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Lecture title: GENES

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Gene: the genetics unit that carries genetic information.

Or it is a sequences of nucleotides encoded into a polypeptide or RNA.

The gene in its location cannot do or produce anything, it only stores information in the form of nucleotide sequences, and without an external effect on it the gene does not start any action on its own.

Genome: is the complete genetic material of the organism, consisting of coded and non-coded genes of DNA (sometimes RNA!!)

Allele: Various sequences of the same gene, such as the polymorphism of different parts of DNA.

Introns: Intervening sequences that are eliminated during the mRNA processing (splicing).

Exons: Expressing sequences that make up mature mRNA, usually the gene begins and ends with exons.

Organism	Size of DNA (1000 bp)	No. of genes
E. coli	4.6×10^6	4288
Drosophila melanogaster	1.3×10^8	13600
Mus musculus	2.7×10^9	20210

Information carried by genes

- Genetic information in the cell can be compared to the language. The letters and their sequences are the words that represent the information.
- The gene is composed of words of nucleotides (four letters A, T, G and C).
- In the genes the different words consist of **only three** characters, called **codon**.
- There are two types of information that is encoded during protein synthesis:

1. Every three letters of nucleotides represent a certain amino acid.
2. The location of each amino acid within the protein.

The two types of information above determine the **primary** structure of the protein (the linear arrangement of amino acids within the protein structure).

In addition, both sides of the gene have sequences of regulatory functions called regulatory sequence. These sequences do not have a final product, but they are responsible for regulatory chemical signals.

Different types of these regulatory sequences are the most important:

1. Promoters.
2. Operators.
3. Attenuators.
4. Enhancers.

Regulation of gene expression

1. Gene expression is subjected to regulation throughout the life span of prokaryotic and eukaryotic cells.

2. Total DNA or whole genome is never expressed at any given time.

3. Expression of genes is **turned on** or **turned off** depending on the biological needs, developmental stage or time, environmental (external) and nutritional factors.

For example, when cell prepares for division only genes that have role in cell division are expressed like genes which code replication enzymes and genes that code proteins which are essential for proper cell division. After cell divides expression of these genes is turned off and expression of genes that are involved in various housekeeping activities or maintenance of cell is turned on.

Furthermore, the extent of expression of genes that are involved in various cellular processes also differs. For example, enzymes of glycolysis and TCA cycle are expressed several times whereas enzymes of gluconeogenesis are expressed only when needed. When cell completes its lifespan apoptosis occurs due to expression of certain genes associated with it.

4. Development of embryo to infant involves turning on or tuning off of several genes.

5. **Gene expression is tissue specific and species specific.** Even though all eukaryotic cells contain same information, its expression depends on tissue. For example, insulin is expressed in pancreas but not in other tissues. Similarly, urea cycle enzymes are expressed in liver only.

6. Depending on environmental or nutritional factors, expression of certain genes is increased or decreased. For example, expression of key enzymes of gluconeogenesis is increased in starvation.

7. In eukaryotes, gene expression is affected by hormones.

8. Cancer is due to altered gene expression.

Regulation of gene expression by hormones and vitamins:

Many steroid hormones, retinoic acid and vitamin D exert their action by affecting gene expression. They induce synthesis of enzymes and proteins in target cells. They form complex with receptor after entering target cell. These complexes migrate to nucleus where they bind to specific sequence of DNA known as hormone responsive element (HRE) or enhancer. This leads to activation of promoters and binding of RNA polymerase to DNA and thus transcription.