



# GENETICS BRIEF

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## Genetic Testing

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Genes are the fundamental and functional unit of heredity passed down through generations that make individuals unique while at the same time tying families together. Genetic tests analyze an individual's genes, gene products or chromosomes—structures composed of DNA that lie inside a cell's nucleus, and test results reveal the presence or absence of a particular DNA sequence or gene.

Tests for predisposition to adult-onset disorders like breast or ovarian cancer and Huntington disease already are widely used, and well-established state programs screen newborns for a variety of genetic diseases such as phenylketonuria (PKU), hypothyroidism and sickle cell disease. However, these applications only begin to tap into the potential of genetic testing technology. Currently, genetic tests can screen or test for characteristics associated with more than 900 diseases.

### How Genetic Tests Benefit Patients

Genetic tests serve as a useful tool in a number of ways. Medical practitioners might request a genetic test for the following reasons:

- To confirm a diagnosis of an existing condition
- To conduct prenatal testing
- To perform pre-implantation genetic diagnosis to select healthy embryos for implantation
- To screen newborns
- To identify carriers of genetic mutations
- To determine whether an individual may have a genetic predisposition to a disease

No state requires health insurance coverage of genetic testing for adult onset disorders, which may cost more than a thousand dollars, but test results can provide important medical information to the individual undergoing the test and his or her family members. Furthermore, genetic tests can assist couples with reproductive planning by informing prospective parents of genetic risk

#### Genetic Testing in Practice

**Breast and Ovarian Cancer**—Genetic tests for cancer predisposing mutations on the BRCA1 or BRCA2 gene may confirm a diagnosis of hereditary cancer or identify individuals with a mutation. The likelihood of developing cancer varies depending on the penetrance of the mutation within the family and ethnicity but may be as high as 85% by age 70 for some with an alteration in the BRCA1 or 2 gene. Test results combined with family history can help patients choose appropriate preventative measures or treatment.

**Cystic Fibrosis (CF)**—Genetic tests for dozens of mutations on the CFTR gene can be used to confirm a diagnosis, identify carriers of a mutation or conduct prenatal testing. Test results can assist individuals with reproductive planning or make early intervention possible, which may improve the clinical outcome for CF patients, who typically suffer from loss of pulmonary function.

**Hereditary Hemochromatosis**—Genetic tests for two disease-causing mutations on the HFE gene to confirm a diagnosis can alert patients to excess iron absorption, which ultimately may cause organ failure. Early diagnosis and treatment of routine blood removal in pre-symptomatic individuals can prevent complications.

Source: Adapted from GeneTests-GeneClinics: Medical Genetic Information Resource [database online]. University of Washington, Seattle. 1995-. Updated weekly at <http://www.geneclinics.org>. Accessed December 12, 2001.

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factors that offspring may face. Using the knowledge gained from a genetic test to make decisions, however, is often a difficult process. Medical, emotional and financial factors may make the initial choice to test at all very stressful. Because of the complex issues genetic tests raise for individuals and their family members, genetic counseling will likely play a more prominent role in health care as use of the technology grows.

**Limits of Genetic Tests.** Genetic counselors also can help patients understand the limits of genetic tests. In almost all cases, genetic test results do not predict with certainty whether an individual will manifest a condition later in life, with the rare exception of disorders such as Huntington disease. Rather, test results indicate the increased likelihood of illness—if any—based on an individual's genetic makeup. Second, while the number of tests for single-gene disorders—or diseases that are strongly correlated with the presence of a mutation on a single gene—is expanding, testing technology for complex, yet common, genomic disorders like diabetes or heart disease, in which multiple genes and pathways may contribute to the onset of illness, remains at its infancy. Furthermore, genetic testing provides information relative only to the genetic factors that may lead to a disorder, but genetics is merely one determinant of disease. In addition to hereditary factors, environmental variables—such as exposure to toxic substances—and lifestyle choices—like smoking and poor nutrition—play a critical role in the onset of illness. Finally, a genetic test may fail to identify a patient with a mutation because of a false-negative result.

## Legislative Action

**Discrimination and Privacy.** Consumers, health care professionals, policymakers, employers and insurers have expressed concerns about the effects of genetic testing technology—particularly in light of public misconceptions about its true capabilities and limits—on privacy rights, employment and insurance. As a result, the majority of states prohibit genetic discrimination in employment while the federal government forbids genetic discrimination in its workplace. In addition, most states enforce anti-discrimination in health insurance laws; these measures are backed by the Health Insurance Portability and Accountability Act of 1996, which bans genetic discrimination in federally regulated health plans. Finally, two dozen states mandate informed consent requirements to handle genetic information under certain circumstances—either to perform a genetic test, to require a genetic test or to disclose test results.

**Genetic Counselors.** Two states, California and Utah, have anticipated the need for counselors with an expertise in the psychological and medical implications of genetic testing and have passed genetic counselor licensing laws. Some commentators argue that licensing requirements could have a negative impact on the availability of genetic counseling services in the near-term as a result of counselor shortages. Others believe that licensing will help further the profession. Neither any state nor the federal government require insurers to offer or cover the cost of genetic counseling services.

Legislative action in genetics is not expected to slow in the years to come. Not only will states revisit genetics laws as the technology develops—as they have done already—but also policymakers increasingly will be faced with the need to balance patients' desire to access new technologies with consumer safety and ethical concerns as medical professionals incorporate genetic testing into general health practice.

## Selected References

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