Chapter 7

Genetic Technologies

7.2 Genetic Testing

Many prospective parents have questions about different inherited diseases and the possibility of genetic testing. For example, what is their risk of having a child with a genetic disease? What tests are there that show whether a developing fetus has a genetic disease? What is it like to raise a child with a genetic disease? Trained genetic counselors can help people answer questions like these.

Genetic counseling is especially important for couples with a family history of genetic disease. Genetic counselors often begin by creating a *pedigree*, a family tree that shows which relatives are affected by genetic disease. A sample family tree for the occurrence of Tay–Sachs disease within a family is shown in Figure 7.1. Information about the inheritance pattern of the disease in question—for example, whether the disease allele is dominant or recessive—can then be used to assess a couple's risk of having an affected child.

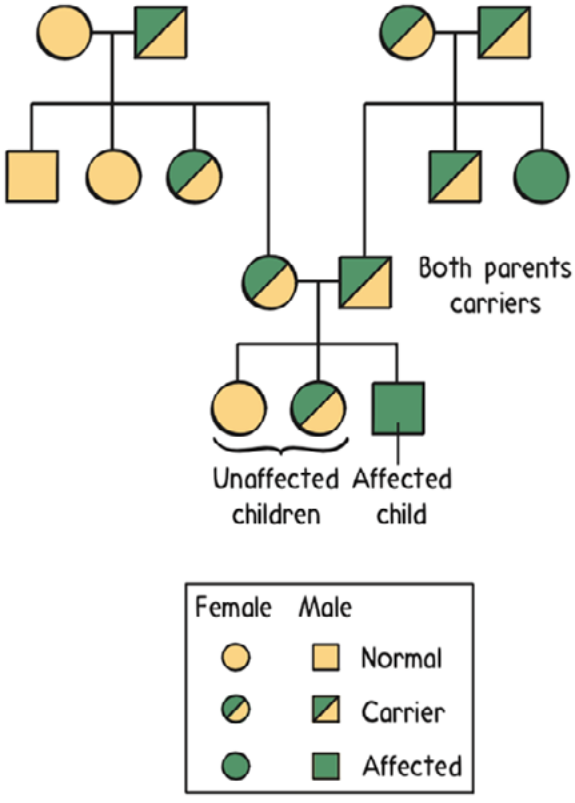

FIGURE 7.1

This three-generation family tree (pedigree) shows the occurrence of Tay–Sachs disease within a family. The allele for Tay–Sachs disease behaves as a recessive—that is, only people who are homozygous for the allele are affected. This type of analysis allows a genetic counselor to assess the risk of genetic disease in a family.

For some diseases, medical tests can provide additional information. Those with a family history of Tay–Sachs disease (a fatal disease of the central nervous system) or cystic fibrosis (a serious disease characterized by mucus buildup in the lungs, digestive system, and other organs) can undergo simple tests to determine whether they are *carriers* of the disease—that is, whether they possess a disease allele in addition to a normal allele. Because these diseases affect only people who have two recessive alleles, a couple is at risk of having an affected child only if both parents are carriers.

If both parents are in fact carriers, procedures such as *chorionic villus sampling* or *amniocentesis*, in which fetal cells are collected and examined during pregnancy, can be used to determine whether a fetus is affected. Couples may also have the option of undergoing *in vitro* fertilization, testing the embryos, and implanting only unaffected ones.

Genetic testing is also useful for families without a history of genetic disease. For Down syndrome (Figure 7.2), a condition associated with mental retardation and other health issues,
risk depends primarily on the age of the mother. Older women have a greater risk of having children with Down syndrome. Down syndrome is caused by trisomy 21, the presence of three copies of chromosome 21 instead of two. Tests during pregnancy can look at the chromosomes in a fetus's cells to determine whether they show trisomy 21.

FIGURE 7.2

Down syndrome occurs in about one out of every 1000 births. It is caused by trisomy 21.

READING CHECK

What does it mean for someone to be a "carrier" of a genetic disease?

CHECK YOUR ANSWER

For genetic diseases that are caused by recessive alleles, a carrier is a person who possesses one disease allele and one normal allele. Carriers are not affected by the disease themselves, but they are able to pass a disease allele to their offspring, who will inherit the disease if they also inherit a disease allele from the other parent.

For more information on family pedigrees as well as the opportunity to create your own, check out the following website:

https://www.genome.gov/Pages/Education/Modules/YourFamilyHealthHistory.pdf