

14.2 Human Genetic Disorders

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- _____
- ▶ cystic fibrosis- _____
- ▶ Huntington's disease- _____

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 _____ (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- _____
- ▶ Turner's syndrome- _____
- ▶ Klinefelter's syndrome- _____

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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Table of Contents Biology Study Guide Genetic Disorders Answers

1. Understanding the eBook Biology Study Guide Genetic Disorders Answers
 - The Rise of Digital Reading Biology Study Guide Genetic Disorders Answers
 - Advantages of eBooks Over Traditional Books
2. Identifying Biology Study Guide Genetic Disorders Answers
 - 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 Biology Study Guide Genetic Disorders Answers
 - User-Friendly Interface
4. Exploring eBook Recommendations from Biology Study Guide Genetic Disorders Answers
 - Personalized Recommendations
 - Biology Study Guide Genetic Disorders Answers User Reviews and Ratings
 - Biology Study Guide Genetic Disorders Answers and Bestseller Lists
5. Accessing Biology Study Guide Genetic Disorders Answers Free and Paid eBooks
 - Biology Study Guide Genetic Disorders Answers Public Domain eBooks
 - Biology Study Guide Genetic Disorders Answers eBook Subscription Services
 - Biology Study Guide Genetic Disorders Answers Budget-Friendly Options
6. Navigating Biology Study Guide Genetic Disorders Answers eBook Formats
 - ePub, PDF, MOBI, and More
 - Biology Study Guide Genetic Disorders Answers Compatibility with Devices
 - Biology Study Guide Genetic Disorders Answers Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Biology Study Guide Genetic Disorders Answers
 - Highlighting and Note-Taking Biology Study Guide Genetic Disorders Answers
 - Interactive Elements Biology Study Guide Genetic Disorders Answers

8. Staying Engaged with Biology Study Guide Genetic Disorders Answers
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Biology Study Guide Genetic Disorders Answers
9. Balancing eBooks and Physical Books Biology Study Guide Genetic Disorders Answers
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Biology Study Guide Genetic Disorders Answers
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Biology Study Guide Genetic Disorders Answers
 - Setting Reading Goals Biology Study Guide Genetic Disorders Answers
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Biology Study Guide Genetic Disorders Answers
 - Fact-Checking eBook Content of Biology Study Guide Genetic Disorders Answers
 - 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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