

## 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.

# Chapter 14 Human Genome Answer Key

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## **Table of Contents Chapter 14 Human Genome Answer Key**

1. Understanding the eBook Chapter 14 Human Genome Answer Key
  - The Rise of Digital Reading Chapter 14 Human Genome Answer Key
  - Advantages of eBooks Over Traditional Books
2. Identifying Chapter 14 Human Genome Answer Key
  - Exploring Different Genres
  - Considering Fiction vs. Non-Fiction
  - Determining Your Reading Goals
3. Choosing the Right eBook Platform
  - Popular eBook Platforms
  - Features to Look for in an Chapter 14 Human Genome Answer Key
  - User-Friendly Interface
4. Exploring eBook Recommendations from Chapter 14 Human Genome Answer Key
  - Personalized Recommendations
  - Chapter 14 Human Genome Answer Key User Reviews and Ratings
  - Chapter 14 Human Genome Answer Key and Bestseller Lists
5. Accessing Chapter 14 Human Genome Answer Key Free and Paid eBooks
  - Chapter 14 Human Genome Answer Key Public Domain eBooks
  - Chapter 14 Human Genome Answer Key eBook Subscription Services
  - Chapter 14 Human Genome Answer Key Budget-Friendly Options
6. Navigating Chapter 14 Human Genome Answer Key eBook Formats
  - ePub, PDF, MOBI, and More
  - Chapter 14 Human Genome Answer Key Compatibility with Devices
  - Chapter 14 Human Genome Answer Key Enhanced eBook Features
7. Enhancing Your Reading Experience
  - Adjustable Fonts and Text Sizes of Chapter 14 Human Genome Answer Key
  - Highlighting and Note-Taking Chapter 14 Human Genome Answer Key
  - Interactive Elements Chapter 14 Human Genome Answer Key

8. Staying Engaged with Chapter 14 Human Genome Answer Key
  - Joining Online Reading Communities
  - Participating in Virtual Book Clubs
  - Following Authors and Publishers Chapter 14 Human Genome Answer Key
9. Balancing eBooks and Physical Books Chapter 14 Human Genome Answer Key
  - Benefits of a Digital Library
  - Creating a Diverse Reading Collection Chapter 14 Human Genome Answer Key
10. Overcoming Reading Challenges
  - Dealing with Digital Eye Strain
  - Minimizing Distractions
  - Managing Screen Time
11. Cultivating a Reading Routine Chapter 14 Human Genome Answer Key
  - Setting Reading Goals Chapter 14 Human Genome Answer Key
  - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Chapter 14 Human Genome Answer Key
  - Fact-Checking eBook Content of Chapter 14 Human Genome Answer Key
  - Distinguishing Credible Sources
13. Promoting Lifelong Learning
  - Utilizing eBooks for Skill Development
  - Exploring Educational eBooks
14. Embracing eBook Trends
  - Integration of Multimedia Elements
  - Interactive and Gamified eBooks

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