

Chromosomes, Genes, Alleles and Mutations (4.1)

Define the following terms (4.1.2 / 4.1.3 / 4.2.2 / 4.3.1 / 4.4.11)

Gene: A heritable factor that controls a specific characteristic

Allele: One specific form of a gene, occupying the same gene locus as other alleles of the gene

Genome: The whole of the genetic information of an organism

Gene Mutation: A change in the nucleotide sequence of a section of DNA coding for a polypeptide

Homologous Chromosomes: Chromosomes that have the same structural features and the same genes

Genotype: The alleles of an organism

Phenotype: The characteristics of an organism (combination of genotype and environmental factors)

Dominant Allele: An allele that is expressed in the phenotype in a homozygous or heterozygous state

Recessive Allele: An allele that is only expressed in the phenotype in a homozygous state

Codominant Alleles: A pair of alleles which both affect the phenotype in a heterozygote

Locus: The particular position on homologous chromosomes of a gene

Homozygous: Having two identical alleles of a gene

Heterozygous: Having two different alleles of a gene

Carrier: A heterozygote who has one copy of a recessive allele that causes a genetic disease

Test Cross: Testing a suspected heterozygote by crossing with a known homozygous recessive

Clone: A group of genetically identical organisms or a group of cells derived from a single parent cell

Organisation of chromosomes (4.1.1)

- Eukaryotic chromosomes consist of DNA wrapped around histone proteins
- This forms the basic structure of the nucleosome, which are packaged into chromatin
- Chromatin containing active genes remains in an expanded form (euchromatin)
- Chromatin with genes not expressed by the cell becomes condensed (heterochromatin)
- Chromatin only supercoils into chromosomes during cell division (mitosis / meiosis)



Consequences of a base substitution mutation in sickle cell anaemia (4.1.4)

Cause:

- Base substitution: GAG → GTG
- Amino acid change: Glu → Val
- Location of change: Haemoglobin beta chain (chromosome 11)
- Mode of inheritance: Autosomal recessive

Consequence:

- Structural effect: Haemoglobin forms strands; red blood cells to become sickle-shaped
- Functional effect: Causes anaemia (low RBC count) as sickle-cells destroyed faster
Blood is less effective at transporting oxygen, causing fatigue
- Complications: Altered shape causes clotting, leading to organ failure (e.g. kidneys)

Genes And Chromosomes Reinforcement And Study Guide

Rachel Sandford



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Table of Contents Genes And Chromosomes Reinforcement And Study Guide

1. Understanding the eBook Genes And Chromosomes Reinforcement And Study Guide
 - The Rise of Digital Reading Genes And Chromosomes Reinforcement And Study Guide
 - Advantages of eBooks Over Traditional Books
2. Identifying Genes And Chromosomes Reinforcement And Study Guide
 - 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 Genes And Chromosomes Reinforcement And Study Guide
 - User-Friendly Interface
4. Exploring eBook Recommendations from Genes And Chromosomes Reinforcement And Study Guide
 - Personalized Recommendations
 - Genes And Chromosomes Reinforcement And Study Guide User Reviews and Ratings
 - Genes And Chromosomes Reinforcement And Study Guide and Bestseller Lists

5. Accessing Genes And Chromosomes Reinforcement And Study Guide Free and Paid eBooks
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 - Genes And Chromosomes Reinforcement And Study Guide eBook Subscription Services
 - Genes And Chromosomes Reinforcement And Study Guide Budget-Friendly Options
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 - Highlighting and Note-Taking Genes And Chromosomes Reinforcement And Study Guide
 - Interactive Elements Genes And Chromosomes Reinforcement And Study Guide
8. Staying Engaged with Genes And Chromosomes Reinforcement And Study Guide
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Genes And Chromosomes Reinforcement And Study Guide
9. Balancing eBooks and Physical Books Genes And Chromosomes Reinforcement And Study Guide
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Genes And Chromosomes Reinforcement And Study Guide
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Genes And Chromosomes Reinforcement And Study Guide
 - Setting Reading Goals Genes And Chromosomes Reinforcement And Study Guide
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Genes And Chromosomes Reinforcement And Study Guide
 - Fact-Checking eBook Content of Genes And Chromosomes Reinforcement And Study Guide
 - 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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