

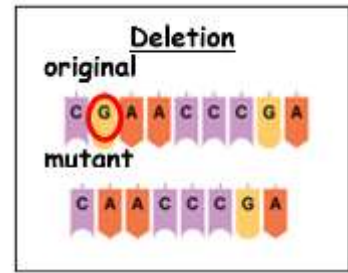
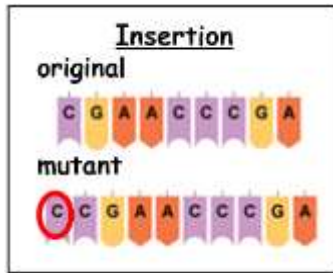
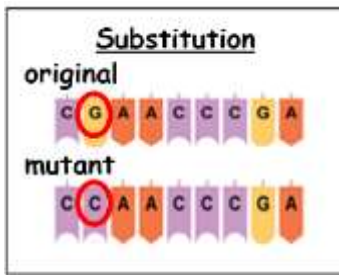
## 1.6. Mutations

When a cell divides, the process of DNA replication is very carefully controlled to preserve the \_\_\_\_\_ encoded within the \_\_\_\_\_ base sequence.

However, changes in the genome, known as \_\_\_\_\_, do occur and can result in there being no \_\_\_\_\_ or an altered protein being \_\_\_\_\_. Mutations arise \_\_\_\_\_ and at \_\_\_\_\_ but only occur \_\_\_\_\_.

### Single gene mutations

A single gene (\_\_\_\_\_) 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 \_\_\_\_\_ for another, this usually brings about only a \_\_\_\_\_ change to the protein, as it only affects one \_\_\_\_\_ therefore one \_\_\_\_\_. However, if the substitution occurs at a critical point, it can result in \_\_\_\_\_ changes such as Sickle cell anaemia.











<b>Types of single-nucleotide substitutions</b>	
	After a substitution, the altered codon codes for an <u>amino acid</u> which <u>still makes sense</u> but not the original sense.
	As a result of a substitution, a codon that used to code for an amino acid becomes changed into one that acts as a <u>stop codon</u> . 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 <u>non-functional</u> .
	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. <u>Splice-site mutations can alter post-transcriptional processing</u> .

### **Frame-shift mutations**

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

## Chromosome Mutations

Chromosome mutations alter the \_\_\_\_\_ of one or more \_\_\_\_\_.

<b>Types of chromosome mutations:</b>	
<p style="text-align: center;"><b><u>Duplication</u></b></p> <p>original </p> <p>mutant </p>	<p>A segment of genes is _____. Some duplication of genes may have a detrimental effect or be of an advantage.</p>
<p style="text-align: center;"><b><u>Deletion</u></b></p> <p>original </p> <p>mutant </p>	<p>A segment of genes becomes _____ and the two remaining ends _____ giving a shorter chromosome lacking the detached genes. Deletion normally has <b>drastic</b> effects on the living organism involved.</p>
<p style="text-align: center;"><b><u>Inversion</u></b></p> <p>original </p> <p>mutant </p>	<p>A segment of genes is _____. This results in non-viable gametes.</p>
<p style="text-align: center;"><b><u>Translocation</u></b></p> <p>original  </p> <p>mutant  </p>	<p>A section of one chromosome _____ off and _____ to another chromosome that is not its matching partner. This results in non-viable gametes.</p>

### Importance of mutations and gene duplication in evolution

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

**Gene duplication** is important for the evolution of a species as it can facilitate the creation of new \_\_\_\_\_. Normally, changes to genes result in \_\_\_\_\_ of function and so are \_\_\_\_\_ by natural selection. However, when genes are \_\_\_\_\_ 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 \_\_\_\_\_ function.

### Polyploidy

Polyploidy is the result of an error occurring during \_\_\_\_\_ or \_\_\_\_\_ (nondisjunction) and all the matching chromosomes fail to separate. Polyploidy is a mutation where cells receive one or more extra sets of chromosomes.

<b>Importance of polyploidy in evolution</b>	<b>Importance of polyploidy in human food crops</b>
<p>Polyploid organisms may have an evolutionary advantage over diploid organisms because the <b>extra</b> 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.</p>	<p>Closely related plant species can be crossed and polyploidy induced to create polyploids. Polyploid plants are important in crop production since:</p> <ul style="list-style-type: none"> <li>- they are normally _____</li> <li>- they produce a higher _____ of fruit</li> <li>- they show a greater _____ to disease.</li> </ul>