

them understand the chances that a subsequent pregnancy will be affected and will not be affected, and ensure they have been given information about their options for future children (preimplantation diagnosis, use of donor egg or sperm, prenatal diagnosis, or adoption). Families often try to reason that some unrelated event caused the abnormality (e.g., a fall, a urinary tract infection, or “one glass of wine”) before the mother was aware that she was pregnant. These misconceptions need to be assessed and dispelled.

After a genetics visit, and sometimes before the visit, parents often use the Internet to find answers to their questions. During the initial genetics evaluation, a diagnosis may not be possible. Instead, findings in medical, developmental, and family histories lead the professional to order genetic tests and other diagnostic procedures. Diagnoses under consideration are discussed briefly with the parents. Some parents are satisfied with the brief information and do not care to find out more until the actual diagnosis is established. Other parents go home and seek as much information as they can about the diagnoses under consideration. The information they find can be terrifying and overwhelming and inaccurate or misleading. Nurses can play an important role in helping parents identify reliable, accurate resources for information at whatever time they desire it. It is also important to stress that everything that is described for a genetic condition may not be relevant to their child. Before the follow-up genetics visit when test and procedure results are discussed, nurses can help parents identify and write down the questions and concerns they need addressed before leaving the clinic.

After a genetic diagnosis is made or a genetic predisposition to a delayed-onset disorder is identified, nurses need to have frequent contact with patients and families as they attempt to incorporate recommended therapies or disease-prevention strategies into their daily lives. For example, a disorder such as PKU requires conscientious diet management; therefore, it is important to make certain that the family understands and follows instructions and is able to navigate the health care system to access the essential formula and low-phenylalanine food products. An infant evaluated for cleft palate and cardiac defect and subsequently found to have VCFS requires surgical intervention for the congenital