

# Genetics.

classmate

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**Genetics:-** Genetics is a branch of biology which concerned with the study of genes, genetic variation and heredity in organism.

**Genetic materials:-** The substance that gets transferred from one generation to the next generation in order to express its characteristics in parental characters.

The function of genetic materials are-

- It should be capable of replicating and being inherited to offspring.
- It should be able to carry out all the information necessary for the function of a cell.
- It should be able to change its sequence by mutation.

DNA and RNA are the principal genetic materials of living organism and chemically called nucleic acids.

**Gene:-** The unit of heredity transmitted from one generation to the next generation is called a gene. A gene represents the heredity unit which is responsible for inheritance of genetic ~~or~~ characters of sex organism.

The term gene was coined by Danish Gene theorist, Wilhelm Johannsen in 1909. Genes (so called factors) transmits individual traits (called characters according to Mendel).

## classical concept of gene

Based on the classical concept, the gene is regarded as

- (1) The unit of function. Acc to this definition gene are consider as the unit of chromosome responsible for the expression of a trait.  
Example:- blue or brown eyes in humans.
- (2) The unit of mutation. Acc to this definition gene are the smallest segment of chromosomes capable of undergoing mutation.

## Basic requirements for DNA replication

The enzymes, protein factor and metabolites required for this process are:-

- (a) DNA helicase: It is an enzyme which breaks the hydrogen bond and separates the DNA strands. Thus, a fork is formed at the junction known as replication fork.
- (b) Single stranded binding (SSB) proteins: These are the molecules which attach tightly to the exposed single stranded DNA in order to stabilize the single-stranded DNA long enough for replication.
- (c) Primase:- It is an enzyme responsible for synthesis of short RNA primers. A RNA primer is a small strand of RNA which guides the process of replication.
- (d) DNA polymerase:- It is an enzyme responsible for causing nick in the DNA. (Nick means cutting or breaking of phosphodiester bond)
- (e) DNA polymerase: It is an enzyme responsible for catalyzing the synthesis of DNA.
- (f) Topoisomerase:- It is an enzyme responsible for causing nick in the DNA. (Nick means cutting or breaking of phosphodiester bond)
- (g) RNase: It is an enzyme which digests the RNA primers after the DNA synthesis is over.
- (h) DNA ligase: It is an enzyme which seals the gap in the synthesized DNA fragments.
- (i) Substrates:- The four deoxyribonucleotide triphosphate (dNTPs) such

as dATPs, dGTPs, dCTPs, dTTPs.

- ⑨ Folic acid: It is an essential requirement for the synthesis of nitrogenous base.
- ⑩  $Mg^{2+}$  and  $Mn^{2+}$  ions: These ions are essential for DNA synthesis.

The mechanism of DNA replication is a complex phenomenon which involves the following steps:-

① Origin of replication site: DNA is long chain of poly nucleotides. It has many replication units called replicons. Replication is initiated at each replicon by producing a nick (cut) in one of the strands at a specific initiation point called 'ori-site'. The nick is produced by an enzyme called endonuclease.

② Activation of deoxyribo nucleotides: Free nucleotides present in the nucleoplasm as deoxyribo nucleotide monophosphates (dNMPs) - dAMPs, dGMPs, dCMPs, dTMPs are activated into triphosphates (dATPs, dGTPs, dCTPs, dTTPs) with the help of an enzyme phosphorylase in the presence of ATP. This process is known as phosphorylation.

③ Unwinding of DNA helix: The unwind of two DNA strands occurs in the presence of an enzyme helicase which breaks the hydrogen bonds between the nucleotides. Due to the unwinding of two DNA strands, a Y-shaped fork called replication fork is formed. Now, both separated DNA strands are called templates.

④ Formation of RNA primers:

- Replication is guided by a RNA primer. RNA primer is a small strand of RNA which is synthesized by an enzyme primase. Primers is attached on the template DNA at a site called initiation site from where the DNA synthesis begins

- Replication always initiated from 5' direction to 3' direction.

⑤ Elongation of new strand.

- Once, the primer strands is formed, DNA replication begins with the help of DNA polymerase III (In prokaryotes) & DNA polymerase (In eukaryotes) along with ATP and  $Mg^{2+}$ .

- Nucleotide chain formation proceeds from the initiation site by addition of new bases.
- Replication proceeds on both the template DNA strands: therefore, the replication process is bidirectional. Replication is continuous on one template strand i.e. single DNA primer is responsible for the formation of whole strand, thus new strand is formed which is called a leading strand.
- After a formation of a new nucleotide chain is completed, RNA primer is removed and the gaps get filled with the complementary bases.

⑥ Termination: - The replication is terminated whenever two replication forks meet.

⑦ Proof-reading and DNA repair: - Sometimes, the wrong bases may be ~~inserted~~ inserted during replication. It is an error. The probability of which error is about once per 100000 nucleotides. The error is corrected by inserting the correct nucleotides. This process is called proof-reading.

The replication in which the half of the original DNA is ~~are~~ conserved and half is the newly synthesized one is called semi-conservative mode of replication of DNA.

DNA or RNA carries all the genetic information. DNA or RNA is formed by the four alphabets A, G, C, T or U, and the alphabet for the DNA molecules are the nitrogenous bases such as Adenine (A), Guanine (G), Cytosine (C) and Thymine (T). Similarly, the nitrogenous bases of RNA are Adenine (A), Guanine (G), Cytosine (C) and Uracil (U). These four alphabets encode the hereditary messages and are called as code letters or codons. The linear arrangement of nitrogenous bases in the RNA determines the sequence of amino acid.

Therefore, the genetic code is defined as a dictionary of nucleotide bases (A, G, C and U) that determines the sequence of amino acids in the protein of a cell. It is also defined as the dictionary that helps in translating the language of the nucleic acid into the language of protein. The group of nucleotides that specifies or codes for one amino acid is known as codon or code word.

There are four nucleotide bases; namely the Adenine (A), Guanine (G), Cytosine (C) and Uracil (U). These four bases form 64 different combinations ( $4 \times 4 \times 4 = 4^3 = 64$ ) of three base codons. Out of 64 codons, 61 codons code for the 20 amino acids found in proteins. The remaining three codons UAA, UAG and UGA do not code for amino acids. Rather they act as stop signals in the protein synthesis.

1. The code is triplet: A codon is triplet in nature. It means that for any amino acid it is necessary to have a triplet codon. A triplet codon codes for a particular amino acid.

2. The code is degenerate: Most of the amino acids have more than one triplet codon. For example: glucose has four triplet codons. Similarly, arginine has six triplet codons. These are called degenerate codons.
3. The code is not overlapping: A base is a part of only one codon. The same base letter is not used for two different codons. For example: a nucleotide sequence CAT, GAT is read as CAT and GAT. It represents only two codons i.e. CAT and GAT which is not overlapped.
4. The code is commaless or punctuationless: The genetic code is continuous and doesn't possess any pause or gap after the triplets. If a nucleotide is deleted or added, the whole genetic code and read differently.
5. The code is non-ambiguous: It means that there is no ambiguity about a particular codon. A particular codon will always code for the same amino acid, hence the genetic code is highly specific or unambiguous.
6. The code is universal: - The genetic code is applicable universally i.e. a codon specifies the same amino acid from a virus to a plant or human being.
7. The code is collinear: - The genetic code works in the principle of collinearity i.e. it explains the specific relationship between DNA, RNA and polypeptide chain. The linear order of nucleotides in DNA determines the linear order of codons in mRNA which in turn determines the linear order of amino acids in a polypeptide chain.

First Base (5' end)	Second Base				Third Base TA (3' end)
	U	A	C	G	
U	UUU	UAU	UCU	UGU	U
	UUA	UAA	UCA	UGA	A
	UUC	UUA	UCC	UGC	C
	UUG	UAG	UCG	UGG	G
	AUU	AAU	ACU	AGU	U
	AUA	AAA	ACA	AGA	A
	AUA	AAC	ACC	AGC	C
A	AUG	AAG	ACG	AGG	G
	CUU	CUA	CCU	CGU	U
	CUA	CAA	CCA	CGA	A
C	CUC	CAC	CCC	CCG	C
	CUG	CAG	CCG	CGG	G
	GUU	GAU	GUC	GUU	U
G	GUU	GAA	GCA	GUA	A
	GUG	GAC	GCC	GUC	C
	GUC	GAC	GCG	GGU	U
	GUG	GAG	GCG	GGG	G
	GUG	GAG	GCG	GGG	G

Genetic code (Triplet)