Mutations

What Are Mutations?

- Changes in the nucleotide sequence of DNA
- May occur in somatic cells (aren't passed to offspring)
- May occur in gametes (eggs & sperm) and be passed to offspring
- Changing amino acid sequence can mess up protein function or the organism's phenotype

Are Mutations Helpful or Harmful?

- Mutations happen regularly
- Almost all mutations are neutral
- Chemicals & UV radiation cause mutations
- Many mutations are repaired by enzymes (DNA Polymerase)

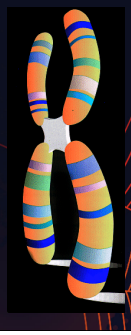
Are Mutations Helpful or Harmful?

- Some type of skin cancers and leukemia result from somatic mutations
- Some mutations may improve an organism's survival (beneficial)

Types of Mutations

Chromosome Mutations

- May Involve:
  - Changing the structure of a chromosome
  - The loss or gain of part of a chromosome

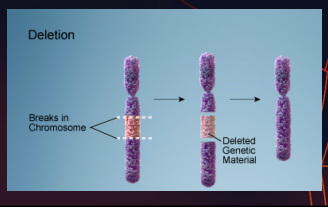


Chromosome Mutations

- Five types exist:
  - Deletion
  - Inversion
  - Translocation
  - Nondisjunction
  - Duplication

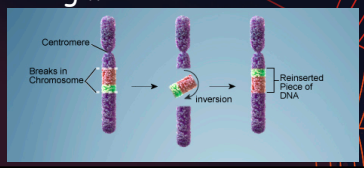
Deletion

- Due to breakage
- A piece of a chromosome is lost



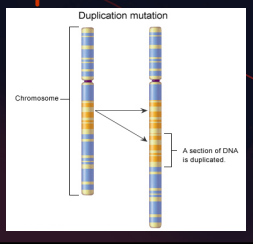
Inversion

- Chromosome segment breaks off
- Segment flips around backwards
- Segment reattaches



Duplication

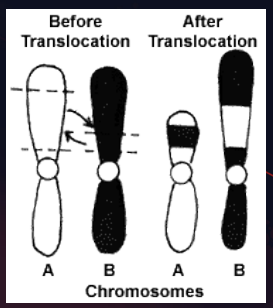
- Occurs when a gene sequence is repeated



Translocation

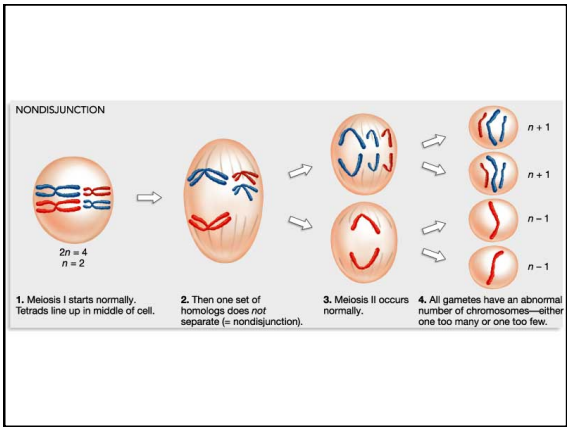
- Involves two chromosomes that aren't homologous
- Part of one chromosome is transferred to another chromosomes

Translocation

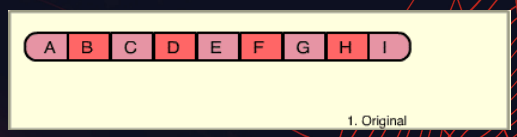


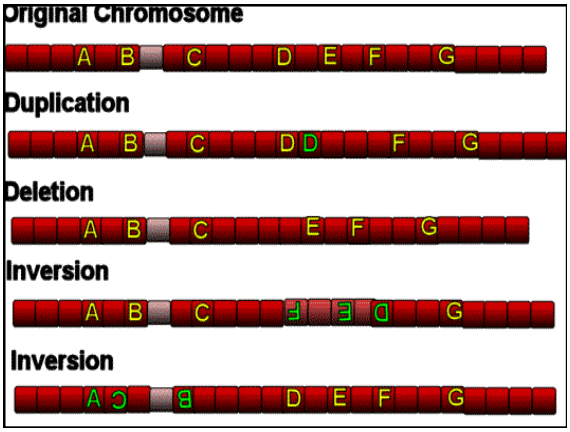
Nondisjunction

- Failure of chromosomes to separate during meiosis
- Causes gamete to have too many or too few chromosomes
- Disorders:
  - Down Syndrome – three 21st chromosomes
  - Turner Syndrome – single X chromosome
  - Klinefelter's Syndrome – XXY chromosomes



Chromosome Mutation Animation





Gene Mutations

- Change in the nucleotide sequence of a gene
- May only involve a single nucleotide
- May be due to copying errors, chemicals, UV radiation, viruses, etc.

Types of Gene Mutations

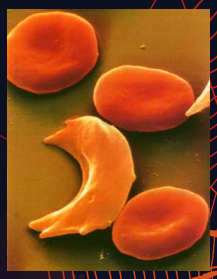
- Include:
  - Point Mutations
  - Substitutions
  - Insertions
  - Deletions
  - Frameshift

Point Mutation

- Change of a **single** nucleotide
- Includes the deletion, insertion, or substitution of **ONE** nucleotide in a gene

Point Mutation

- **Sickle Cell disease** is the result of one nucleotide substitution
- Occurs in the **hemoglobin gene**



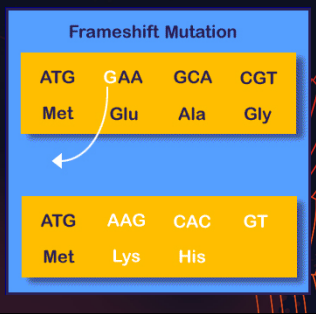
Frameshift Mutation

- **Inserting or deleting** one or more nucleotides
- Changes the "**reading frame**" like changing a sentence
- **Proteins** built **incorrectly**

Frameshift Mutation

- Original:
  - The fat cat ate the wee rat.
- Frame Shift ("a" added):
  - **The fat caa tat eth ewe era t.**

Amino Acid Sequence Changed



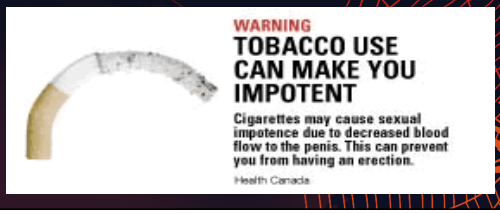
Gene Mutation Animation

Definitions

- **Carcinogens**
  Substances and preparations which, if they are inhaled or ingested or if they penetrate the skin; may induce cancer or increase its incidence and can affect any cells or tissues
- **Mutagens**
  may induce hereditary genetic defects or increase their incidence and effect the germ cells (gonads)
- **Teratogens**
  may induce non-hereditary congenital malformations or increase their incidence and effect the growing fetus

Mutagens

- Tobacco products
- Nitrous Acid
- Mold Toxins
- X-rays
- Gamma Rays
- UV Radiation
- Some Artificial Sweeteners



Cancer

- Cells gone wild!
  - Overproduction, underproduction, or production of proteins at wrong time
  - Can be influenced by environmental factors
    - Carcinogens