

## Mutations

1. A mutation is a change in the genetic material of a cell - a change in the sequence of nucleotides of DNA.
2. **Effects of a mutation:**
  - changes to the base sequence results in coding for different amino acid sequences and consequently the production of a different polypeptide.
  - this may then result in changes in cell activity.
  - Harmful, beneficial or neutral effect
  - Impaired or reduced original function
  - Total dysfunction
  - Entirely new function
3. **Types of mutations:**
  - Change in genetic material:
    - **Change in DNA sequence:** substitution of a single base on a DNA strand causes a point mutation, deletion or insertion into a DNA sequence causes a frameshift mutation which changes the codon sequence along the DNA strand
    - **Gene mutations:** mutations affect a single gene e.g. cystic fibrosis, sickle cell anemia
    - **Chromosome mutation:** blocks of genes are moved to another part of the chromosome or another chromosome altogether, when someone has an abnormal chromosome number or aneuploidy, a pair of chromosomes failed to separate during meiosis resulting in one extra or one less chromosome e.g. Down Syndrome
  - Effect of mutation on phenotype:
    - **Somatic mutations:** changes in DNA sequences that may occur in somatic body cells, present in the individual only as cells carrying mutation divide by mitosis e.g. mutations leading to skin cancer
    - **Gametic mutations:** mutation occurs in the sexual reproductive cells which produce gametes, are inheritable and can be passed on to future generations e.g. cystic fibrosis (gene mutation) or Down Syndrome (chromosome mutation)
  - Origin of mutation:
    - **Spontaneous mutations:** arise randomly as a result of an error in a natural process such as DNA replication in cells.
    - **Induced mutations:** arise as a result of an environmental agent such as a chemical or radiation that increases the changes to nucleotide sequences e.g. cancer
4. A mutagen is an environmental agent that causes mutations.
5. Types of mutagens:
  - ❖ Chemical mutagens:
    - Alcohol
    - Cigarette smoke

- Pesticides
  - Cleaning products
  - ❖ Biological mutagens:
    - Viruses
    - Microorganisms
    - E.g. hepatitis B, HIV, HPV
  - ❖ Mutagenic radiation:
    - Ionising radiation from radioactive materials
      - X-rays, gamma rays
      - E.g. from atomic bombs, toxic spills
    - UV radiation from sunlight
6. Evidence of the mutagenic nature of radiation:
- Scientists involved in the study of radiation including Marie Curie who were exposed to large amounts over radiation for long periods of time developed cancer related illnesses.
  - Muller performed an experiment in which he irradiated the reproductive cells of fruit flies. He found that the irradiated fruit flies were more likely to gain a mutation than non-irradiated fruit flies. His experiments provided direct evidence of the link between exposure to radiation and mutations.
  - Survivors of the bombings of Hiroshima suffered physical mutations as a result of the radioactive output of the nuclear explosion. Many children suffered from microcephaly.
  - Victims of the Chernobyl leak suffered infertility and genetic mutations.