the obligation to avoid or refrain from intentionally harming the patient
- **Justice:** on an individual level, we treat our patients in a fair, equitable, and appropriate manner in light of what benefits are due or owed them

#### Legal System

On the **federal** level, there are several sources of laws that govern different aspects of health and medical care. These provide such broad protections as antidiscrimination (e.g., race, gender, disability) and privacy of personal health information. Areas of law that are left solely to the **states** include public health, insurance, and professional licensing.

A **tort** is a civil wrong committed against a person or property. In medicine, negligence torts are most common. There are 4 major components to a negligence tort: duty, breach of duty, causation, and damages incurred. The professional's duty to the patient, and therefore what defines a breach of duty, is commonly the central issue. The duty is typically defined by the standard of care or what is expected of an individual in a given situation.

### **Ethical and Legal Issues in Medical Genetics**

You may want to print or bookmark the following reviews on some of the major ethical and legal issues encountered in the genetic testing process.

- Informed consent
- Genetic privacy and confidentiality
- Genetic discrimination
- Duty to warn
- Testing minors
- Duty to recontact

View this list of steps you can take to protect yourself:

### **Protecting Yourself**

- Be familiar with and adhere to professional standards.
- Provide adequate verbal and written informed consent. Document the content of those discussions and any decisions made.
- Respect the patient's autonomy in decision making. Be aware of your personal biases and how they impact your presentation of information and options.
- Anticipate potential conflicts, such as disclosing risk status to relatives, addressing concerns about privacy, and testing minors. Establish a plan for dealing with these issues **prior** to initiating genetic testing.
- Establish reciprocity in the professional-patient relationship. The professional agrees to keep genetic counseling information current, while the patient agrees to make periodic appointments for updates and to provide current contact information. Document this agreement in chart notes, consent forms, and/or patient letters.
- Refer to or consult with specialists when knowledge, time, or personal values limit ability to manage genetic cases.
- The first responsibility of the health care provider is to identify clients who would benefit from genetic services. This applies whether or not a patient actively seeks medical advice regarding a family history of a possibly genetic condition.
- Once an at-risk individual has been identified, the patient must understand the risk and the options for dealing with this risk.

- If genetic testing and/or other management strategies are available, the patient must provide informed consent. Informed consent requires that a patient be competent, the patient is provided and understands adequate and accurate information about all management options. The consent must be voluntary. This consent process should be well documented.
- Potential conflicts that may be encountered in genetic decision-making should be anticipated and steps taken to avoid poor outcomes. Management goals include:
  - Recognize the complex nature of genetic decision-making, particularly when there is no clear medical benefit driving the decision, and respect a patient's autonomy in the decision-making process.
  - Protect the privacy of genetic information thereby reducing the risk of discrimination and stigmatization
  - Recognize the implications of genetic information for other family members and promote the sharing of risk information within the family. Also, recognize when there exists a duty to warn that requires going outside of the traditional provider-patient confidential relationship.
  - Prepare patients for the possibility of unexpected findings (e.g. ambiguous or otherwise unanticipated results, uncovering misattribution of paternity).
  - Recognize when genetic testing in minors is and is not appropriate and be able to effectively convey this reasoning to parents.
  - Be knowledgeable about the reproductive and prenatal diagnostic options for families with various genetic conditions.
  - Develop an understanding with your patients about their responsibility in maintaining contact and thereby learning of any significant medical advances that may have bearing on them.

These goals may sometimes appear to be in conflict with one another. Ethical consultations can help resolve such dilemmas.

- A healthcare provider that is not comfortable with any of the preceding duties should refer to an appropriate specialist.

## RESOURCES AVAILABLE THROUGH THIS MODULE:

- [American Academy of Family Physicians: Policy and Advocacy](#)[Edit](#)
- [American Academy of Pediatrics Policy Statements Authored by the Committee on Genetics](#)  
The Committee on Genetics studies and makes recommendations to the Board of Directors on recent advances in genetics and provides support to chapters on state legislative issues as they relate to genetics. This is a description of current AAP policy statements authored by the COG.
- [American College of Medical Genetics: Membership Directory](#)  
The American College of Medical Genetics is composed primarily of doctoral (MD, PhD, DO) and master's level (genetic counselors) medical genetics professionals. This is a searchable database of all ACMG members by name or location.
- [Americans with Disabilities Act of 1990](#)  
The website for the Americans with Disabilities Act of 1990 contains a section for other federal agencies with ADA responsibilities, general ADA publications, and other resources.

- [Code of Ethics from the National Society of Genetic Counselors](#)  
Genetic counselors are health professionals with specialized education, training, and experience in medical genetics and counseling. The National Society of Genetic Counselors (NSGC) is the leading voice, authority and advocate for the genetic counseling profession. As such, the NSGC is an organization that furthers the professional interests of genetic counselors, promotes a network for communication within the profession, and deals with issues relevant to human genetics. With the establishment of this code of ethics the NSGC affirms the ethical responsibilities of its members and provides them with guidance in their relationships with self, clients, colleagues, and society. NSGC members are expected to be aware of the ethical implications of their professional actions and to adhere to the guidelines and principles set forth in this code.
- [Constitution of the United States](#)  
The Constitution of the United States comprises the primary law of the U.S. Federal Government. It also describes the three chief branches of the Federal Government and their jurisdictions. In addition, it lays out the basic rights of citizens of the United States. The Constitution of the United States is the oldest Federal constitution in existence and was framed by a convention of delegates from twelve of the thirteen original states in Philadelphia in May 1787. The Constitution is the landmark legal document of the United States. Files are available in ASCII text and Adobe Portable Document Format (PDF)
- [Duty to Recontact](#)   
An article discussing the duty to recontact patients who may benefit from additional knowledge
- [Duty to Warn](#)   
Guidelines to help make the decision to breach patient's confidentiality.
- [Eugenics](#)   
A description of the place of eugenics in genetics knowledge.
- [GeneTests](#)  
The GeneTests website offers an outstanding series of expert-authored GeneReviews that provide important information for clinicians to know about diagnosis, natural history, and genetic testing for genetic conditions. GeneTests.org also maintains databases of genetic testing laboratories and medical genetics clinics. There is no cost to use this website.
- [Genetics and Health Insurance State Anti-Discrimination Laws](#)  
A table which provides a current summary of state laws pertaining to the use of genetic information in health insurance. Restrictions on the use of genetic information in health insurance may address the use of genetic information in individual insurance, group insurance or both.
- [International Code of Medical Ethics of the World Medical Association](#)  
A code of medical ethics which describes the duties of doctors in general, to the sick, and to each other.
- [National Cancer Institute](#)  
National Cancer Institute provides various information. For the general public, patients, and health professionals, they offer consumer-oriented information on a wide range of topics as well as comprehensive descriptions of their research programs and clinical trials. Scientists will find detailed information on specific areas of research interest and funding opportunities. (From Their Website)

- [National Guideline Clearinghouse](#)  
National Guideline Clearinghouse is a public resource for evidence-based clinical practice guidelines. Users may start a search by typing keywords into the search box, or using the NGC Browse or Detailed Search features.
- [Overview of the Privacy Act of 1974](#)  
The "Overview of the Privacy Act of 1974," prepared by the Department of Justice's Office of Privacy and Civil Liberties (OPCL), is a discussion of the Privacy Act's disclosure prohibition, its access and amendment provisions, and its agency recordkeeping requirements. Tracking the provisions of the Act itself, the Overview provides reference to, and legal analysis of, court decisions interpreting the Act's provisions.
- [Principles of Medical Ethics](#)  
The code of medical ethics, from the American Medical Association, discusses opinions on 9 main topics, including social policy, inter professional relationships, hospital relations, fees, physician records, and professional rights and responsibilities.
- [The 5 Elements of Informed Consent](#)   
The 5 Elements of Informed Consent
- [The Genetic Information Nondiscrimination Act of 2008](#)  
A fact sheet on the Genetic The Genetic Information Nondiscrimination Act of 2008, intended for general informational purposes only, provides an explanation of the statute to assist those involved in clinical research to understand the law and its prohibitions related to discrimination in health coverage and employment based on genetic information. The information should not be considered legal advice. In addition, some of the provisions discussed involve issues for which the rules have not yet been finalized, and this information is subject to revision based on publication of regulations.
- [The Health Insurance Portability and Accountability Act of 1996](#)  
The website for the The Health Insurance Portability and Accountability Act of 1996 contains a section for understanding the HIPAA privacy act, statute and rules, how to file a complaint, a news archive, and an FAQ section.
- [The ICN Code of Ethics for Nurses](#)  
The ICN Code of Ethics for Nurses, most recently revised in 2006, is a guide for action based on social values and needs. The Code has served as the standard for nurses worldwide since it was first adopted in 1953.

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