



CHAPTER 6 : INDICATIONS FOR A GENETIC REFERRAL

Referrals to genetic specialists should be considered if a physician suspects a patient is at risk of or is affected with a genetic disorder. Genetic specialists can help identify the appropriate tests to order (genetic or additional laboratory tests), consider the family history, and provide information about the treatment and long-term outcomes for patients diagnosed with a genetic disorder, including recommendations 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 Chapter 3 (“Red flags for genetic disease”). For conditions such as cancer and diabetes, specific clinical guidelines are available.

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, 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 and cancer, particularly if onset is at an early age
- Couples who would like testing or more information about genetic conditions that occur with higher frequency in their ethnic group

6.1.2 Developmental Delay/Growth

- 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 routine newborn screening

6.1.3 Reproductive Issues

- Women who are or are planning to be pregnant after age 35
- Women who have experienced multiple pregnancy losses, including babies who died in infancy
- People concerned that their jobs, lifestyles or medical history may pose a risk to outcome of 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 indicates that their pregnancy may be at increased risk for certain complications or birth defects





A genetic specialist can provide assistance through a variety of ways—a formal or informal consultation, a genetic counseling session, or a genetic evaluation. A genetic specialist can provide a more accurate assessment of the risk 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 to be considered for referral are clinical geneticists and genetic counselors. While these specialists can play a major role in the diagnosis and education of family members with a genetic disorder, other medical specialists may be required for

appropriate treatment or intervention such as surgeons, nutritionists, social workers, psychologists, and occupational therapists. 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

Cancer Genetics Service Directory http://www.cancer.gov/search/genetics_services/

March of Dimes—Genetics and Your Practice <http://www.marchofdimes.com/gyponline/index.bm2>

National Society of Genetic Counselors <http://www.nsgc.org/resourcelink.cfm>