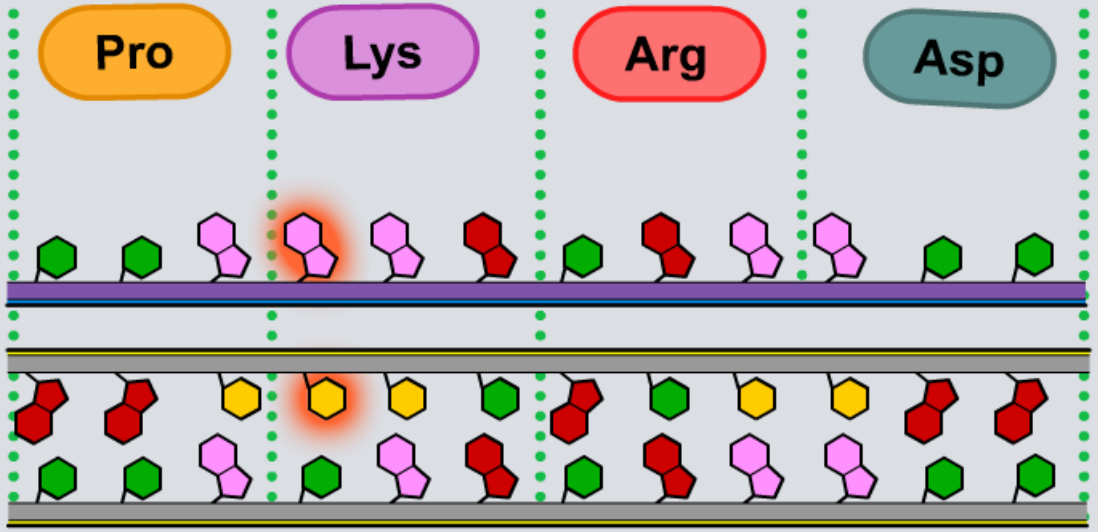
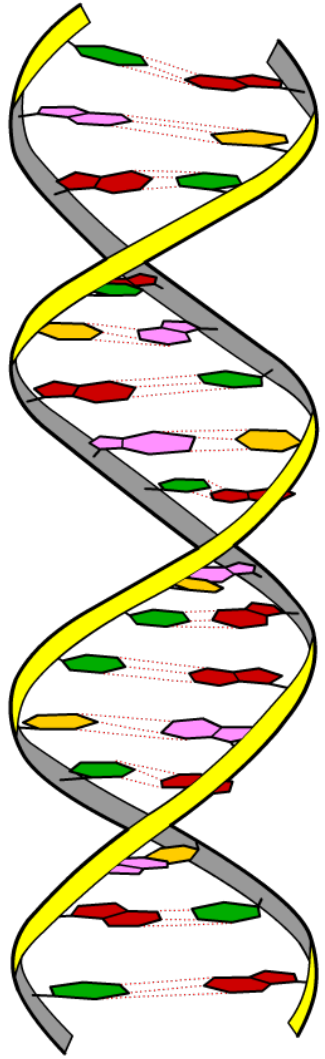
Genetic Mutations



Genetic mutations

A **mutation** is a change in the amount or structure of DNA. There are two types of mutation:



- A **gene** or **point mutation** – a change in the base sequence of a gene, which can cause a change in the polypeptide chain. It is caused by errors that occur during **DNA replication**.

- A **chromosome mutation** – a change in the number or structure of the chromosomes. It is caused by errors that occur during **cell division**.

Different types of point mutation

Mutagenic agents

The natural spontaneous mutation rate is typically around one or two mutations per 100,000 genes per generation.

The mutation rate can be increased by **mutagenic agents**, which include UV radiation and harmful chemicals. These disrupt the structure of DNA or interfere with transcription.

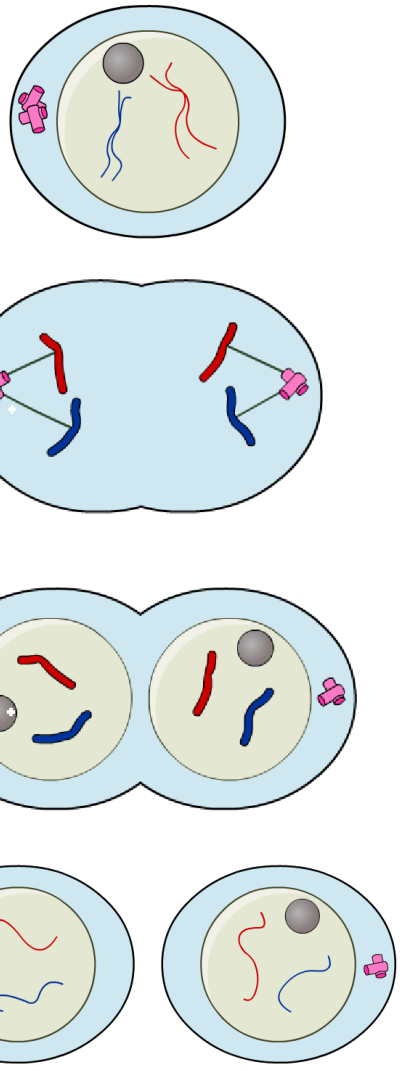
Mutations provide the genetic variation needed for evolution, however they can produce harmful effects.

Control of cell division

Cells divide to ensure that dead or damaged cells are replaced. The rate of cell division is controlled by two genes:

- **Proto-oncogene** – stimulates cell division.
- **Tumor suppressor gene** – slows or inhibits cell division.

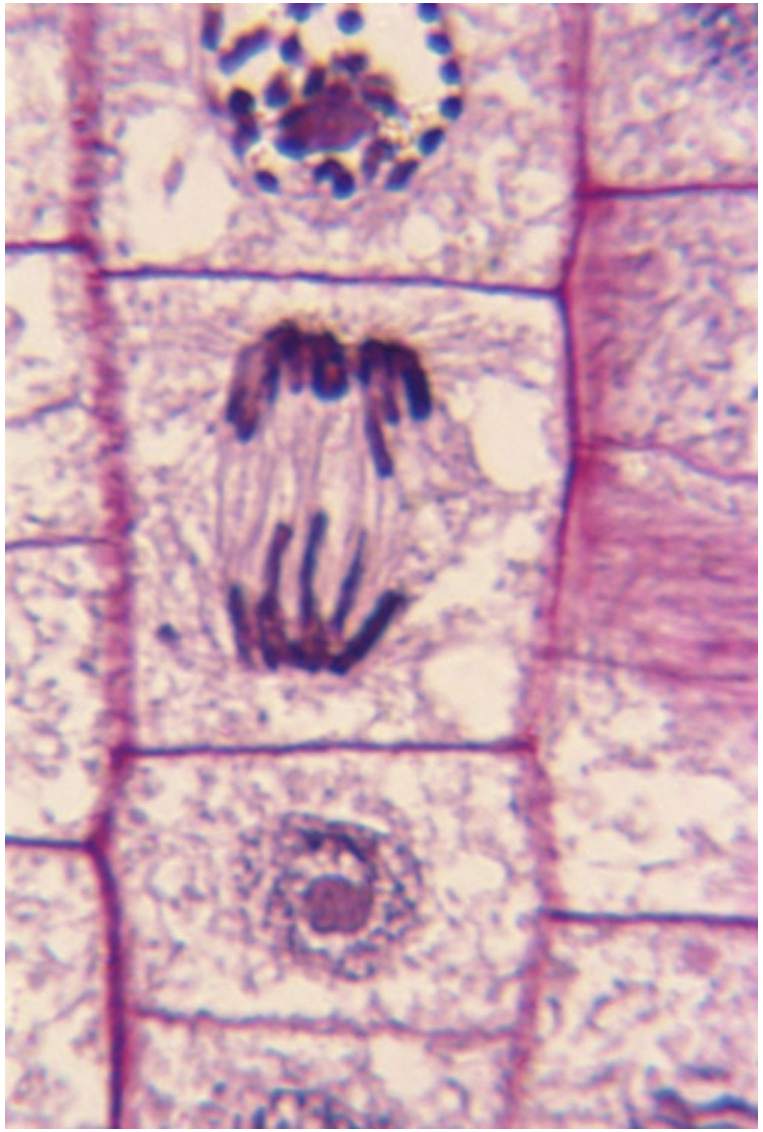
In normal cells the activities of these two genes are balanced; however, if a mutation occurs in one of these genes, problems can arise.



Oncogenes

If a point mutation occurs in a **proto-oncogene** it can form an **oncogene**. This can stimulate excessive cell division, leading to the formation of a tumor.

If a point mutation occurs in a **tumor suppressor gene** it can become inactivated. This allows the rate of cell division to increase unregulated.



Cystic fibrosis

Cystic fibrosis (CF) is a recessive inherited disease. It is caused by a number of different point mutations in the **CFTR** gene, which codes for a transmembrane protein that acts as an ion pump.

The CFTR gene is found on chromosome 7. It codes for 1480 amino acids. There are over 1000 known mutations, which can affect the function of the CFTR gene in different ways.

In around 70% of cases CF is caused by a triplet deletion, resulting in the removal of an amino acid from the polypeptide chain produced.

Mutations