



## CHAPTER 8 : ETHICAL, LEGAL, AND SOCIAL ISSUES

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Over the past decade, many ethical, legal, and social implications (ELSI) associated with genetic testing and research have been raised. In order for genetic testing to be used safely and appropriately, these issues should be discussed with patients so that they are aware of risks and benefits. This chapter provides a brief overview of some of the major ELSI concerns related to genetic testing.

Several concerns have arisen regarding the use and potential misuse of genetic information. Genetic information may differ from other health information because of its long-term implications for an individual and his or her family. Concerns range from the analytical and clinical validity of a genetic test, to potential discrimination by health insurers or employers, to the duty to disclose genetic information to potentially affected family members.

## 8.1 DESCRIPTION OF ELSI ISSUES

To protect patients from additional distress, health care providers should be aware of the relevant ethical, legal, and social issues related to genetics in health care. Genetic specialists may be better able to address patient concerns and questions regarding these issues. A brief discussion of the major ELSI issues related to genetic testing is provided below.

*8.1.1 Communicating Test Results.* It is critical that genetic test results are discussed with patients in an understandable manner. As many genetic tests will not provide simple positive/negative results, but potentially inconclusive results or risk estimates, it is important that patients understand the extent of the information actually provided from a genetic test. Results should be released only to those individuals for whom the test recipient has given consent. The method of communication should be chosen in advance (for example by phone, or in person) to minimize the likelihood that results will be shared with unauthorized persons or organizations. Under no circumstances should results with identifiers be provided to any outside parties, including employers, insurers, or government agencies, without the test recipient's written consent.

*8.1.2 Direct-to-consumer Tests.* A number of companies market genetic tests directly to consumers without requiring physician involvement. Patients should be cautious when considering direct-to-consumer genetic testing and encouraged to discuss this option with their healthcare professional. Some of these companies may play off consumer fears and offer invalidated or bogus tests, or their laboratories may not be properly certified.

*8.1.3 Duty to Disclose.* The results of a genetic test may have implications for a patient's family members. However, health care providers have an obligation to the person being tested not to inform other family members without the permission of the person tested, except in extreme circumstances. If a health professional believes family members may be at risk, the patient may be encouraged to discuss test results with other family members. In general, families are opposed to doctors informing at-risk members without their consent, even in cases where the disease is easily preventable. The duty to inform varies by state, and courts have ruled on differing sides in different cases.



The American Society of Human Genetics suggests that disclosure to at-risk individuals is permissible when the following criteria are met:

- Attempts to encourage disclosure on the part of the patient have failed
- Harm is highly likely, serious, imminent, and foreseeable
- At-risk relatives are identifiable
- Disease is preventable, or medically accepted standards for treatment or screening are available
- The harm from failing to disclose outweighs the harm from disclosure

**8.1.4 Genetic Discrimination.** When considering genetic testing, a major concern often raised is the potential of discrimination based on genetic information. Since genetic test results are typically included in a patient's medical record, patients should be aware that the results may be accessible to others. As a result, genetic test results could affect a person's insurance coverage or employment. More than 30 states have legislation prohibiting genetic discrimination. However, the scope of these protections differs slightly from state to state. As this publication goes to print, no federal legislation has been passed despite several attempts over the last decade.

In addition, members of minority communities often fear that genetic information will be used to stigmatize them. Health providers should be sensitive to the fact that some groups may mistrust the use of genetics as a health tool.

**8.1.5 Informed Consent.** To help ensure that patients understand the risks and benefits of health care choices, informed consent is an important part of the medical decision-making process. For patients considering genetic testing, the following items should be carefully discussed and understood before consent is obtained:

- Testing is voluntary
- Risks, limitations, and benefits of testing or not testing
- Alternatives to genetic testing
- Details of the testing process (for example, what type of sample is required, accuracy of test, turn-around time, etc.)
- Privacy/confidentiality of test results
- Potential consequences related to results including
  - > Impact on health
  - > Possible emotional and psychological reactions
  - > Treatment/prevention options
  - > Ramifications for family

**8.1.6 Privacy.** Genetic information has enormous implications to an individual and his or her family. The privacy of that information is a major concern to patients: in particular, who should have or needs access to that information. In order to protect personal genetic information and to avoid its inclusion in a patient's medical record, some patients may wish to pay for genetic testing out-of-pocket if possible.

**8.1.7 Psychosocial Impact.** Every individual will respond differently to news of his or her genetic test results whether negative or positive. As there is no right or wrong response, health professionals should refrain from judgment and help the patient understand what the test results mean with respect to their own health, available interventions or follow-up, and risks to their family. An individual may respond to genetic information on several levels, the individual level, family level, or on a community and society level. Referrals to genetic counselors, psychologists, or social workers should be made as needed.

**8.1.8 Reproductive Issues.** Genetic information is routinely used to inform reproductive decisions and medical care. Risk factors for genetic conditions for which preconception or prenatal genetic testing may be considered include advanced maternal age, family history, multiple miscarriages, or drug and alcohol exposure. As these procedures carry risks and benefits, parents should carefully consider and discuss these options with a physician or genetic counselor. Providers should take a non-directive stance, especially when the only management option is termination of pregnancy.

**8.1.9 Societal Values.** Genetic information can raise questions about personal responsibility, personal choice versus genetic determinism/fate, and concepts of health and disease. Personal factors, family values, and community and cultural beliefs will influence responses to these issues. While genetic information may influence one individual to change his or her lifestyle or behavior in order to reduce risk or disease severity, others may choose to respond differently. Health professionals should be respectful and sensitive to cultural and societal values and work with the patient to define the appropriate course of action for them with respect to genetic testing and follow-up care.

**8.1.10 Test Utility.** The useful application of genetic tests will depend on the correct interpretation of test results and their utility in guiding medical care and treatment. However, for some genetic conditions, the utility of genetic test results may be limited if no treatment is available or if the results are inconclusive. These issues should be discussed with patients or parents of patients when a genetic test is being considered. Even if a test is not considered to be medically useful, a patient or the family may still gain benefit from testing. Clinical guidelines should be consulted for recommended follow-up care and treatment.

**8.1.11 Test Validity.** Several issues regarding test validity should be considered prior to ordering a genetic test. The analytical and clinical validity of a test are generally measured as test specificity, sensitivity, and predictive value. This information should be shared with the patient as they consider whether or not testing is appropriate for them. Because most genetic tests are offered as services, they are not approved by the Food and Drug Administration. However, genetic tests (or any other clinical laboratory test) should only be ordered from laboratories certified by Clinical Laboratory Improvement Amendments (CLIA).

## SELECTED REFERENCES

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**March of Dimes** Genetics and Your Practice (Financial, Ethical, Legal and Social Issues (FELSI))

<http://www.marchofdimes.com/gyponline/index.bm2>

**U.S. Department of Energy** Ethical, Legal, and Social Issues

[http://www.ornl.gov/sci/techresources/Human\\_Genome/elsi.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/elsi.shtml)

