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14.1 Human Chromosomes

Lesson Objectives

- Identify the types of human chromosomes in a karotype.
- Describe the patterns of the inheritance of human traits.
- Explain how pedigrees are used to study human traits.

Lesson Summary

Karyotypes A **genome** is the full set of all the genetic information that an organism carries in its DNA. Chromosomes are bundles of DNA and protein found in the nucleus of a eukaryotic cell. A **karyotype** is a picture that shows the complete diploid set of human chromosomes, grouped in pairs and arranged in order of decreasing size. A typical human diploid cell contains 46 chromosomes, or 23 pairs:

- Two of the 46 are the **sex chromosomes** that determine an individual's sex: XX = female and XY = male. The X chromosome carries nearly 10 times the number of genes as the Y chromosome.
- The other 44 are **autosomes**, or autosomal chromosomes.

Transmission of Human Traits Human genes follow the same Mendelian patterns of inheritance as the genes of other organisms:

- Many human traits follow a pattern of simple dominance.
- The alleles for many human genes display codominant inheritance.
- Many human genes, including the genes for blood group, have multiple alleles.
- A gene located on a sex chromosome is a **sex-linked gene**. The genes on sex chromosomes show a sex-linked pattern of inheritance, since females have two copies of many genes (located on X chromosomes) while males have just one.
- In females, most of the genes in one of the X chromosomes are inactivated in each cell.

Human Pedigrees A chart used to analyze the pattern of inheritance that shows the relationships in a family is a **pedigree**. Pedigrees can be used to determine the nature of genes and alleles associated with inherited human traits.

Karyotypes

1. **THINK VISUALLY** Make a sketch of a human karyotype. Number the chromosome pairs. Label autosomes and sex chromosomes.

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For Questions 2–8, write the letter of the correct answer on the line at the left.

_____ **2.** The complete set of genetic information an organism carries in its DNA is its

A. karyotype.

B. genome.

C. chromosomes.

D. autosomes.

_____ **3.** From what is a karyotype made?

A. A photograph of cells in mitosis

B. A series of X-diffraction images

C. A preparation of gametes on a microscope slide

D. A Punnett square

_____ **4.** How many chromosomes are in a normal human karyotype?

A. 23

B. 46

C. 44

D. 2 (either XX or XY)

_____ **5.** Which of the following genetic abbreviations denotes a male human?

A. 23, XX

B. 23, XY

C. 46, XX

D. 46, XY

_____ **6.** Why is the ratio of male to female births roughly 50:50?

A. All egg cells carry an X chromosome.

B. Half of all egg cells carry a Y chromosome.

C. All sperm cells carry an X chromosome.

D. Half of all sperm cells carry a Y chromosome.

_____ **7.** How are the X and Y chromosomes different?

A. Only one is an autosome.

B. The X is smaller than the Y.

C. The Y carries fewer genes than the X.

D. Only females have a Y.

_____ **8.** All human cells carry

A. at least one X chromosome.

B. at least one Y chromosome.

C. a pair of X chromosomes.

D. one X and one Y chromosome.

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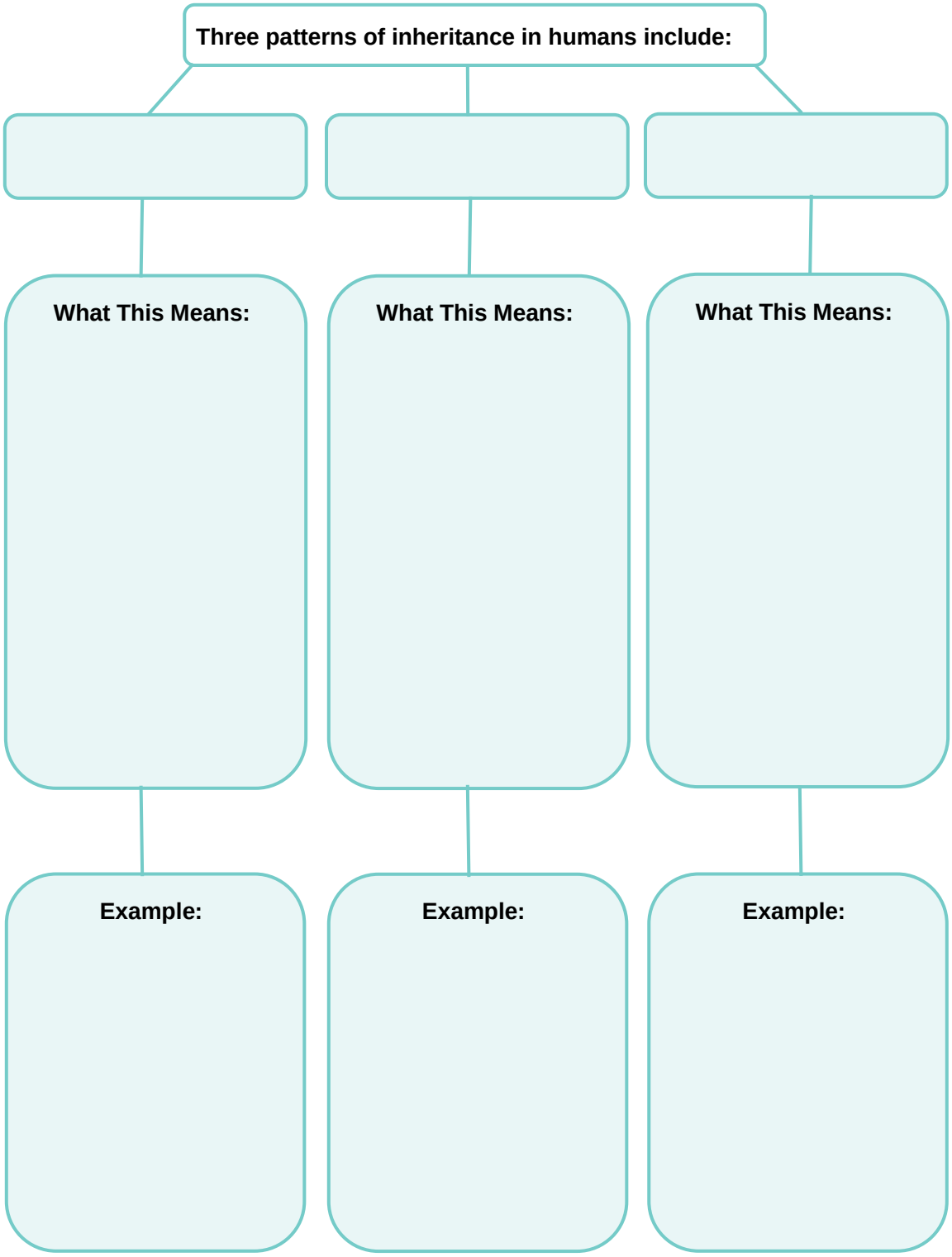
Transmission of Human Traits

9. Complete the graphic organizer to list, describe, and give examples of three types of inheritance patterns in humans:



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10. Colorblindness is a sex-linked trait. Let *C* represent an allele for normal color vision. Let *c* represent an allele for colorblindness. The genotype for a male with normal color vision is X^CY. The genotype for a female heterozygous for normal color vision is X^CX^c.

Complete the Punnett square to show the genotypes and phenotypes of their possible offspring.

	Male Gamete:	**Male Gamete:**
Female Gamete:	Genotype: Phenotype:	Genotype: Phenotype:
Female Gamete:	Genotype: Phenotype:	Genotype: Phenotype:

11. Use your Punnett square to explain why a female with one *c* allele has normal color vision but a male with one *c* allele is colorblind.

_____

_____

12. How does the cell "adjust" to the extra X chromosome in female cells?

_____

_____

13. What is a Barr body?

_____

_____

14. Why don't males have Barr bodies?

_____

_____

15. Is a cat with three colors of spots more likely to be male or female?

_____

_____

_____

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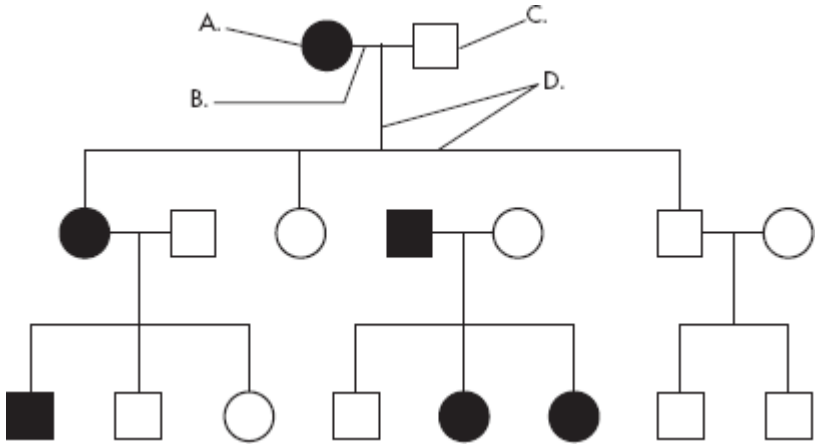
Human Pedigrees

For Questions 16–21, match the labels to the parts of the pedigree chart shown below. Some of the labels may be used more than once.



_____ **16.** A person who expresses the trait

_____ **17.** A male

_____ **18.** A person who does not express the trait

_____ **19.** A marriage

_____ **20.** A female

_____ **21.** A connection between parents and offspring

Apply the Big idea

22. Dimples in the cheeks are inherited as a dominant trait on an autosome. Using the proper form and symbols, draw a pedigree chart, beginning with a heterozygous, dimpled father (*Dd*), and a nondimpled mother (*dd*). Show four children of the expected types: boys, girls, dimples, and no dimples. Label your pedigree with phenotypes and genotypes.

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14.2 Human Genetic Disorders

Lesson Objectives

- Explain how small changes in DNA cause genetic disorders.
- Summarize the problems caused by nondisjunction.

Lesson Summary

From Molecule to Phenotype There is a molecular reason for genetic disorders. A change in DNA can alter an amino acid sequence, which can change a protein and therefore, the phenotype. Some common inherited disorders result from a change in DNA. They include:

- sickle cell disease, in which a defective polypeptide makes hemoglobin in the blood less soluble;
- cystic fibrosis, in which a deletion of three bases in a gene causes cell membranes to lose their ability to transport chloride ions;
- Huntington's disease, in which a single codon for a certain amino acid repeats more than 40 times, causing mental deterioration and uncontrolled movements.

Some alleles that cause disease in the homozygote can provide an advantage in the heterozygote. The geographic associations between sickle cell disease and malaria and between cystic fibrosis and typhoid demonstrate how the heterozygous state reduces the risk of infection.

Chromosomal Disorders Sometimes, during meiosis, homologous chromosomes fail to separate. This **nondisjunction** (not coming apart) can create a gamete with an abnormal number of chromosomes, leading to offspring with missing or extra chromosomes. Examples include:

- Down syndrome, most often a result of three copies of chromosome 21;
- Turner's syndrome, a female with a single X chromosome;
- Klinefelter's syndrome, a male with an extra X chromosome.

From Molecule to Phenotype

1. The boxes below each show a step to explain how genetic disorders have a molecular basis. Number them so that the steps are in the correct order.

A change in phenotype results. _____	A gene's DNA sequence changes. _____	The amino acid sequence that alters a protein changes. _____

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For Questions 2–7, write the letter of the correct answer on the line at the left.

_____ **2.** How many human genetic disorders are known?

A. three

B. about 20

C. about 100

D. thousands

_____ **3.** The inherited disease in which hemoglobin molecules clump into long fibers, changing the shape of blood cells is

A. cystic fibrosis.

B. sickle cell disease.

C. Huntington's disease.

D. Klinefelter's syndrome.

_____ **4.** What happens to the CFTR gene in individuals who have cystic fibrosis?

A. The entire gene is deleted.

B. The entire gene is duplicated.

C. Three bases are deleted, causing one amino acid to be missing.

D. Three bases are duplicated, causing one amino acid show up about 40 times.

_____ **5.** Why are individuals who are heterozygous for the cystic fibrosis allele unaffected by the disease?

A. They have an extra copy of the allele on their X chromosome.

B. Cystic fibrosis only occurs in males, so females are unaffected.

C. They make enough of a particular protein to allow their cells to work properly.

D. Their cells can transport chloride ions through diffusion channels.

_____ **6.** How might the allele that causes a disease stay in the population if it is fatal to those who have the disease?

A. It is present only in heterozygotes.

B. It makes the heterozygote resistant to a fatal disease.

C. It disappears but is continuously replaced by mutations.

D. It occurs only in certain geographic areas.

_____ **7.** What advantage do individuals with one sickle cell allele have?

A. a stronger resistance to malaria

B. immunity to typhoid fever

C. more rigid red blood cells

D. no advantage

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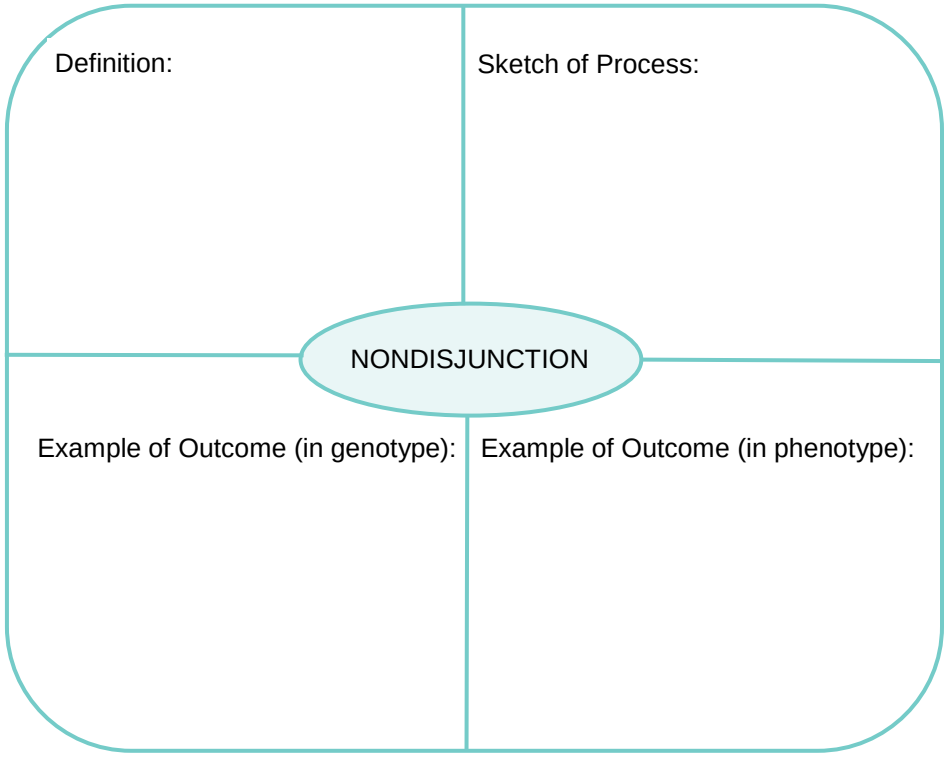
Chromosomal Disorders

8. Complete this graphic organizer to explain the process and outcomes of nondisjunction.



9. What is trisomy?

_____

_____

10. What happens when a male has XXY sex chromosomes?

_____

_____

Apply the Big idea

11. Most of the genetic disorders you have learned about are the result of a change in DNA sequence, as with cystic fibrosis, or the presence of an extra chromosome, as with Down syndrome. The exception is Turner's syndrome. Women with Turner's syndrome have only 45 chromosomes. They are missing an X chromosome. This disorder is the *only* case in which a person can survive with one less chromosome. What does this tell you about how genetic information is inherited in humans?

_____

_____

_____

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14.3 Studying the Human Genome

Lesson Objectives

- Summarize the methods of DNA analysis.
- State the goals of the Human Genome Project and explain what we have learned so far.

Lesson Summary

Manipulating DNA Since the 1970s, techniques have been developed that allow scientists to cut, separate, and replicate DNA base-by-base. Using these tools, scientists can read the base sequences in DNA from any cell.

- **Restriction enzymes** cut DNA into smaller pieces, called restriction fragments, which are several hundred bases in length. Each restriction enzyme cuts DNA at a different sequence of bases.
- **Gel electrophoresis** separates different-sized DNA fragments by placing them at one end of a porous gel, then applying an electrical voltage. The electrical charge moves the DNA.
- Using dye-labeled nucleotides, scientists can stop replication at any point along a single DNA strand. The fragments can then be separated by size using gel electrophoresis and "read," base-by-base.

The Human Genome Project was a 13-year international effort to sequence all 3 billion base pairs in human DNA and identify all human genes. The project was completed in 2003.

- The researchers identified markers in widely separated strands of DNA.
- They used "shotgun sequencing," which uses a computer to match DNA base sequences.
- To identify genes, they found promoters, exons, and other sites on the DNA molecule.
- To locate and identify as many haplotypes (collections of linked single-base differences) in the human population as possible, the International HapMap Project began in 2002.
- The Human Genome Project identified genes associated with many diseases and disorders. From the project came the new science of **bioinformatics**, the creation and use of databases and other computing tools to manage data. Bioinformatics launched **genomics**, the study of whole genomes.
- The human genome project pinpointed genes and associated particular sequences in those genes with numerous diseases and disorders. It also found that the DNA of all humans matches base-for-base at most sites, but can vary at 3 million sites.
- The 1000 Genomes Project, launched in 2008, will catalogue the variation among 1000 people.

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Manipulating DNA

For Questions 1–4, write True if the statement is true. If the statement is false, change the underlined word to make the statement true.

_____ **1.** Bacteria produce restriction enzymes that cut the DNA molecule into smaller pieces.

_____ **2.** Restriction fragments are always cut at a particular sequence of proteins.

_____ **3.** The technique that separates differently sized DNA fragments is gel electrophoresis.

_____ **4.** The enzyme that copies DNA is DNA restrictase.

5. Complete the graphic organizer to summarize the steps used to determine the sequences of bases in DNA.

Purpose	Tool or Technique Used	Outcome
Cutting DNA		
Separating DNA		
Reading DNA		

For Questions 6–10, complete each statement by writing in the correct word or words.

6. By using tools that cut, separate, and then replicate DNA, scientists can now read the _____sequence in DNA from any cell.

7. Restriction enzymes cut pieces of DNA sometimes called restriction _____.

8. Each restriction enzyme cuts DNA at a different sequence of _____.

9. The smaller the DNA, the _____ and farther it moves during gel electrophoresis.

10. After chemically dyed bases have been incorporated into a DNA strand, the order of colored _____ on the gel reveals the exact sequence of bases in DNA.

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The Human Genome Project

For Questions 11–16, write the letter of the correct answer on the line at the left.

_____ **11.** What technology made the Human Genome Project possible?

A. DNA sequencing

B. RNA replication

C. protein synthesis

D. enzyme activation

_____ **12.** What were the "markers" that the researchers of the Human Genome Project used?

A. restriction enzymes

B. gel electrophoresis

C. base sequences

D. restriction fragments

_____ **13.** What does "shotgun sequencing" do?

A. separate fragments using gel electrophoresis

B. find overlapping areas of DNA fragments

C. cut DNA into millions of "puzzle pieces"

D. bind colored dyes to base sequences

_____ **14.** What are SNPs?

A. points where a restriction enzyme cuts a DNA molecule

B. missing sequence of base pairs in a restriction fragment

C. proteins formed by a mutated gene

D. differences in a base between two individuals

_____ **15.** Bioinformatics would not have been possible without

A. microscopes.

B. genes.

C. computers.

D. genomics.

_____ **16.** In humans, single-base differences

A. occur at about 3 million sites.

B. occur rarely in the sex chromosomes.

C. seldom occur in normal DNA.

D. cannot be identified from DNA analysis.

17. What were the goals of the Human Genome Project?

_____

_____

_____

_____

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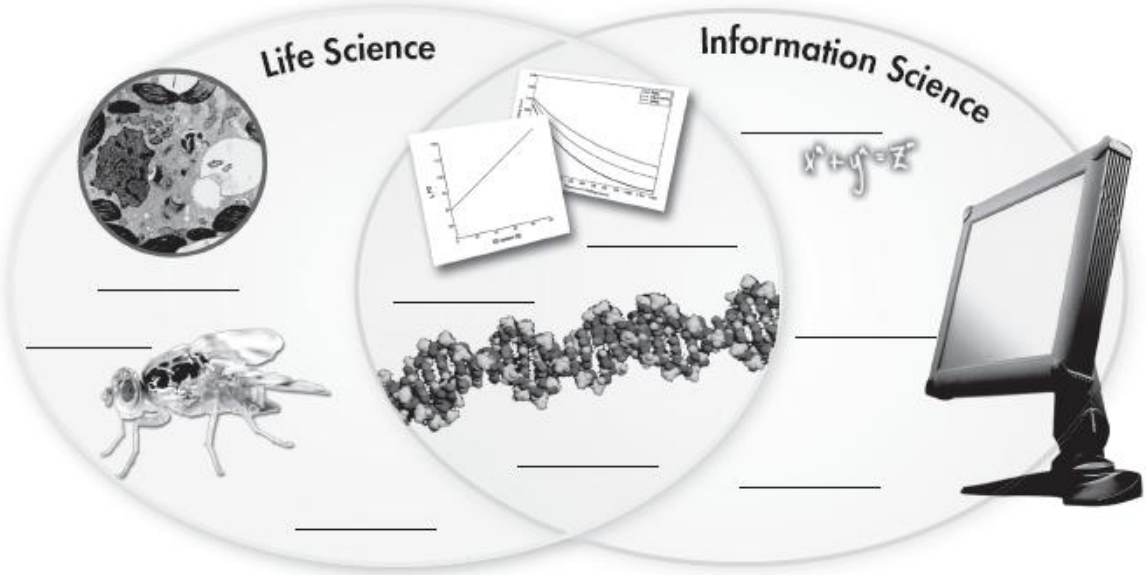
18. **THINK VISUALLY** The field of bioinformatics combines both life sciences and modern technology. Fill in the Venn diagram to show how.



Apply the Big idea

19. The Icelandic people have always placed high importance on knowing about their ancestors. In fact, 80% of all the Icelandic people who have ever lived can be added to a family tree. Medical records are just as detailed. The population is quite isolated, so the gene pool is considered to be homogeneous. Why would these conditions make the genome of the Icelandic population ideal for studying rare inherited disorders associated with gene sequencing errors?

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Chapter Vocabulary Review

For Questions 1–11, match the term with its definition.

Definition

_____ **1.** The X chromosome or the Y chromosome

_____ **2.** A gene on the X chromosome or the Y chromosome

_____ **3.** The failure of homologous chromosomes to separate during meiosis

_____ **4.** A technology used to separate fragments of DNA

_____ **5.** A chart that shows family relationships and inheritance of traits

_____ **6.** A field of study that includes the operation of databases

_____ **7.** An enzyme that cuts a DNA molecule into small pieces

_____ **8.** The study of whole genomes, including genes and their functions

_____ **9.** A picture that shows chromosomes arranged in pairs

_____ **10.** Any chromosome that is not a sex chromosome

_____ **11.** The full set of genetic information in an organism's DNA

Term

A. genome

B. karyotype

C. sex chromosome

D. autosome

E. sex-linked gene

F. pedigree

G. nondisjunction

H. restriction enzyme

I. gel electrophoresis

J. bioinformatics

K. genomics

For Questions 12–19, complete each statement by writing in the correct word or words.

12. A circle represents a female in a(n) _____.

13. The protein that cuts DNA into pieces is a restriction _____.

14. An inherited disorder that appears more often in males than females is probably caused by a _____.

15. The 23 pairs of human chromosomes are arranged from largest to smallest in a _____.

16. Humans have 22 pairs of _____.

17. The cause of Down syndrome is _____ during meiosis.

18. Humans have 3 billion base pairs in their _____.

19. The new field of _____ resulted from the Human Genome Project.