Any change to the quantity or the base sequence of the DNA of an organism is known as a mutation. Mutations occurring through the formation of gametes may be inherited. Any change to one or more nucleotide bases, or a change in the sequence of the bases, in DNA is known as a gene mutation. Gene mutations can arise spontaneously during DNA replication and include base substitution and base deletion.

Substitution of bases:

This is where a nucleotide in DNA is replaced by another nucleotide that has a different base.

Example: DNA triplet of bases guanine-thymine-cytosine (GTC) that codes for glutamine. If the final base (cytosine) is replaced by guanine, then code becomes GTG that codes for histidine and replaces the original amino acid glutamine.

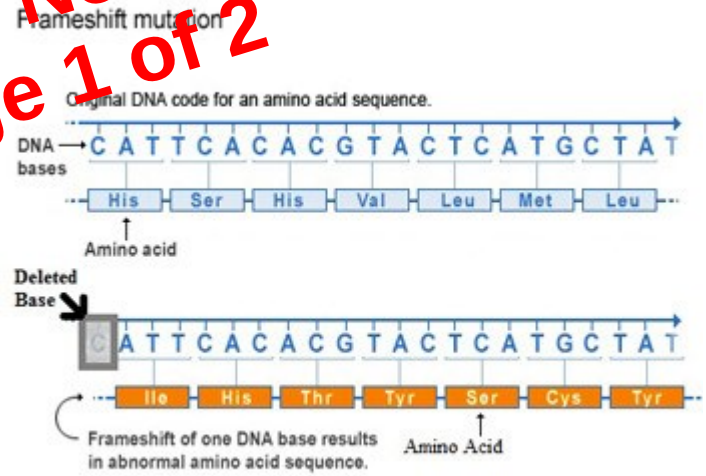
The polypeptide produced will differ in a single amino acid and the significance of this difference will depend upon the precise role of the original amino acid.

Replacement amino acid may not form the same bonds that determine the tertiary structure of a protein/ may be a different shape and therefore not function properly (like an enzyme with an inaccurate active site. The effect of mutation is different if the new triplet still codes for the same amino acid (due the degenerate nature of genetic code).

Gene Mutation

Deletion of bases:

A gene mutation by deletion arises when a nucleotide is lost from the normal DNA sequence. Usually the amino acid sequence of the polypeptide is entirely different and so the polypeptide is unlikely to function correctly. This is because the sequence of bases in DNA is read in units of three bases (triplet). One deleted nucleotide



Chromosome mutations:

This is changes in the structure or number of whole chromosomes. They take two forms:

1. Changes in whole sets of chromosomes: occur when organisms have three or more sets of chromosomes rather than the usual two. This condition is called polyploidy and occurs mostly in plants.
2. Changes in the number of individual chromosomes: sometimes individual homologous pairs of chromosomes fail to separate during meiosis. This is known as non-disjunction and usually results in a gamete having either one more or one fewer chromosome. On fertilisation with a gamete that has the normal amount of chromosomes, the resultant offspring have more or fewer chromosomes than normal in tall their body cells. An example of non-disjunction is Downs syndrome (an additional chromosome).