Referrals to genetic specialists should be considered if a healthcare provider suspects a patient is at risk for or affected with a genetic disorder. Genetic specialists can help identify the appropriate tests to order, consider the family history, and provide information about the treatment and long-term outcomes for patients diagnosed with a genetic disorder. They may recommend a referral or referrals to other medical specialists. This chapter provides a brief overview of points to consider when deciding if a referral to a genetic specialist is appropriate.

A referral to or consultation with a genetic specialist may be indicated for several reasons. In general, a consultation with a genetic specialist should be considered if a hereditary condition is suspected. Symptoms that may suggest a genetic disorder are listed in Section 2.2: Red Flags for Genetic Disease. Specific clinical guidelines are available for some conditions such as cancer and diabetes.
6.1 When to Refer to a Genetic Specialist

Patients meeting any of the following criteria should be considered for referral to a genetic specialist:

6.1.1 Family History

- One or more members with mental retardation, developmental disability, an inherited disorder, or a birth defect
- One or more members with early deaths due to known or unknown medical conditions
- One or more members with adult-onset health conditions such as cardiovascular disease, dementia, or cancer, particularly if onset is early in adulthood
- Couples who would like testing or more information about genetic conditions that occur with higher frequency in their ethnic group

6.1.2 Delayed Growth and Development

- Those who have or are concerned that their child has developmental delays that may be due to an inherited disorder or birth defect
- Parents whose infant has a genetic disease diagnosed by newborn screening

6.1.3 Reproductive Issues

- Women who are interested in genetic testing or screening
- Women who have experienced multiple pregnancy losses or babies who died in infancy
- People concerned that their jobs, lifestyles, or medical history may pose a risk to the outcome of a pregnancy. Common causes of concern include exposure to radiation, medications, illegal drugs, chemicals, or infections
- Couples who are first cousins or other close blood relatives
- Pregnant women whose ultrasound examinations or blood testing indicate that their pregnancy may be at increased risk for certain complications or birth defects

A genetic specialist can provide assistance in several ways: a formal or informal consultation, genetic counseling session, or genetic evaluation. A genetic specialist can provide an accurate assessment or confirm the diagnosis of a genetic disease. A diagnosis may be made primarily through genetic testing or a combination of testing, clinical examination, and family history. Genetic specialists are able to provide management options or referrals.
to specialists as needed; provide advice to primary-care practitioners about a genetic condition, prognosis, treatment, and long-term outcome; and recommend educational materials to patients and families.

The primary genetic specialists considered for referral are clinical geneticists and genetic counselors. Although these specialists can play a major role in the diagnosis and education of family members with a genetic disorder, other medical specialists such as surgeons, cardiologists, or ophthalmologists may be required for appropriate treatment or intervention. The services of other allied health professionals such as nutritionists, social workers, psychologists, and occupational and physical therapists may also be necessary. The requirements for a referral will vary from system to system. In general, though, a genetic referral requires the following information:

- Patient information
- Name and address of the referrer
- Reason for the referral
- Information about the suspected diagnosis, if known
- Family history

**Selected References**

GeneTests  
[www.genetests.org](http://www.genetests.org)

March of Dimes, Genetics and Your Practice  

National Cancer Institute, Cancer Genetics Service Directory  
[www.cancer.gov/search/genetics_services](http://www.cancer.gov/search/genetics_services)

National Society of Genetic Counselors  
[www.nsgc.org/resourcelink.cfm](http://www.nsgc.org/resourcelink.cfm)