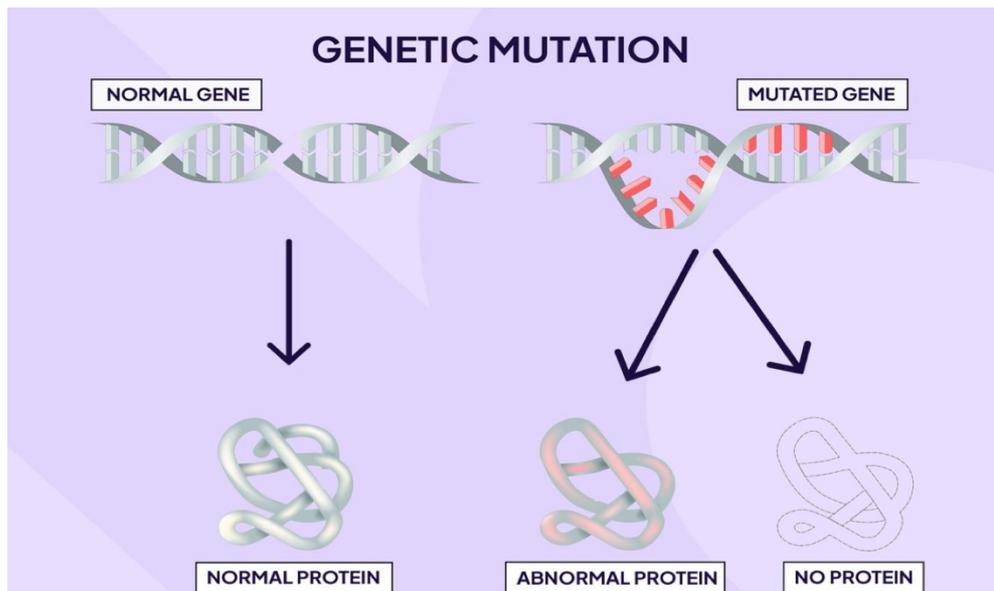


# Mutations

- **Mutations** are random changes in the sequence of an organism's **DNA** bases and happen continuously. They can be **inherited**, caused by exposure to certain substances and **radiation** or just occur **spontaneously**.
- **DNA** codes for specific **amino acid** sequences which create specific shaped **proteins**.
- Some **mutations** can have detrimental effects on the organism:
  - **Shape of Enzyme:** If the shape of an enzyme is altered, it will no longer be **complementary** to its **substrate**. They will not be able to bind and form an **enzyme-substrate complex** and it will not be able to **catalyse** the reaction.
  - **Shape of Structural Protein:** If **structural proteins** are altered, they could lose their **strength** and ability to provide support.
- Most **mutations** have little to no effect on the **protein** created, e.g. changing its shape only slightly so it is still functional.



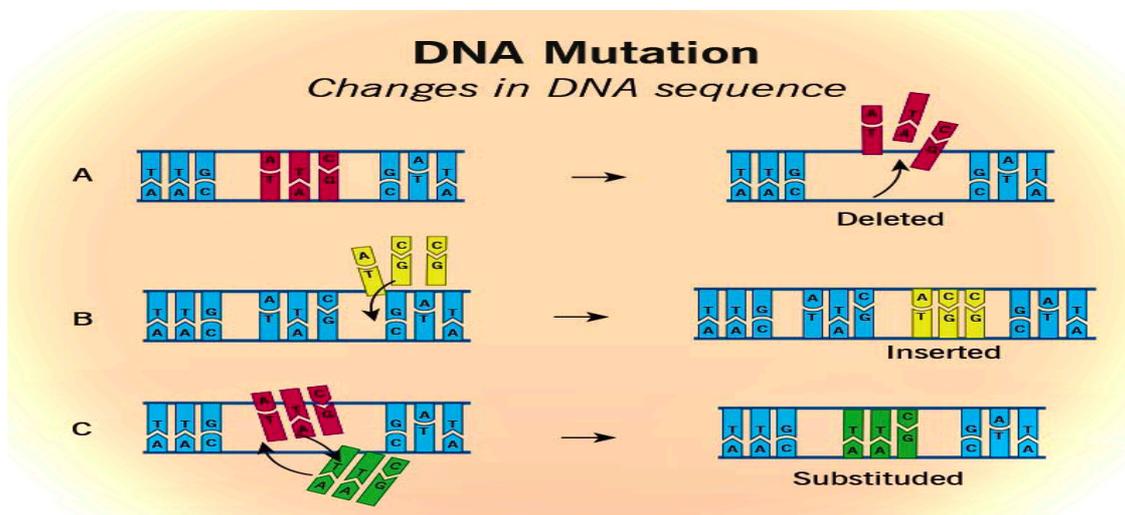
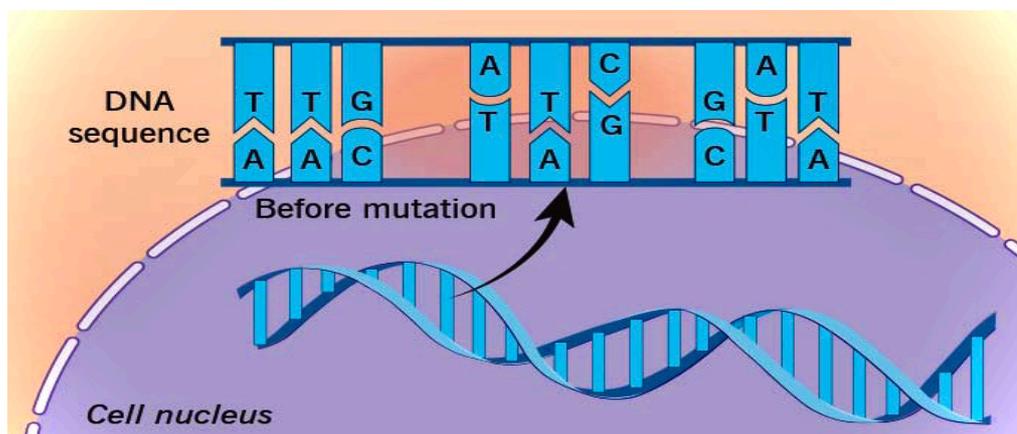
## Types of Mutation:

- Mutations in **non-coding** sections of **DNA** can change how genes are **expressed**.
- Types of **mutations** include:

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1. **Insertions**– A new base is **inserted** into the sequence where it is not supposed to be. As DNA is read in groups of **3 bases**, this can cause **knock-on effects** further down the sequence and cause major changes to the **amino acids** coded for and the **protein** produced.
2. **Deletions**– A base is **removed** from the sequence which will have **knock-on effects** further down the sequence and cause major changes to the **amino acids** coded for and the **protein** produced.
3. **Substitutions**– A base is **swapped** for another base, which can be **harmless** or have more serious effects depending on the base that is changed.



## **Genetic Disorder**

- ▶ Genetic disorders occur when a mutation (a harmful change to a gene, also known as a pathogenic variant) affects your genes or when you have the wrong amount of genetic material.
- ▶ Genetic order can be:
  1. **Chromosomal:** This type affects the structures that hold your genes/DNA within each cell (chromosomes). With these conditions, people are missing or have duplicated chromosome material.
  2. **Complex (multi factorial):** These disorders stem from a combination of gene mutations and other factors. They include chemical exposure, diet, certain medications and tobacco or alcohol use.
  3. **Single gene ( monogenic):** This group of conditions occurs from a single gene mutation.

### **- Genetic Disorders -**

**Common genetic conditions by type:**

#### **Single-gene:**

- Sickle Cell
- Cystic Fibrosis
- Tay-Sachs
- Marfan Syndrome
- PKD

#### **Chromosomal:**

- Down's syndrome
- Edwards' syndrome
- Patau's syndrome
- Turner syndrome

#### **Multifactorial:**

- Heart disease
- Alzheimer's disease
- Diabetes
- Cancer
- Arthritis

