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)

DNA → **Nucelosome** → **Chromatin**

Consquences 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)