



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 that may indicate when a genetics referral 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.

Patients meeting any of the following criteria should be considered for recommendation to a genetics specialist.

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 early
- Couples who would like testing or more information about genetic conditions that occur frequently in their ethnic group

DEVELOPMENTAL DELAY/GROWTH

- Those who have or are concerned that their child might have an inherited disorder or birth defect due to developmental delay or failure to reach milestones
- Couples whose infant has a genetic disease diagnosed by routine newborn screening

REPRODUCTIVE ISSUES

- Women who are pregnant or planning to be 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 the 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 indicate that their pregnancy may be at increased risk for certain complications or birth defects





A genetics specialist can provide assistance through a variety of ways—a formal or informal consultation, a genetic counseling session, or a genetic evaluation. A genetics 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. They are able to provide management options or referrals to specialists as needed depending on the disease, advice to primary care practitioners about a genetic condition, prognoses, treatment and long-term outcome, and recommend educational materials to patients and families.

The primary genetics 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 of 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 genetics referrals require 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>

