# 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)



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

#### Causer

- 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 Study Guide**

**Mark Steyvers** 

**Genes And Chromosomes Study Guide:** 

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In a digitally-driven world where screens reign supreme and instant interaction drowns out the subtleties of language, the profound secrets and mental nuances hidden within phrases often move unheard. However, located within the pages of **Genes And Chromosomes Study Guide** a fascinating literary value blinking with raw thoughts, lies a fantastic journey waiting to be undertaken. Penned by a talented wordsmith, that marvelous opus encourages visitors on an introspective trip, delicately unraveling the veiled truths and profound influence resonating within ab muscles cloth of each word. Within the mental depths of the touching review, we can embark upon a honest exploration of the book is core themes, dissect its interesting publishing type, and yield to the strong resonance it evokes deep within the recesses of readers hearts.

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