

# 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

**Yi-Tong Ma**



## **Genes And Chromosomes Reinforcement And Study Guide:**

## Whispering the Secrets of Language: An Emotional Journey through **Genes And Chromosomes Reinforcement And Study Guide**

In a digitally-driven earth where displays reign supreme and quick interaction drowns out the subtleties of language, the profound secrets and psychological nuances concealed within words usually go unheard. Yet, located within the pages of **Genes And Chromosomes Reinforcement And Study Guide** a charming literary prize pulsating with fresh emotions, lies a fantastic journey waiting to be undertaken. Published by an experienced wordsmith, that charming opus attracts visitors on an introspective trip, gently unraveling the veiled truths and profound influence resonating within the very cloth of each word. Within the psychological depths of the emotional review, we can embark upon a honest exploration of the book is key subjects, dissect its charming publishing model, and yield to the effective resonance it evokes strong within the recesses of readers hearts.

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