
The Genetic Counselor as a Partner in Reproductive Health

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Genetic counselors affect peoples' lives in very personal and profound ways. For pregnant women and their partners, the counselor's role is to critically evaluate a patient's family history, provide risk analysis, and present current information on genetic testing options. This often involves complex medical information that must be presented in terms that can be clearly understood so that patients can make informed decisions. Prenatal genetic testing has made it possible to provide expectant parents with reassurance regarding specific genetic disorders. However, the risk for birth defects is 3-5%, and many of these cannot be detected prenatally.

In the early days of prenatal diagnosis, chromosome abnormalities and a few metabolic disorders could be detected. Now, there are hundreds of disorders for which prenatal diagnosis may be available, either through chorionic villus sampling, amniocentesis or targeted ultrasound. While this allows a couple to make informed decisions based on the best available information, it also means that many more couples face difficult decisions when a fetal abnormality is suspected or confirmed. When a particular genetic disorder is identified, the genetic counselor can provide options for this pregnancy and for future pregnancies.

Genetic counselors are non-directive, meaning that they do not impose their own views, but rather help couples through the decision making process. Parents may be encouraged to envision different scenarios, and consider which alternatives are the most or least acceptable based on personal or religious reasons. Options for future pregnancies may include preimplantation genetic diagnosis, gamete donation, prenatal diagnosis, the choice not to bear children, or adoption.

There are many reasons why a woman and her partner might seek genetic counseling. In some cases, the decision to start a family or learning that she is pregnant may prompt a woman to ask certain questions. For example:

- I have a brother with mental retardation and some autistic behaviors. Could this have any significance for my child?
- I am 40 years of age and I have heard that the risk for certain birth defects increases with age. What is my risk and what are my options?
- I just found out that I am pregnant and I am concerned about the medications that I take. Will they harm the baby?

- My husband and I are both of Ashkenazi Jewish descent. I read that certain disorders are seen more commonly in infants born to Jewish couples. Should we be tested? We already have two healthy children.
- My sister's baby was just diagnosed with cystic fibrosis. What is the chance that my baby will have CF?
- I want to have children, but my mother has Huntington disease, and I definitely do not want to pass this gene on to my children, but on the other hand, I don't think I am ready to find out if I inherited the gene. What can I do?

When these questions come up in a busy obstetric practice, there may not be enough time to adequately address them. Genetic counselors have time not only to provide the information being sought, but also to explore what the information means to the parents so that they can make the best possible decision for themselves and their family.

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