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TURNING ASPIRANTS INTO ACHIEVERS

SCIENCE & TECHNOLOGY

FOR UPSC PRELIMS & MAINS

EXAM-ORIENTED

CONCEPT -CENTRIC

STRUCTURED & SIMPLIFIED

"Innovation is the ability to see change as an opportunity—not a threat." —

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SECTION I: BIOTECHNOLOGY

Chapter 1: GENETICS

Genetics is the branch of biology that studies **genes, genetic variation, and heredity** in living organisms.

In essence, it explores how traits and characteristics are passed down from parents to offspring.

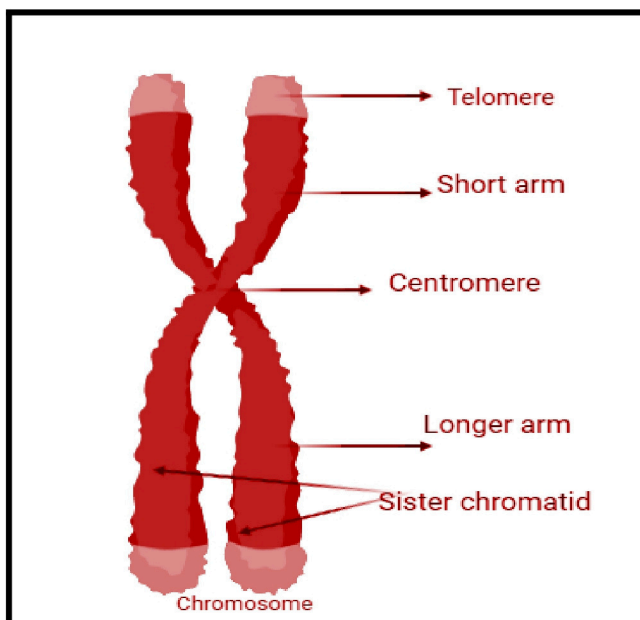
1.1. Key Concepts in Genetics

Heredity: The process by which traits are passed from one generation to the next.

Gene: The basic physical and functional unit of heredity. Genes are specific sequences of DNA that provide instructions for making proteins.

Chromosome: A long, thread-like structure of DNA, tightly coiled around proteins, found inside the nucleus of a cell. Humans have 23 pairs of chromosomes.

Structure of chromosome:



- A human cell has a pair of homologous chromosomes. A homologous chromosome is one with chromosomes of the same length and centromere positions for the genes corresponding to the same loci.

- These chromosomes are made up of chromatin, which contains the DNA or Deoxyribonucleic Acid and other associated proteins.

- This DNA is tightly coiled around histones proteins which makes the structure of Chromosomes.

- These chromosomes contain thousands of genes which can precisely code for several proteins in the cell.

Types of Chromosomes

Autosomes Chromosomes	Sex Chromosomes
These are non-sex chromosomes and thus control the somatic characteristics	They determine the gender of the individual
There are 22 pairs of autosomal chromosomes in humans.	Only one pair of sex chromosomes is present in humans.

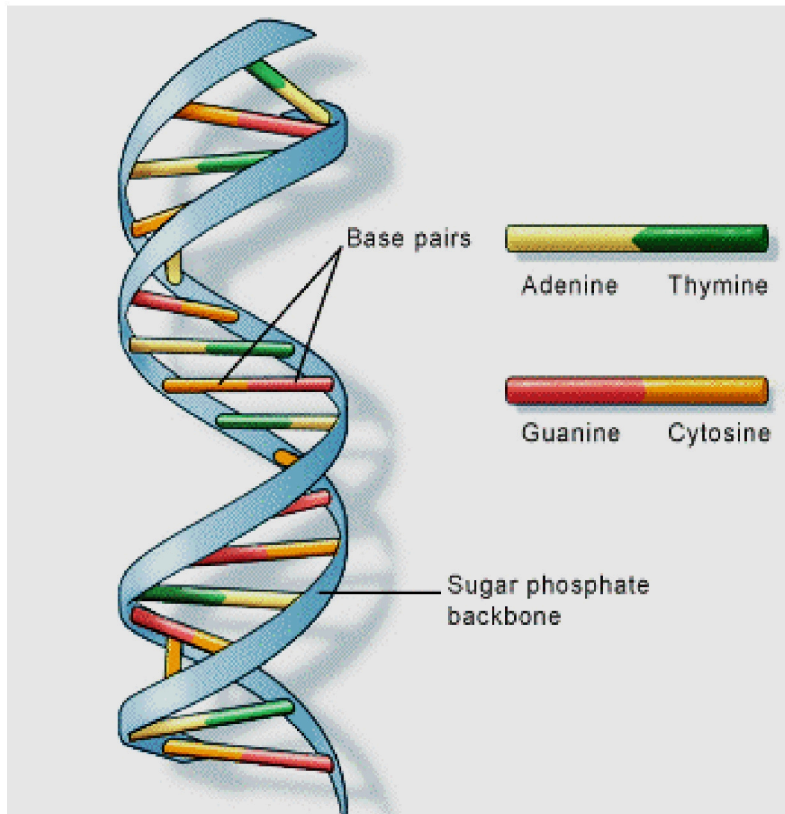
Nucleic Acid

They are complex compounds in the cells. It includes DNA (Deoxyribonucleic Acid) and RNA (Ribonucleic Acid). It contains genetic information in the form of genes which are made up of nucleotide bases.

DNA

Deoxyribonucleic Acid is the genetic material of humans. It was discovered by Frederic Meischer and the structure of the DNA was given by Watson and Crick and is famously known as the Watson and Crick model of DNA.

Structure of DNA



It has three components:

1. Nitrogenous base: DNA is made up of 4 nitrogenous bases namely:

- a. Adenine
- b. Thymine
- c. Cytosine
- d. Guanine

Adenine bonds with Thymine and Cytosine bonds with Guanine by double and triple hydrogen bonds respectively.

2. Sugar
3. Phosphate

Apart from the nucleus DNA is also present in the Mitochondria of a cell and is called mtDNA.

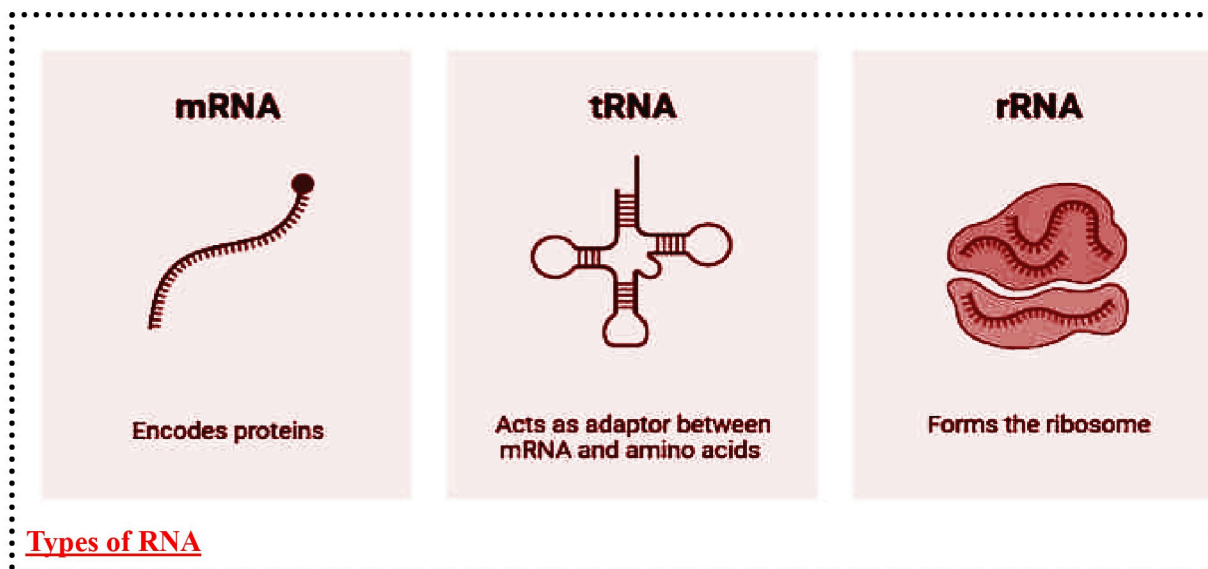
Nuclear DNA	Mitochondrial DNA
It has approximately 20000-25,000 genes.	It has 37 genes.
No. of base pairs are 3 billion.	No. of base pairs is 16,569.
	It gets replicates independently of Nuclear DNA

RNA: Ribonucleic Acid

RNA is generally a single-stranded nucleic acid. Various double-stranded RNA includes rotavirus and Reovirus. RNA makes the protein.

Types of RNA

- **Messenger RNA (mRNA)** transfer message from DNA found in the Nucleus to the cytoplasm in a coded form.
- **Ribosomal RNA (rRNA)** is the site of protein synthesis
- **Transfer RNA (tRNA)** is the carrier of amino acid and transfers it to the ribosome.



Difference between DNA and RNA

Feature	DNA	RNA
Sugar	Deoxyribose	Ribose
Genome	Generally double stranded	Generally single stranded
Nucleotide Bases	Adenine, Guanine, Cytosine, Thymine	Adenine, Guanine, Cytosine, Uracil
Location in human cell	Nucleus	nucleus and cytoplasm.

- **Allele:** A specific variant or version of a gene. For example, the gene for blood type has different alleles (A, B, O).
- **Genotype vs. Phenotype:**
 - **Genotype:** The complete set of genes an organism possesses.
 - **Phenotype:** The observable physical or biochemical traits of an organism, which result from the interaction of its genotype with the environment (e.g., eye color, height, blood type).

1.2. Mutation

It is the process of alteration in the nucleic acid sequence of the genome. It is responsible for variations and evolution. The term mutation was coined by **Hugo de Vries** in 1909. Anything that can cause mutation/ agent of mutation is called mutagen.

Types of Mutation:

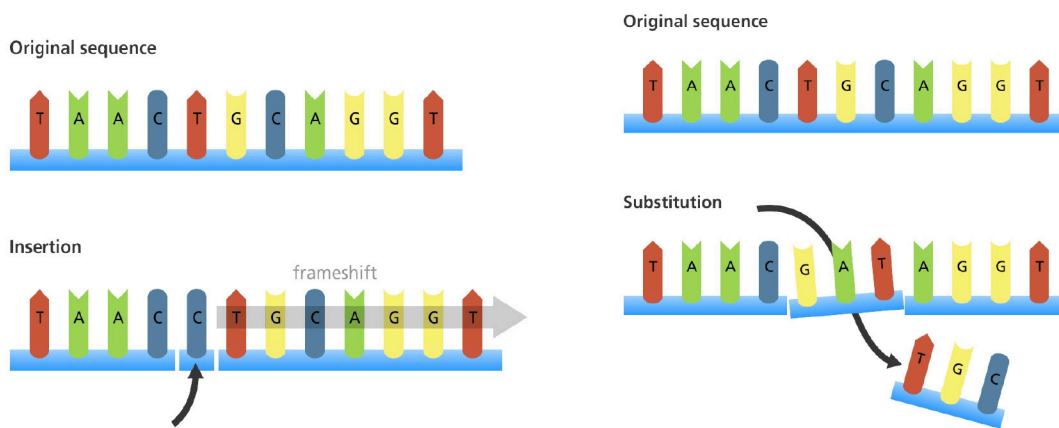
➤ On the basis of types of cells

- Somatic cell mutation** refers to change/alteration in a person's DNA that occurs to any cell other than the germ cell (egg or sperm cell).
 - They don't pass from parents to their children (not hereditary).
 - It occurs happen sporadically or randomly, without the mutation existing in a person's family history.

- iii. They don't play any role in evolution.
- iv. Example: Skin cancer, Lung cancer etc.
- b. **Germline mutation** occurs in a parent's reproductive cells (egg or sperm) of the organism. These mutations change the genetic material are hereditary and can play a role in evolution as well. Example Sickle cell anaemia etc.

➤ **On the basis of types of size**

- c. **Point mutation** refers to a change in one base by either insertion, deletion or substitution in the DNA sequence. For example, single base pair in the beta-globin chain of haemoglobin pigment in the blood leads to sickle cell anaemia.
 - i. **Substitution mutation** refers to an alteration in one or more bases in a sequence that is replaced by another



nucleotide base.

- ii. **Insertion mutation** refers to the addition of one nucleotide base in a sequence.
- iii. **Deletion mutation** refers to the removal of one nucleotide base in a DNA sequence.
- b. **Gross mutations** are mutations which involve large portions of the genetic material.

➤ **On the basis of the cause of mutation**

Spontaneous Mutation	Induced mutation
Occur naturally in the genome.	Induced mutations do not occur naturally.
Generally occur due to errors in replication or cell division etc.	Are induced through various chemical and physical agents known as mutagens , which enhance the frequency of mutation, such as Alkylating agents, radiations etc.

➤ **On the basis of Impact of Mutation:**

- **Neutral mutations:** The mutation which does not have any impact on the individual/ species is called a neutral mutation, they are neutral as they don't impact the protein synthesis in the body.
- **Negative mutation:** These mutations are harmful mutations, for example, a change in a gene causes Cystic fibrosis which causes thick and sticky mucus that clogs lungs etc.