

It is nurses' responsibility to learn basic genetic principles, to be alert to situations in which families could benefit from genetic evaluation and counseling, to know about special services that can help manage and support affected children, and to be familiar with facilities in their areas where these services are available. In this way, nurses are able to direct individuals and families to needed services and be active participants in the genetic evaluation and counseling process. A regularly updated resource for locating genetics clinics can be found at <http://ghr.nlm.nih.gov/handbook/> (click on link for Genetic Consultation). In addition, state health departments either offer services or can help identify health professionals with specialty training in genetics.

Early identification of a genetic disorder allows anticipation of associated conditions and implementation of available preventive measures and therapy to avoid potential complications and to enhance the child's health. It may also prevent the unexpected birth of another affected child in the immediate or extended family. Nurses have an important role in identifying patients and families who have or are at risk for developing or transmitting a genetic condition (see [Box 3-4](#)). When facilitating genetics consultations, nurses should share with the genetics professional the findings in the histories they collected that triggered the consultation. Nurses can also help the referral process by determining and communicating the family's initial concerns, their state of knowledge about the reason for referral, and their attitudes and beliefs concerning genetics.

Genetic evaluation for diagnostic purposes may occur at any point in the life span. In the newborn period, birth defects and abnormal newborn screen results are obvious reasons for referral. Beyond the newborn period, indicators for referral include metabolic disorders, developmental delays, growth delays, behavioral problems, cognitive delays, abnormal or delayed sexual development, and medical problems known to be associated with genetic diseases. For example, a preschooler with hyperactivity and autistic-like behaviors may need evaluation for fragile X syndrome, and a 17-year-old girl with primary amenorrhea and short stature should be evaluated for Turner syndrome.

With so many recent advances in genetic testing, it is not unusual for a child or adult with longstanding medical problems, including