status on their present and future lifestyles; those who test negative may manifest survivor guilt. Parents who are found to have a disease-associated mutation often express considerable anxiety and despair as they address the issue of risk to their children. In addition, some individuals consider options such as preimplantation genetic diagnosis in their reproductive decision making.

When a condition does not manifest until adulthood, clinicians and parents are faced with the question of whether at-risk children should be offered genetic testing and, if so, at what age. Although the matter is debated, several professional organizations have cautioned that genetic testing for adult-onset disorders should not be offered to children. Many of these conditions have no known interventions in childhood to prevent disease; consequently, such information can pose significant psychosocial risk to the child. In addition, there is concern that testing during childhood violates a child's right to make an informed decision regarding testing upon reaching adulthood. On the other hand, testing should be offered in childhood for disorders that may manifest early in life, especially when management options are available. For example, children with multiple endocrine neoplasia 2 (MEN 2) may develop medullary thyroid cancer early in life and should be considered for prophylactic thyroidectomy **(Chap. 408)**. Similarly, children with familial adenomatous polyposis (FAP) due to a mutation in *APC* may develop polyps in their teens with progression to invasive cancer in the twenties, and therefore, colonoscopy screening is started between the ages of 10 and 15 years **(Chap. 110)**.

INFORMED CONSENT

Informed consent for genetic testing begins with education and counseling. The patient should understand the risks, benefits, and limitations of genetic testing, as well as the potential implications of test results. Informed consent should include a written document, drafted clearly and concisely in a language and format that is understandable to the patient. Because molecular genetic testing of an asymptomatic individual often allows prediction of future risk, the patient should understand all potential long-term medical, psychological, and social implications of testing. There have long been concerns about the potential for genetic discrimination. The Genetic Information Nondiscrimination Act (GINA) was passed in 2008 and provides some protections related to job and health insurance discrimination. It is important to explore with patients the potential impact of genetic test results on future health as well as disability and life insurance coverage. Patients should understand that alternatives remain available if they decide not to pursue genetic testing, including the option of delaying testing to a later date. The option of DNA banking should be presented so that samples are readily available for future use by family members, if needed.

FOLLOW-UP CARE AFTER TESTING

Depending on the nature of the genetic disorder, posttest interventions may include: (1) cautious surveillance and awareness; (2) specific medical interventions such as enhanced screening, chemoprevention, or risk-reducing surgery; (3) risk avoidance; and (4) referral to support services. For example, patients with known deleterious mutations in *BRCA1* or *BRCA2* are strongly encouraged to undergo risk-reducing salpingo-oophorectomy and are offered intensive breast cancer screening as well as the option of risk-reducing mastectomy. In addition, such women may wish to take chemoprevention with tamoxifen, raloxifene, or exemestane. Those with more limited medical management and prevention options, such as patients with Huntington's disease, should be offered continued follow-up and supportive services, including physical and occupational therapy and social services or support groups as indicated. Specific interventions will change as research continues to enhance our understanding of the medical management of these genetic conditions and more is learned about the functions of the gene products involved.

Individuals who test negative for a mutation in a disease-associated gene identified in an affected family member must be reminded that they may still be at risk for the disease. This is of particular importance for common diseases such as diabetes mellitus, cancer, and coronary artery disease. For example, a woman who finds that she does not carry the disease-associated mutation in *BRCA2* previously discovered in the family should be reminded that she still requires the same breast cancer screening recommended for the general population.

GENETIC COUNSELING AND EDUCATION

Genetic counseling should be distinguished from genetic testing and screening, although genetic counselors are often involved in issues related to testing. Genetic counseling refers to a communication process that deals with human problems associated with the occurrence of risk of a genetic disorder in a family. Genetic risk assessment is complex and often involves elements of uncertainty. Counseling, therefore, includes genetic education as well as psychosocial counseling. Genetic counseling can be useful in a wide range of situations **(Table 84-1)**. The role of the genetic counselor includes the following:

1. Gather and document a detailed family history.
2. Educate patients about general genetic principles related to disease risk, both for themselves and for others in the family.
3. Assess and enhance the patient's ability to cope with the genetic information offered.
4. Discuss how nongenetic factors may relate to the ultimate expression of disease.
5. Address medical management issues.
6. Assist in determining the role of genetic testing for the individual and the family.
7. Ensure the patient is aware of the indications, process, risks, benefits, and limitations of the various genetic testing options.
8. Assist the patient, family, and referring physician in the interpretation of the test results.
9. Refer the patient and other at-risk family members for additional medical and support services, if necessary.

Genetic counseling is generally offered in a nondirective manner, wherein patients learn to understand how their values factor into a particular medical decision. Nondirective counseling is particularly appropriate when there are no data demonstrating a clear benefit associated with a particular intervention or when an intervention is considered experimental. For example, nondirective genetic counseling is used when a person is deciding whether to undergo genetic testing for Huntington's disease. At this time, there is no clear benefit (in terms of medical outcome) to an at-risk individual undergoing genetic testing for this disease because its course cannot be altered by therapeutic interventions. However, testing can have an important impact on the individual's perception of advanced care planning and his or her interpersonal relationships and plans for childbearing. Therefore, the decision to pursue testing rests on the individual's belief system and values. On the other hand, a more directive approach is appropriate when a condition can be treated. In a family with FAP, colon cancer screening and prophylactic colectomy should be recommended for known *APC* mutation carriers. The counselor and clinician following this family must ensure that the at-risk family members have access to the resources necessary to adhere to these recommendations.

Genetic education is central to an individual's ability to make an informed decision regarding testing options and treatment. An adequate knowledge of patterns of inheritance will allow patients to understand the probability of disease risk for themselves and other family members. It is also important to impart the concepts of disease penetrance and expression. For most complex adult-onset genetic

TABLE 84-1 INDICATIONS FOR GENETIC COUNSELING

Advanced maternal age (>35 years)
Consanguinity
Previous history of a child with birth defects or a genetic disorder
Personal or family history suggestive of a genetic disorder
High-risk ethnic groups
Documented genetic alteration in a family member
Ultrasound or prenatal testing suggesting a genetic disorder