

In the fourth month of pregnancy, a hypodermic needle is used to obtain a small sample of amniotic fluid from the mother. The amniotic fluid, which bathes the fetus, contains free-floating cells derived from the fetus. Once removed, these cells can be cultured in the laboratory and used to test for many of the most common genetic disorders.

During amniocentesis, the position of the needle in relationship to the fetus is observed by means of a technique called *ultrasound*. Pulses of high-frequency sound waves are sent into the body and are reflected back in various patterns depending on the tissues or fluids the waves hit. These patterns of sound-wave reflections are then mapped to produce a picture of inner tissues (Figure 14.19b). This allows the person performing the amniocentesis to avoid damaging the fetus. Ultrasound also allows the fetus to be examined for the presence of major abnormalities.

Today, using ultrasound to guide them, physicians are able to sample fetal skin and certain other tissues. Cells from the outermost fetal membrane, the chorion, can also be sampled by a technique known as *chorionic villus sampling (CVS)*. To perform CVS, a physician inserts a thin tube or a needle into the vagina and up into the uterus, vacuuming up fingerlike projections of the chorion called villi. This procedure can be performed in the second to third month of pregnancy.

The options for treating fetal abnormalities, though increasing, are still limited. Open fetal surgery was first performed at the University of California at San Francisco in 1981. Various surgical pro-

cedures are conducted today to treat fetal urinary obstructions, certain respiratory problems, and certain growths. The use of laser scalpels and new fetal imaging technologies such as fetoscopic cameras are allowing surgeons to treat such disorders less invasively. In some cases, surgery can be performed without opening the uterus. Gene therapy, the treatment of a genetic disorder by the insertion of “normal” copies of a gene into the cells of a patient carrying “defective” copies of the gene (see Chapter 11), may also be used.

At this time, however, most of the problems that can be diagnosed by pedigree analysis, amniocentesis, chorionic villus sampling, and ultrasound techniques cannot be treated. Sometimes the only options available to a couple are to continue the pregnancy and deal with the problems after birth or to have a therapeutic abortion. Continued progress in developing gene therapies and laser technologies may, in the future, have a dramatic effect on a physician’s ability to treat genetic disorders before birth.

CONCEPT CHECKPOINT

14. When a genetic test is performed, for what does it test?
15. How is amniocentesis performed? How does the use of ultrasound help in the procedure? Is amniocentesis a genetic test?

CHAPTER REVIEW

Summary of Key Concepts and Key Terms

How are human traits inherited?

- 14.1 Some human traits are caused by dominant or recessive alleles and are inherited in a Mendelian fashion.
- 14.2 In traits exhibiting **incomplete dominance** (p. 000), alternative forms of a trait are neither dominant nor recessive; heterozygotes are phenotypic intermediates.
- 14.3 In traits exhibiting codominance (**codominant alleles**, p. 000), alternative forms of an allele are both dominant; heterozygotes exhibit both phenotypes.
- 14.4 Some genes consist of a system of alleles, or **multiple alleles** (p. 000), which are usually codominant to one another.
- 14.4 The ABO system of human blood types is an example of multiple alleles, with two of the alleles (*A* and *B*) codominant to one another and one allele (*O*) recessive to the other two.
- 14.5 Many traits are controlled by more than one gene, which is called **polygenic inheritance** (p. 000).
- 14.5 Polygenic inheritance can be identified by its continuous nature and includes traits such as skin color, height, and eye color.
- 14.6 Environmental conditions can influence the phenotypic expression of a gene.

How are human genetic disorders inherited and studied?

- 14.7 A few human genetic disorders are caused by dominant alleles.
- 14.7 Dominant alleles that lead to severe genetic disorders are less common; in some of the more frequent ones, the expression of the alleles does not occur until after the individuals have reached their reproductive years.
- 14.8 Many of the most common genetic disorders are associated with recessive alleles, the functioning of which may lead to the production of defective versions of enzymes that normally perform critical functions.
- 14.8 Because recessive traits are expressed only in homozygotes, the alleles are not eliminated from the human population.
- 14.9 Some human genetic disorders are caused by **sex-linked (X-linked) alleles** (p. 000).
- 14.9 Human cells contain 46 chromosomes: 44 **autosomes** (AW-tuh-somes, p. 000) and 2 **sex chromosomes** (p. 000); the autosomes form 22 pairs of homologous (matched) chromosomes.
- 14.10 Patterns of inheritance observed in family histories, or **pedigrees** (PED-uh-greez, p. 000), can be used to determine the mode of inheritance of a particular trait.
- 14.10 Pedigree analysis can often determine whether a trait is associated with a dominant or a recessive allele and whether the gene determining the trait is sex linked.