

Human Genetics

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Human genetics is both a fundamental and applied science. As a fundamental science, it is part of genetics – the branch of science that examines the laws of storage, transmission, and realization of information for the development and function of living organisms.

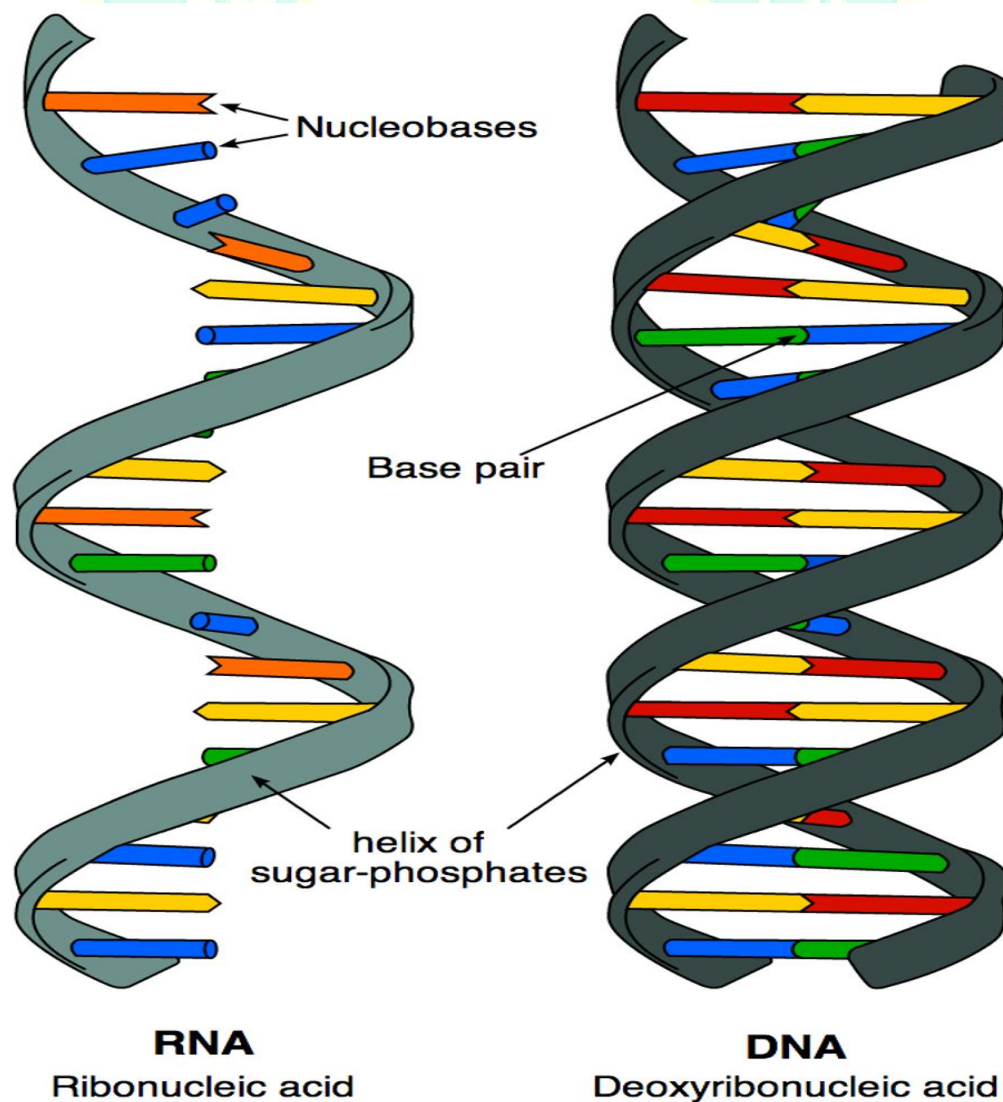
Our bodies contain millions of cells, which are the building blocks that make up all tissues in the body (eg skin, bones and muscles).

Inside cells is DNA, which tells cells how to function. DNA can be thought of as an instruction manual that tells the body how to function.

Cells work differently depending on which parts of the DNA are used to obtain the information, individual genetic information that determines characteristics (eg the color of your eyes and hair or your blood type).

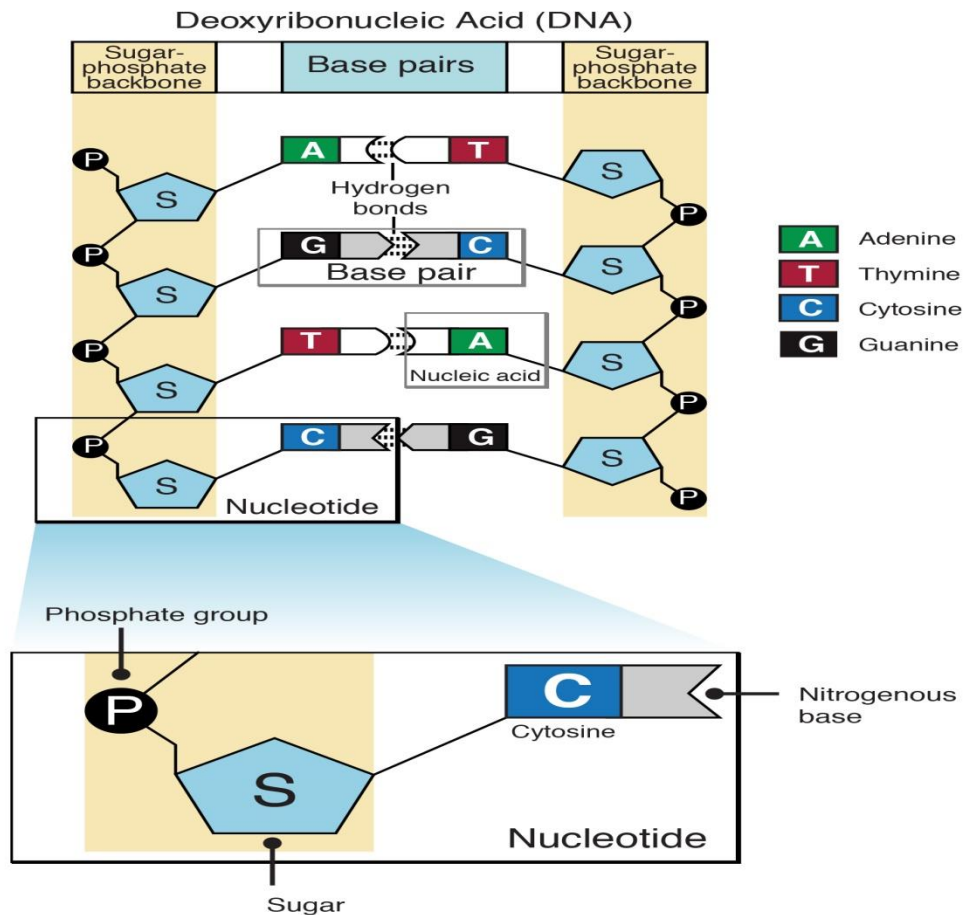
Nucleic acids

A substance called deoxyribonucleic acid (DNA) is the substance of which genes are made , another substance called ribonucleic acid (RNA), are together referred to as **nucleic acids**.



Deoxyribonucleic acid (DNA):-

- ✓ DNA is a double stranded molecule consists of 2 polynucleotide chains running in opposite directions.
- ✓ Both strands are complementary to each other.
- ✓ DNA consist of three different components. These are the dexoyribose sugar, a phosphate group, and a nitrogen base.
- ✓ The bases are on the inside of the molecules and the 2 chains are joined together by double H-bond between A and T and triple H-bond between C and G.



Ribonucleic acid (RNA):-

Three major kinds of RNA have been identified in cells. These are referred to as:-

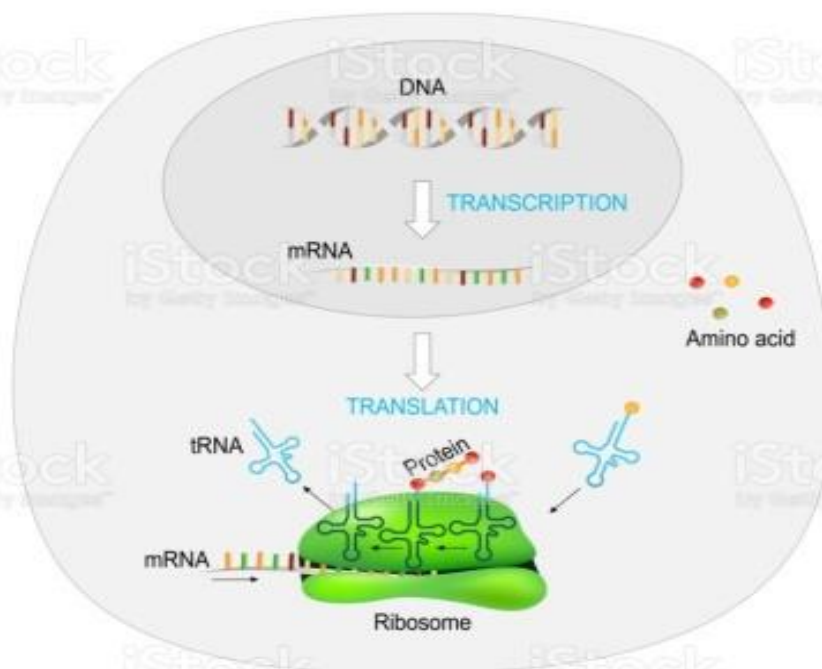
1. Messenger RNA (mRNA),
2. Ribosomal RNA (rRNA), and
3. Transfer RNA (tRNA).

Each type of RNA has a specific role in protein synthesis.

RNA

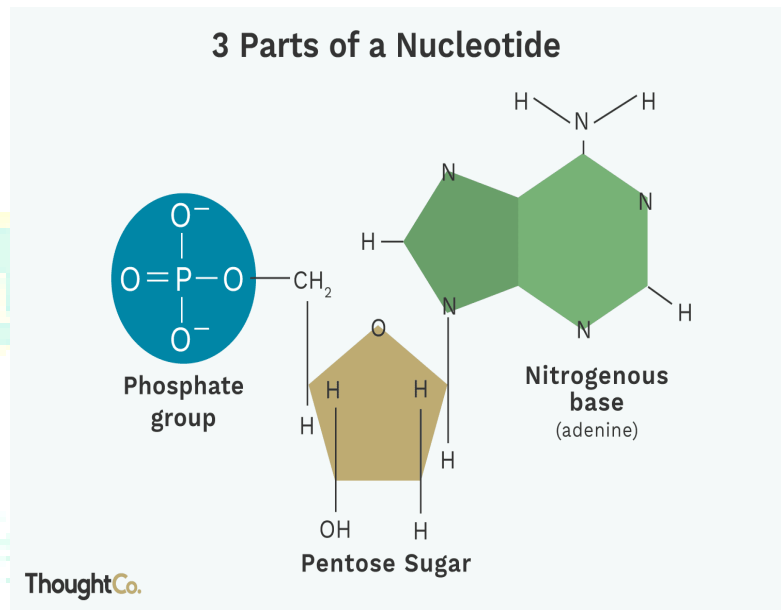


Protein synthesis



Nucleotides:

Nucleotides are the structural units of nucleic acids. Nucleotides are named according to their nitrogenous base.



Each nucleotide has three parts:

1. A nitrogen-containing base

Purines and pyrimidines: The nitrogen containing bases are cyclic compounds made up of carbon, hydrogen, oxygen, and nitrogen atoms. The bases are named adenine (A), thymine (T), cytosine (C), guanine (G), and uracil (U). A and G are double-ring structures called purines, whereas T, C, and U are single ring structures referred to as pyrimidines.

2. A pentose (five-carbon) sugar called deoxyribose or ribose
3. A phosphate group (phosphoric acid)

Genetics: Genetics is the study of genes, their structure and function, heredity and variation.

Genomics: The study and analysis of the nucleotide sequence of DNA is called genomics.

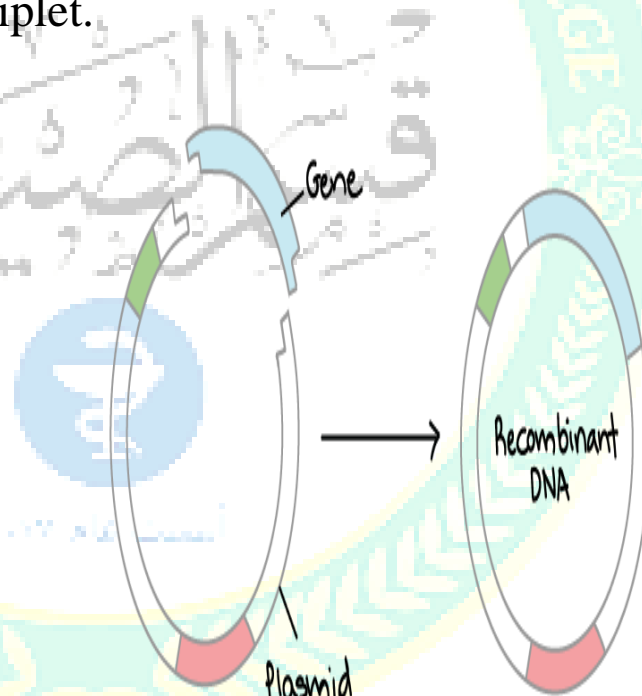
Genome: The complete set of genetic information for a cell is referred to as its genome.

Code:- Genetic information is stored in DNA as a code.

Codon:- The unit of code is. It consists of a sequence of three bases. Therefore, code is triplet.

Gene:- a segment of DNA carrying a number of codons specifying for a particular polypeptide is known as gene.

Plasmids:- most bacteria possess extra chromosomal genetic element in addition to chromosomal DNA elements known as plasmid.



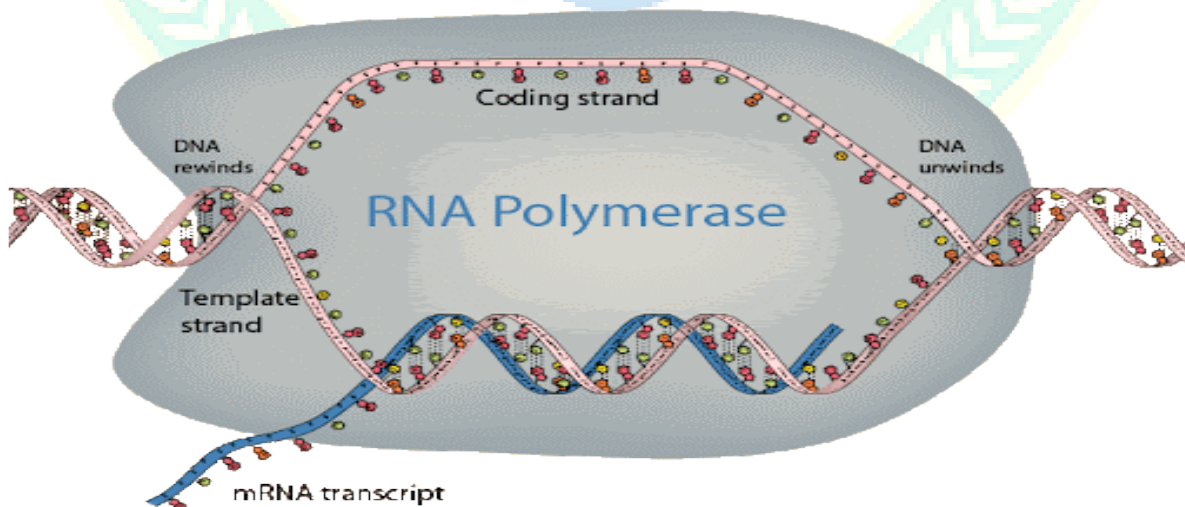
Gene Expression

Gene expression involves two separate but interrelated processes transcription and translation.

1. **Transcription:** Transcription is the process of synthesizing RNA from a DNA template. The DNA acts as a template for the transcription of RNA by RNA polymerase for subsequent protein production within the cell.

RNA polymerase attaches itself to the beginning of a gene on DNA and synthesizes mRNA, using one of the strands in DNA as a template. This process is known as transcription. The bases in mRNA will be complementary to one strand of DNA since DNA acts as a template for synthesis of mRNA.

2. **Translation:** Translation is the process of decoding the information carried on the mRNA to synthesize the specified protein.



Mutation

It is a random, undirected, heritable variation caused by an alteration in the nucleotide sequence at some point of the DNA of the cell.

Types of Mutation

Mutations can be divided conveniently into:

1. **Spontaneous mutation:** Many mutations occur spontaneously in nature in the absence of any mutation-causing agents.
2. **Induced mutation:** The frequency of mutation is greatly enhanced by exposure of cells to several agents (mutagens) which may be physical or chemical.

