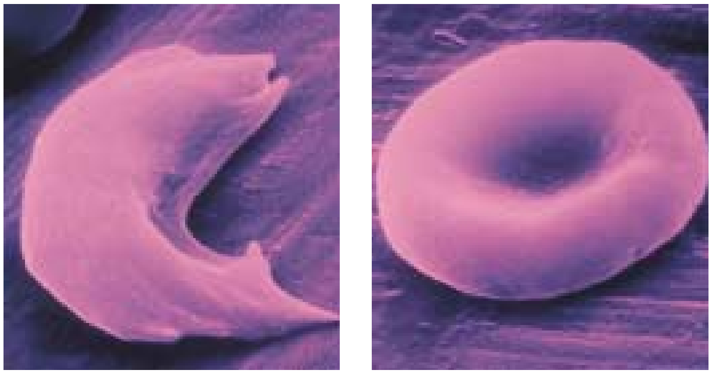

(*a*) (*b*)

Figure 14.7 (*a*) Sickle-shaped red blood cell from person afflicted with sickle cell anemia. (*b*) Normal human red blood cell.

thin mucus and move it toward the larger airways where it can be cleared by coughing or suctioning.

Sickle cell anemia

Sickle cell anemia is a recessive disorder most common among African blacks and their descendants. This disorder is caused by a mutant HBB gene, which provides instructions for making a part of the hemoglobin molecule in red blood cells. In the United States, about 9 percent of blacks are heterozygous for this mutant allele; about 0.2 percent are homozygous and therefore have sickle cell anemia. In some groups of people in Africa, up to 45 percent of the individuals are heterozygous for this allele. Heterozygous carriers of the sickle cell (mutant HBB) gene do not have the disease but can pass on the gene to their children.

Individuals affected with sickle cell anemia are unable to transport oxygen to their tissues properly because the oxygen-carrying hemoglobin molecules are defective. Red blood cells that contain large proportions of such defective molecules become sickle shaped (**Figure 14.7*a***) and stiff; normal red blood cells are disk shaped (Figure 14.7*b*) and much more flexible. As a result of their stiffness and irregular shape, the sickle-shaped red blood cells are unable to move easily through capillaries. Therefore, they tend to accumulate in blood vessels, reducing the blood supply to the organs they serve and causing pain, tissue destruction, and an early death.

The gene for sickle cell anemia is most prevalent in the regions of Africa where malaria is prevalent. Malaria is a disease caused by microorganisms that live in a person's red blood cells. These microbes are injected into a person's bloodstream by the bite of a female *Anopheles* mosquito. A long-lasting disease, malaria affects the physical and mental development of its victims, causing damage to many body organs. Scientists have discovered that the defective hemoglobin molecules of the person with sickle cell anemia produce conditions that are unfavorable to the growth of the malaria organism, but these persons eventually die of their anemia. However, persons heterozygous for the sickle cell gene, though unaffected by sickle cell anemia, are more resistant to malaria than persons with no sickle cell gene. Heterozygotes, then, have a survival advantage with respect to malaria and do not have sickle cell anemia, although their offspring may be affected by the disease if they reproduce with persons also carrying the defective gene.

Tay-Sachs disease

Tay-Sachs disease is an incurable, fatal recessive disorder caused by a mutation in a gene called hex A that codes for the production of the enzyme hexosaminidase A. This enzyme is necessary for breaking down certain fatty substances in brain and nerve cells. In persons with the mutation, these substances build up and and gradually destroy brain and nerve cells, until the entire central nervous system stops working.

Although rare in most human populations, Tay-Sachs has a high incidence among Jews of Eastern and Central Europe and among American Jews, 90 percent of whom are descendants of Eastern and Central Europeans. Geneticists estimate that, within these Jewish populations, 1 in 28 individuals carries the allele for this disease and approximately 1 in 3600 infants is born with the disorder.

The most common form of the disease affects infants. Affected children appear unaffected at birth but begin to show signs of mental deterioration at about 8 months of age. As the brain begins to deteriorate, affected children become blind; they usually die by the age of 5 years.

CONCEPT CHECKPOINT

7. Explain why parents who are heterozygous for a dominant disorder could be expected to pass on the trait to 50 percent of their children.
8. Draw a Punnett square to show the inheritance of sickle cell anemia in the offspring of parents who are both carriers of the sickle cell allele. Use *s* to show the sickle cell allele and *S* to show the normal allele. What is the probability of their having a child with the disease?

14.9 Some human genetic disorders are caused by sex-linked alleles.

Humans have 23 pairs of chromosomes: a pair of **sex chromosomes** and 22 pairs of **autosomes** (**AW**-tuh-somes). The sex chromosomes determine a person's sex; a female has two X chromosomes and a male has an X and a Y chromosome, just as Morgan discovered in fruit flies (see Chapter 13). Autosomes, or non-sex chromosomes, carry the majority of an individual's genetic information but do not determine sex. They are the same in both sexes. Thus far in this chapter, the genes coding for the traits and disorders discussed have all been autosomal.

Almost all **sex-linked alleles** and disorders are really *X-linked alleles and disorders*, because the alleles for these disorders are found on the X chromosome. Y-linked disorders are rare. The reason is that the human Y chromosome is much smaller than the X chromosome and carries few genes. Therefore, recessive X-linked disorders appear more frequently in males than in females because males need to inherit only one mutant allele, not two, for the trait to be expressed. Some recessive X-linked disorders are red-green color blindness, hemophilia, and Duchenne muscular dystrophy.

Persons with *red-green color blindness* cannot distinguish red from green. This problem is due to inheriting a mutant allele located on the X chromosome that causes a defect in either the red or