1.6. Mutations

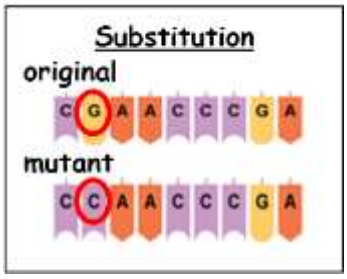
When a cell divides, the process of DNA replication is very carefully controlled to preserve the **genetic information** encoded within the **nucleotide** base sequence.

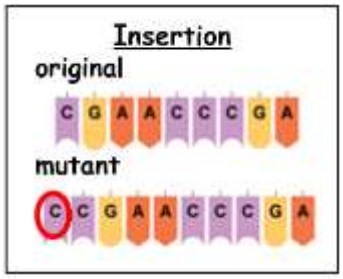
However, changes in the genome, known as **mutations**, do occur and can result in there being no **protein** or an altered protein being **expressed**.

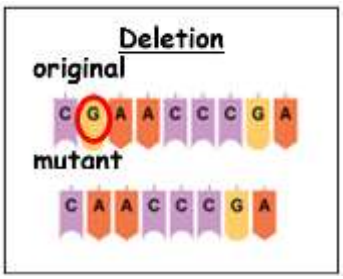
Mutations arise **spontaneously** and at **random** but only occur **rarely**.

Single gene mutations

A single gene (**point**) mutations involve the alteration of a DNA nucleotide sequence as a result of the **substitution**, **insertion** or **deletion** of nucleotides.







Substitution

Substitution is when one base is **substituted** for another, this usually brings about only a **minor** change to the protein, as it only affects one **codon** therefore one **amino acid**. However, if the substitution occurs at a critical point, it can result in **major** changes such as Sickle cell anaemia.

Types of single-nucleotide substitutions	
Missense	After a substitution, the altered codon codes for an amino acid which still makes sense but not the original sense.
Nonsense	As a result of a substitution, a codon that used to code for an amino acid becomes changed into one that acts as a stop codon. It causes protein synthesis to be halted prematurely and results in the formation of a polypeptide chain which is shorter than the normal one and often non-functional.
Splice-site	A molecule of primary mRNA transcript is spliced to remove introns and seal exons together. A splice-site mutation substitutes, inserts or deletes one or more nucleotides at a site where introns are normally removed, this results in introns being left in. Splice-site mutations can alter post-transcriptional processing.

Insertions and deletions

Insertions and deletions of a single base pair brings about **major** changes since they cause a large portion of a gene's DNA to be **misread**. The protein produced differs from the normal protein by many amino acids and it is usually **non-functional**. Frame-shift mutations can also result in an expansion of a **nucleotide sequence repeat**.

Chromosome Mutations

Chromosome mutations alter the **structure** of one or more **chromosomes**.

Types of chromosome mutations:	
Duplication **original**  **mutant** 	A segment of genes is **repeated**. Some duplication of genes may have a detrimental effect or be of an advantage.
Deletion **original**  **mutant** 	A segment of genes becomes **detached** and the two remaining ends **join** giving a shorter chromosome lacking the detached genes. Deletion normally has **drastic** effects on the living organism involved.
Inversion **original**  **mutant** 	A segment of genes is **reversed**. This results in non-viable gametes.
Translocation **original**  **mutant** 	A section of one chromosome **breaks** off and **attaches** to another chromosome that is not it's matching partner. This results in non-viable gametes.

Importance of Mutations and gene duplication in evolution

Mutations are the only source of new variation. New alleles of genes arise. Most mutations are **harmful** or **lethal** but rarely, can be **advantageous**. Mutations are the driving force of **evolution**.

Gene duplication is important for the evolution of a species as it can facilitate the creation of new **genes**. Normally, changes to genes result in **loss** of function and so are **deleted** by natural selection. However, when genes are **accidentally** duplicated by mutation, it creates a gene that can accumulate further mutations, which over time can give rise to related genes but with a new **specialised** function.

Polyploidy

Polyploidy is the result of an error occurring during **gamete formation** or **cell division** and all the matching chromosomes fail to separate. Polyploidy is a mutation where cells receive one or more extra sets of chromosomes.

Importance of polyploidy in evolution	**Importance of polyploidy in human food crops**
Polyploid organisms may have an evolutionary advantage over diploid organisms because the **extra** sets of chromosomes have the ability to mask any conditions caused by recessive alleles. In addition, these duplicated chromosomes are free to accumulate mutations that may eventually result in a new beneficial trait.	Closely related plant species can be crossed and polyploidy induced to create polyploids. Polyploid plants are important in crop production since: - they are normally **larger** - they produce a higher **yield** of fruit - they show a greater **resistance** to disease.