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

Concerns have arisen regarding the use and potential misuse of genetic information. The unease relates to a range of misuse: from the analytical and clinical validity of a genetic test, to the possible stigma of carrying a genetic difference, to the duty of disclosing genetic information to potentially affected family members.
8.1 Description of Ethical, Legal, and Social Issues

To protect patients from additional distress, healthcare providers should be aware of the relevant ethical, legal, and social issues related to genetics in healthcare. Genetic specialists may be able to address specific patient concerns and questions regarding these issues.

8.1.1 Communicating Test Results. It is critical that genetic test results are discussed with patients in an understandable and compassionate 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 personal 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 offer genetic tests directly to consumers without requiring physician involvement. Patients should be cautious when considering direct-to-consumer genetic testing and are encouraged to discuss this option with their healthcare professional. Some of these companies may play off consumer fears, offer tests with little clinical utility, or not be properly certified or licensed.

8.1.3 Duty to Disclose. The results of a genetic test may have implications for a patient’s family members. However, healthcare 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 differently in response to distinct 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
- Harm from failing to disclose outweighs the harm from disclosure

8.1.4 Genetic Discrimination. When considering genetic testing, the potential for discrimination based on genetic information is a major concern often raised. This fear can impact an individual’s decision to utilize genetic testing services. Since genetic test results are typically included in an individual’s medical record, people should be aware that the results could be accessible to others.

On May 21, 2008, President Bush signed the Genetic Information Nondiscrimination Act (GINA) into law. As the first major new civil rights bill of the new century, GINA protects individuals from discrimination on the basis of genetic information in health insurance and
employment. The health insurance provisions of the law take effect 12 months after the date of signing, in May 2009, and the employment protections take effect 18 months after the date of signage, in November 2009.

In summary, GINA prevents health insurers from denying coverage or adjusting premiums on the basis of genetic information or requesting that an individual undergo a genetic test. Similarly, employers are prohibited from using genetic information to make hiring, firing, or promotion decisions. The law also limits an employer’s right to request, require, or purchase an employee’s genetic information. GINA does not apply to life, disability, or long-term care insurance. Before the federal protections of GINA, more than 40 states established legislation prohibiting genetic discrimination. However, the scope of these protections differs from state to state. GINA does not overturn broader protections provided in some state regulations.

In addition to fears of discrimination in employment and health insurance, members of some communities often fear that genetic information will be used to stigmatize them. Healthcare providers should be sensitive to the fact that some groups may distrust the use of genetics as a health tool.

8.1.5 Informed Consent. To help ensure that patients understand the risks and benefits of healthcare 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:

- Risks, limitations, and benefits of testing or not testing
- Alternatives to genetic testing
- Details of the testing process (e.g., what type of sample is required, accuracy of test, and turn-around time)
- Privacy/confidentiality of test results
- The voluntary nature of testing
- Potential consequences related to results, including: (1) impact on health; (2) emotional and psychological reactions; (3) treatment/prevention options; and (4) ramifications for the family

8.1.6 Privacy. Genetic information has enormous implications for the individual and the family. The privacy of that information is a major concern to patients—in particular, who should have or needs access to that information. To protect personal genetic information and avoid its inclusion in a patient’s medical record, some patients pay for genetic testing out-of-pocket.

8.1.7 Psychosocial Impact. Every individual will respond differently to news of his/her genetic test results, whether negative or positive. As there is no right or wrong response, healthcare professionals should refrain from judgment and help the patient understand the test results with respect to his/her own health, available interventions or follow-up, and risks to his/her family. An individual may respond to genetic information on several levels: individual, family, or community and society. 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, and 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 nondirective stance and support the patients’ decisions.
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. Genetic information may influence one individual to change his or her lifestyle or behavior to reduce risk or disease severity; whereas, 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 him/her 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 treatment is unavailable or 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 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 he or she considers whether or not testing is appropriate for him/her. 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) or another governmental certifying entity.

Selected References


Coalition for Genetic Fairness www.geneticfairness.org


March of Dimes, Genetics and Your Practice www.marchofdimes.com/gyponline/index.bm2