## Gene mutations

| | |
|---|---|
| *Substitution / point mutation* | **One base switches place with another.** This does not always lead to a change because DNA code is degenerate. (A change in the third base is the least likely to lead to change.) |
| *Deletion* | **Base deleted.** Results in frame shift - every codon after deletion is changed, so big change in protein synthesis. |
| *Insertion* | **Base inserted.** Leads to a frame shift. |

## Mutagens

| | | |
|---|---|---|
| **Physical mutagens** | *e.g. ionising radiations* | Break one/both DNA strands, mutations can occur in the process. |
| **Chemical mutagens** | *e.g. deaminating agents* | Chemically alter based in DNA (e.g. cytosine to uracil). |
| **Biological agents** | *e.g. alkylating agents* | Methyl/ethyl group added to base --> incorrect pairing. |
| | *e.g. base analogs* | Incorporate in DNA in place of usual base. |
| | *e.g. viruses* | Viral DNA inserted in genome. |

Mutagens are factors that increase the rate of mutations.

## Chromosome mutations

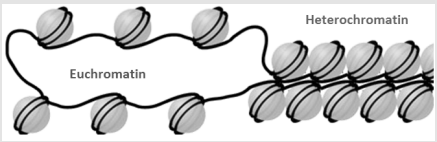
| | |
|---|---|
| *Deletion* | Sections of chromosomes break off. |
| *Duplication* | Section gets duplicated. |
| *Translocation* | One section of a chromosome breaks off and joins to non-homologous chromosome. |
| *Inversion* | Section breaks off, reverses and joins back on. |

## Chromatin remodelling

| | |
|---|---|
| *Heterochromatin* | Tightly wound DNA around histones. |
| *Eurochromatin* | Loosely wound DNA. RNA polymerase can only access genes when DNA is eurochromatin. |

Chromatin is a DNA-protein complex. Also note the dark green boxes will be related to **transcriptional control**.

## Eurochromatin v. Heterochromatin

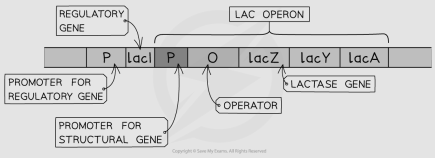



## Histone modification

| | |
|---|---|
| *Acetylation / Phosphorylation* | Lowers the positive charge of histones, so DNA is looser and genes can be transcribed. |
| *Methylation* | Makes histones more hydrophobic so DNA is wound tighter. Genes cannot be transcribed. |

DNA is negatively charged and histones are positively charged, which is how they attract. Changing charges will change the attraction.

## Operons - Lac Operon




## Lac Operon

An operon is a group of genes under control of the same regulatory mechanism --> switch genes in and off depending on environment.
Lac Operon --> digests lactose if glucose concentration is low in E. Coli.
In the presence of glucose, a regulatory genes produces repressor proteins which bind to the operator so RNA polymerase cannot bind and read genes.
In the presence of lactose, lactose binds to the repressor protein, changing its shape so it no longer binds to the operator and the structural genes that code for lactose-digesting enzymes can be transcribed.
cAMP acts as a secondary messenger. It speeds up the transcriptional of lactose genes if glucose is low.

## Post-transcriptional control

| | |
|---|---|
| *RNA processing* | Transcriptional makes pre-mRNA and transforms it in mature mRNA. Once DNA has been transcribed, introns are removed from the pre-mRNA. |
| *RNA editing* | Change in base sequence to make different proteins (deletion, addition...). |

## Translational control

| | |
|---|---|
| *Degradation of mRNA* | Higher resistance of mRNA = longer lasting in the cytoplasm so higher quantity of protein synthesis. |
| *Inhibitory proteins* | Prevent binding to ribosomes. |
| *Initiation factors* | Aid binding to ribosomes. |
| *Protein kinases* | Adds phosphate group to protein, changing tertiary structure and function. Many proteins activated by phosphorylation, important for cell regulation. |

## Post-translational control

Addition of non-protein group

Modifying amino acids + bonds

Folding / shortening of proteins

Modification by cAMP (e.g. Lac operon)

## Homeobox gene

Regulatory genes that code for a homeodomain protein which switches genes on and off.

Code for body plan - basic layout of animals, fungi and plants.

Highly conserved across each species.

180 base pairs long.

## Hox genes

Homeobox gene only present in animals.

Codes for correct placement of body parts.

Found in gene clusters (4 in mammals with 39 Hox genes divided in 4 clustered in humans).

## Body plans

| | |
|---|---|
| *Body plans* | The way the body is arranged. |
| *Somites* | Segments in embryo from which individual vertebrae develop. Hox genes in the mouth area will code for mouthparts, in the thorax for arm placement... |
| *Symmetry* | Radial e.g. in jellyfish |
| | Bilateral e.g. humans |
| | None e.g. sponges |

## Mitosis and apoptosis

| | |
|---|---|
| *Mitosis* | Increase number of cells for growth |
| *Apoptosis* | Programmed cell death to remove unwanted tissue (e.g. webbing between fingers in womb). |

## Apoptosis