Repeated pregnancy loss or infertility

Newly diagnosed abnormality or genetic condition

Before undertaking genetic testing and after receiving results, particularly when testing for susceptibility to late-onset disorders, such as cancer or neurologic disease

As follow-up for a positive newborn test, as with phenylketonuria, or a heterozygote screening test, such as Tay-Sachs disease

From Nussbaum R, McInnes R, Willard H: *Thompson and Thompson genetics in medicine,* ed 6, Philadelphia, 2007, Saunders/Elsevier.

Maintaining contact with the family or referring the family to an agency that can provide a sustained relationship, usually the public health agency in their locality, is one of the most important aspects in the care of the patient and family. In a disorder that requires conscientious diet management, such as PKU or galactosemia, it is important to make certain that the family understands and follows the advice. A vital role for nurses is to advocate for the child and family as they make their way through the various specialty clinics. This is especially important for families that are more vulnerable because of cognitive, hearing, language, or financial issues and those who otherwise may have difficulty accessing health services. Nurses can reinforce the genetic information or arrange for additional genetic counseling if a family has additional questions or misunderstandings.

One of the current ethical concerns is the testing of healthy children for carrier status of a genetic condition that either will not have adverse consequences until adulthood or only has reproductive implications. The American Academy of Pediatrics, Committee on Bioethics (2001, reaffirmed 2008) policy statement does not support the broad use of carrier testing or screening in children or adolescents. When there is no clear medical benefit to testing in childhood, the child should be permitted to wait until adulthood to choose whether or not to be tested. Genetic counseling is recommended to help the family weigh all of the issues.

Psychological Aspects of Genetic Disease