Chromosomes, Genes, Alleles and Mutations

Karyotyping

Eukaryotic chromosomes are made of DNA and protein.

Karyotyping

Pair of chromosomes showing matching banding pattern.

Karyotyping is arranging the chromosomes in pairs according to their structure.

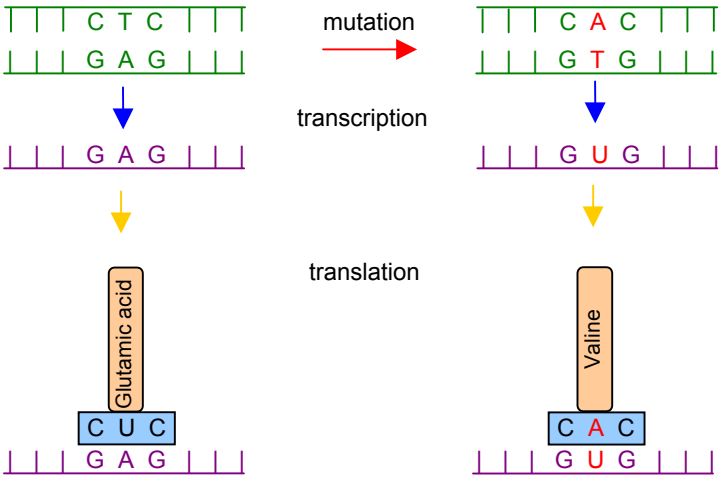
Uses

- Observing major changes in chromosome structure due to mutation;
- Observing changes in chromosome number as in Downs syndrome.

Sickle-cell Anaemia

Base Substitution Mutation - Sickle Cell Anaemia

- Caused by a mutation in one of the haemoglobin genes.
- An amino acid called glutamic acid has been replaced by one called valine.



Refer to the codon table on page 109 and look up the codons for these two amino acids.

If you substitute the middle A of the glutamic acid triplet with a U you will get a codon for valine.

Sickle-cell and Malaria

Sickle Cell and Malaria

Key points

- The sickle cell allele (Hb^s) is codominant (see p51);
- Two mutated alleles produce sickle cell anaemia;
- One mutated allele and one normal allele (the heterozygous condition – see p102) produces sickle cell trait;
- Sickle cell trait results in resistance to the malaria parasite that invades red blood cells;
- Populations that live in areas where malaria is common – Africa, Middle East, India – have a higher frequency of the sickle cell allele.

Define gene.

Define allele.

Define genome.