Genetic Evaluation and Counseling

Genetic counseling is a communication process concerned with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. It involves relaying information about the diagnosis, treatment options, recurrence risk, and availability of prenatal diagnosis. With the completion of the Human Genome Project, the international project to determine the total genetic information in humans, a new era of human genetics is unfolding (International Human Genome Sequencing Consortium, 2004), and it will lead to a better understanding of specifically how genetic variation contributes to health and disease. It is essential that nurses master the basic principles of heredity, understand how heredity contributes to disorders, and be aware of the types of genetic testing available (Table 8-14).

TABLE 8-14

Types of Genetic Testing

Test and Method	Specimen	Indication	Comments
Chromosome analysis (karyotyping)	Blood, skin, amniocytes, bone marrow	Detection of chromosomal abnormality, sex determination, cancer classification	Almost 100% accuracy for whole or partial chromosomal abnormality; will not detect microdeletions or duplication (submicroscopic chromosome segments), single-gene defects, or multifactorial disorders
Fluorescence in situ hybridization (FISH)	Blood, skin, amniocytes, bone marrow	Detection of microdeletion or duplications of chromosome segments (not visible by chromosome analysis)	A technique that is a cross between chromosome analysis and single-gene DNA tests
Direct DNA mutation detection (polymerase chain reaction, Southern blot, gene sequencing)	Blood, skin, amniocytes	Detection of gene mutation(s) in affected individual for diagnosis, in unaffected carrier, or for presymptomatic diagnosis	Gene location must be mapped, and disease-producing mutations must be characterized; can test single individual
Indirect DNA linkage studies (restriction length fragment polymorphisms, microsatellites, genetic markers)	Blood	Prediction of carrier or presymptomatic status based on inheritance of same chromosome segment as in known affected individual	Must test several family members, including one or two confirmed affected individuals, for testing to be valid
Biochemical	Blood, skin, amniotic fluid,	Detection of metabolic pathway errors, enzyme defects,	Results may be difficult to interpret if partial pathway error or modified substrate is present