14.11 A **karyotype** (KAIR-ee-oh-type, p. 000) is a picture of chromosomes paired with their homologues and arranged according to size.
14.11 Researchers often use karyotypes to study the inheritance of genetic disorders caused by the loss of all or part of a chromosome or by the addition of extra chromosomes or chromosome fragments.
14.12 Errors in chromosome number usually result from **nondisjunction** (non-dis-JUNK-shun, p. 000) in meiosis.
14.12 Nondisjunction occurs at either the first or second meiotic division when homologous chromosomes fail to separate.
14.13 In humans, the inheritance of one autosome too few usually results in a nonviable zygote.
14.13 The inheritance of one autosome too many, or a **trisomy** (TRI-some, p. 000), often results in a nonviable zygote (with the exception of chromosomes 13, 18, 21, and 22).
14.13 Individuals with an extra copy of chromosome 21 have Down syndrome, a disorder much more frequent among children born to mothers older than 35 years.
14.14 Persons who inherit abnormal numbers of sex chromosomes often have abnormal features and may exhibit intellectual deficits.
14.14 Examples of genetic disorders caused by abnormal numbers of sex chromosomes are triple X females, XXY males (Klinefelter syndrome), XO females (Turner syndrome), and XYY males.
14.15 Humans may inherit chromosomes having structural changes, such as duplications, translocation, inversions, and deletions.

How can individuals learn more about their genetic makeup?

14.16 **Genetic testing** (p. 000) is the examination of a person's DNA to check for a disease or disorder, or to determine identity.
14.16 **Genetic counselors** (p. 000) provide support for individuals considering or undergoing genetic testing.
14.17 Cells for genetic testing of fetuses can be collected by amniocentesis and chorionic villus sampling.
14.17 Although most genetic disorders cannot yet be cured, research scientists are making progress in developing gene therapies and other technologies that may, in the future, have an important effect on a physician's ability to treat genetic disorders before birth.

Level 1 Learning Basic Facts and Terms

Multiple Choice

1. With incomplete dominance
   a. traits show either dominance or recessiveness.
   b. heterozygotes are phenotypic intermediates.
   c. phenotypic traits have a continuous nature.
   d. genetic counseling is useful.
2. In codominance
   a. a gene is represented by more than two alleles.
   b. a "double dose" of the gene is needed for a trait to be expressed.
   c. each allele contributes equally in the heterozygote.
   d. the phenotype is not completely due to the genotype.
3. Polygenic inheritance of a trait means that
   a. the trait is controlled by more than one gene.
   b. many genes are controlled by more than one trait.
   c. many alleles code for a single trait.
   d. genes for a trait are inherited from more than one parent.

True–False

4. ____ Most human genetic disorders arise from the inheritance of dominant alleles.
5. ____ In recessive disorders, mutant alleles are able to persist in the population among carriers.
6. ____ Tay-Sachs disease has a high incidence among Native American populations.

Matching

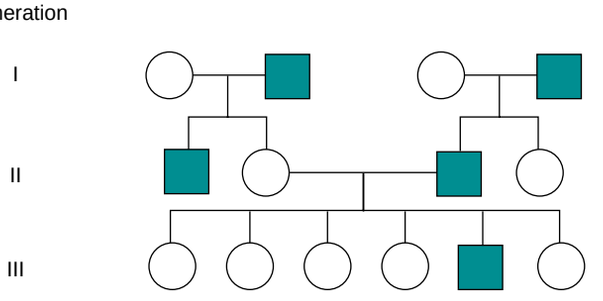
7. ____ autosome	a.	complete complement of chromosomes, paired and ordered
8. ____ pedigree	b.	procedure for prenatal diagnosis of a genetic disorder
9. ____ karyotype	c.	chromosome that does not determine gender
10. ____ trisomy	d.	diagram of genetic relationships among family members
	e.	inheritance of an extra copy of a chromosome

Level 2 Learning Concepts

1. What happens when human offspring inherit abnormal numbers of sex chromosomes? Summarize the four examples discussed in the chapter, and give the genotype of each.
2. What would you conclude about the inheritance pattern of each human trait in each of the following situations?
   a. The trait is expressed more frequently in males than in females.
   b. Offspring who exhibit this trait have at least one parent who exhibits the same trait.
   c. Offspring can exhibit this trait even though the parents do not.
   d. The trait is expressed equally in both males and females.
3. Why do most genetic disorders in humans result from recessive genes? Name several examples.
4. Distinguish between incomplete dominance and codominance. Describe the phenotype of a heterozygote in each case.
5. If a couple wants to have a child but suspects that they may be at risk for a genetic disorder, what can they do? If a pregnancy turns out to be high risk, what options are available?

Level 3 Critical Thinking—Life Applications and Human Genetics Problems

1. Familial hypercholesterolemia is an inherited genetic condition that leads to the development of cardiovascular disease at a relatively young age. Affected men typically have heart attacks in their 40s to 50s, and affected women in their 50s and 60s. Your father has this condition and his mother had it as well, but his father did not. Your mother does not have the condition, and your older brother appears unaffected. What is the inheritance pattern of this condition? What are the chances you inherited this disorder?
2. Suppose that you and your partner were interested in having children. What information would you be able to give a genetic counselor to help him or her identify or exclude you or your offspring from genetically based diseases or birth defects?
3. You were thrilled to have a new baby sister, but excitement turned to anxiety when she began to have digestive problems with persistent vomiting. Doctors quickly learned that she had an autosomal recessive disorder called *galactosemia*. Your sister lacked the enzyme that breaks down the milk sugar galatose, so her pediatrician put her on a special diet free of lactose and galactose. In a short time, your sister was fine.
   a. Neither of your parents is affected with galactosemia. If your sister's genotype is *gl/gl*, what are your parents' genotypes?
   b. You are not affected with galactosemia. What is your genotype or possible genotypes?
   c. Will your sister automatically pass on this disease to her children? Why or why not?
4. The extra chromosome 21 that is found in persons with Down syndrome is the cause of multiple developmental defects. What might this tell you about the interaction of genes on a particular chromosome?
5. A woman whose blood type is AB marries a man with the same blood type. Draw a Punnett square to illustrate the possible genotypes of their children. What blood type will each genotype have?
6. George has the same type of hemophilia as did Queen Victoria and some of her descendants. He marries his mother's sister's daughter Patricia. His maternal grandfather also had hemophilia. George and Patricia have five children: Two daughters are normal, and two sons and one daughter develop hemophilia. Draw the pedigree.
7. A couple with a newborn baby are troubled that the child does not appear to resemble either of them. Suspecting that a mix-up occurred at the hospital, they check the blood type of the infant. It is type O. Because the father is type A and the mother is type B, they conclude that a mistake must have been made. Are they correct?
8. How many chromosomes would you expect to find in the karyotype of a person with Turner syndrome?
9. A woman is married for the second time. Her first husband was blood type A, and her child by that marriage was type O. Her new husband is type B, and their child is type AB. What is the woman's ABO genotype and blood type?
10. Total color blindness is a rare hereditary disorder among humans in which no color is seen, only shades of gray. It occurs in individuals homozygous for a recessive allele and is not sex linked. A non-color-blind man whose father is totally color blind intends to marry a non-color-blind woman whose mother was totally color blind. What are the chances that they will produce offspring who are totally color blind?
11. This pedigree is of a rare trait in which children have extra fingers and toes.



Which one of the following patterns of inheritance is consistent with this pedigree?
   a. autosomal recessive
   b. autosomal dominant
   c. X-linked recessive
   d. X-linked dominant
   e. Y-linked

In The News Critical Thinking

STOP THAT GENE!

Now that you understand more about the science behind the report, reread this chapter's opening story about research into treatments for devastating genetic diseases such as Huntington's. To understand this research better, it may help you to follow these steps.

1. Review your immediate reaction to this research that you wrote when you began reading this chapter.
2. Based on your current understanding, again summarize the main point of the research in a sentence or two.
3. What questions do you now have about this research that are not answered by the opening story?
4. Visit the In The News section of this text's companion web site at www.wiley.com/college/alters and watch the "Stop That Gene!" video. Then use the "summary" link to read the accompanying story and access related links. Use this information, the links provided, and other online and library resources to answer your questions and find updates about this research topic. State the sources of your information. Explain why you believe the information to be accurate. Also determine if the information expresses a particular point of view or is biased in any way.
5. What in your view is the most significant aspect of this research? Why?
6. State how this research could be applied to the daily lives of individuals.