

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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Table of Contents Genetic Disorders Answer Key Section 7 3

1. Understanding the eBook Genetic Disorders Answer Key Section 7 3
 - The Rise of Digital Reading Genetic Disorders Answer Key Section 7 3
 - Advantages of eBooks Over Traditional Books
2. Identifying Genetic Disorders Answer Key Section 7 3
 - 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 Genetic Disorders Answer Key Section 7 3
 - User-Friendly Interface
4. Exploring eBook Recommendations from Genetic Disorders Answer Key Section 7 3
 - Personalized Recommendations
 - Genetic Disorders Answer Key Section 7 3 User Reviews and Ratings
 - Genetic Disorders Answer Key Section 7 3 and Bestseller Lists
5. Accessing Genetic Disorders Answer Key Section 7 3 Free and Paid eBooks

- Genetic Disorders Answer Key Section 7 3 Public Domain eBooks
 - Genetic Disorders Answer Key Section 7 3 eBook Subscription Services
 - Genetic Disorders Answer Key Section 7 3 Budget-Friendly Options
6. Navigating Genetic Disorders Answer Key Section 7 3 eBook Formats
- ePub, PDF, MOBI, and More
 - Genetic Disorders Answer Key Section 7 3 Compatibility with Devices
 - Genetic Disorders Answer Key Section 7 3 Enhanced eBook Features
7. Enhancing Your Reading Experience
- Adjustable Fonts and Text Sizes of Genetic Disorders Answer Key Section 7 3
 - Highlighting and Note-Taking Genetic Disorders Answer Key Section 7 3
 - Interactive Elements Genetic Disorders Answer Key Section 7 3
8. Staying Engaged with Genetic Disorders Answer Key Section 7 3
- Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Genetic Disorders Answer Key Section 7 3
9. Balancing eBooks and Physical Books Genetic Disorders Answer Key Section 7 3
- Benefits of a Digital Library
 - Creating a Diverse Reading Collection Genetic Disorders Answer Key Section 7 3
10. Overcoming Reading Challenges
- Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Genetic Disorders Answer Key Section 7 3
- Setting Reading Goals Genetic Disorders Answer Key Section 7 3
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Genetic Disorders Answer Key Section 7 3
- Fact-Checking eBook Content of Genetic Disorders Answer Key Section 7 3
 - 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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