

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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Table of Contents Chapter 14 The Human Genome Answer Key Wordwise

1. Understanding the eBook Chapter 14 The Human Genome Answer Key Wordwise
 - The Rise of Digital Reading Chapter 14 The Human Genome Answer Key Wordwise
 - Advantages of eBooks Over Traditional Books
2. Identifying Chapter 14 The Human Genome Answer Key Wordwise
 - 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 The Human Genome Answer Key Wordwise
 - User-Friendly Interface
4. Exploring eBook Recommendations from Chapter 14 The Human Genome Answer Key Wordwise
 - Personalized Recommendations
 - Chapter 14 The Human Genome Answer Key Wordwise User Reviews and Ratings

- Chapter 14 The Human Genome Answer Key Wordwise and Bestseller Lists
- 5. Accessing Chapter 14 The Human Genome Answer Key Wordwise Free and Paid eBooks
 - Chapter 14 The Human Genome Answer Key Wordwise Public Domain eBooks
 - Chapter 14 The Human Genome Answer Key Wordwise eBook Subscription Services
 - Chapter 14 The Human Genome Answer Key Wordwise Budget-Friendly Options
- 6. Navigating Chapter 14 The Human Genome Answer Key Wordwise eBook Formats
 - ePub, PDF, MOBI, and More
 - Chapter 14 The Human Genome Answer Key Wordwise Compatibility with Devices
 - Chapter 14 The Human Genome Answer Key Wordwise Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Chapter 14 The Human Genome Answer Key Wordwise
 - Highlighting and Note-Taking Chapter 14 The Human Genome Answer Key Wordwise
 - Interactive Elements Chapter 14 The Human Genome Answer Key Wordwise
- 8. Staying Engaged with Chapter 14 The Human Genome Answer Key Wordwise
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Chapter 14 The Human Genome Answer Key Wordwise
- 9. Balancing eBooks and Physical Books Chapter 14 The Human Genome Answer Key Wordwise
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Chapter 14 The Human Genome Answer Key Wordwise
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Chapter 14 The Human Genome Answer Key Wordwise
 - Setting Reading Goals Chapter 14 The Human Genome Answer Key Wordwise
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Chapter 14 The Human Genome Answer Key Wordwise
 - Fact-Checking eBook Content of Chapter 14 The Human Genome Answer Key Wordwise
 - 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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