

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-labelled 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.

Answer Key For The Human Genome Project

**Deitra Leonard Lowdermilk, Kitty
Cashion, Kathryn Rhodes
Alden, Shannon E. Perry**

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Bioinformatics and the Human Genome Project ,2003 Contains student lessons and teacher support materials on the nature and methods of bioinformatics and the ethical and public policy dilemmas emerging from the use of genetic databases

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Table of Contents Answer Key For The Human Genome Project

1. Understanding the eBook Answer Key For The Human Genome Project
 - The Rise of Digital Reading Answer Key For The Human Genome Project
 - Advantages of eBooks Over Traditional Books
2. Identifying Answer Key For The Human Genome Project
 - 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 Answer Key For The Human Genome Project
 - User-Friendly Interface
4. Exploring eBook Recommendations from Answer Key For The Human Genome Project
 - Personalized Recommendations
 - Answer Key For The Human Genome Project User Reviews and Ratings
 - Answer Key For The Human Genome Project and Bestseller Lists
5. Accessing Answer Key For The Human Genome Project Free and Paid eBooks

- Answer Key For The Human Genome Project Public Domain eBooks
- Answer Key For The Human Genome Project eBook Subscription Services
- Answer Key For The Human Genome Project Budget-Friendly Options

6. Navigating Answer Key For The Human Genome Project eBook Formats

- ePUB, PDF, MOBI, and More
- Answer Key For The Human Genome Project Compatibility with Devices
- Answer Key For The Human Genome Project Enhanced eBook Features

7. Enhancing Your Reading Experience

- Adjustable Fonts and Text Sizes of Answer Key For The Human Genome Project
- Highlighting and Note-Taking Answer Key For The Human Genome Project
- Interactive Elements Answer Key For The Human Genome Project

8. Staying Engaged with Answer Key For The Human Genome Project

- Joining Online Reading Communities
- Participating in Virtual Book Clubs
- Following Authors and Publishers Answer Key For The Human Genome Project

9. Balancing eBooks and Physical Books Answer Key For The Human Genome Project

- Benefits of a Digital Library
- Creating a Diverse Reading Collection Answer Key For The Human Genome Project

10. Overcoming Reading Challenges

- Dealing with Digital Eye Strain
- Minimizing Distractions
- Managing Screen Time

11. Cultivating a Reading Routine Answer Key For The Human Genome Project

- Setting Reading Goals Answer Key For The Human Genome Project
- Carving Out Dedicated Reading Time

12. Sourcing Reliable Information of Answer Key For The Human Genome Project

- Fact-Checking eBook Content of Answer Key For The Human Genome Project
- 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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