9. NATURE OF THE GENE

DNA or deoxyribose nucleic acid was first described by Friedrich Miescher in 1868. The DNA consists of four nucleotides. Each nucleotide is made up of a pentose sugar (deoxyribose type), a phosphate group and a nitrogenous base. A subunit of a nucleotide is the nucleoside. This is made up of only sugar and a nitrogen base. The four nucleosides differ from each other in the type of base which could be adenine (A), guanine (G), thymine (T) or cytosine (C). The Adenine and Guanine are purines while Thymine and cytosine are pyrimidines. In chemical terms, each nuceotide is a deoxy-5-monophosphate i.e., DNA is a polynucleotide.

Structure of DNA : In 1953, Maurice H.F. Wilkins and Rosalind E. Franklin suggested that the DNA is a long molecule consisting of two similar" strands running in parallel and in a helical manner. Watson and Crick proposed that the DNA is a double helix. That is, the DNA consists of two strands which are helically coiled. Each strand consists of a backbone made up of alternating deoxyribose sugar and phosphate. The phosphate joins the two deoxyribose sugars through a phosphouiester bond. (This is a phosphate group forming a bridge between two hydroxyl (i.e., - OH) groups of two adjacent sugars. The base pair of one strand are joined to the base pair of the opposite through hydrogen bonds. The pairing is always such that Adenine pairs with Thymine and Guanine with Cytosine. That is, the pairing is always between the purine and pyrimidine bases.

RNA and its Structure : RNA or ribonucleic acid is another nucleic acid and like DNA, is a polynucleotide. However in RNA. the pentose sugar is ribose but not deoxyribose. It contains Uracil but not Thymine **as** the pyrimidine. RNA serves **as** the genetic material in many viruses. The three major classes of cellular RNA are ribosomal RNA (rRNA), messenger RNA (mRNA) and transfer RNA (iRNA.). All these three moiecuies originate as complementary copies of one of the two strands of a DNA segment that constitutes **a** gene during the process of transcription. That is, each gene (a DNA strand) will throw up an identical R.NA sequence of the nucleotides except that uracil will replace thymine in the RNA sequence. Messenger RNA carries the genetic message from the DNA to the ribosome. Transfer RNA, carries amino acids to the ribosomes during translation. Transfer RNA contains many modified bases.

DNA Replication: According to Watson and Crick, each DNA strand of a helix acts as a template for the synthesis of a daughter strand. The resulting daughter strand will be complementary to the template. The copying of DNA. to make more DNA is called DNA replication. The daughter helix contains one old and one new strand. This mode of DNA replication is called semiconservative replication. In DNA replication, the first requirement is the separation of the two strands making up a DNA helix. The two separated strands can then act as templates for replication.

The most important DNA synthesising enzymne is DNA Polymerase-III. This along with the other DNA polymerases has the capability to elongate an existing DNA strand but cannot initiate the synthesis. To initiate DNA synthesis, a small . segment of RNA called the RNA primer, which is complementary to the template DNA is synthesised by a unique RNA polymerase called primase. To primase, the DNA p'olymerase-III adds the five deoxyribose nucleotides and extends the DNA. Since the two strands of DNA run anti-parallel to each other, DNA synthesis on both strands differs. While on one strand, the DNA synthesis continuous, on the other, the DNA is synthesised in small stretches of DNA, synthesized with the help of RNA primers. The RNA primers are them removed and the gap is then filled by DNA synthesis. That is, the stretches of DNA are joined by the enzyme Ligase. The process of replication ensures accuracy in order to maintain the nucleotide sequence of the original DNA.

Gene Expression: After replication, the second most important attribute of the gene is to store and express the genetic information that will contribute towards the phenotype which will be passed on to successive generations. The idea that genes control the metabolism was put forward by Garrod in 1902.

Genes and Proteins : A specific sequence of four bases in the DNA serves as the storehouse of all genetic information and hence the basis of all life an earth. The unique way the bases are arranged constitutes the genetic code. The genetic code determines "the basic structure and hence function of the whole variety of proteins. The expression of the genetic material generally occurs through the production of proteins. This involves the steps of transcription and translation, hi the process called transcription, the genetic information stored in the DNA is transferred to an RNA. The RNA uses this information to direct the manufacture of proteins during translation.

In 1970, H.M. Temin and D. Baltimore described a process called Reverse Transcription, hi this, the RNA replicates by synthesizing a complementary DNA. This is carried out by a RNA- dependent DNA polymerase called Reverse Transcriptase. Many tumour virus that

contain RNA as the genetic material replicate by synthesising a complementary DNA with the help of Reverse Transcriptase. These viruses are known as reteroviruses. Transcription therefore involves the production of a single-stranded RNA identical in sequence to one of the strands of DNA. Transcription is accomplished by an enzyme called RNA polymerase which gets physically associated with DNA.

Translation : This is a process by which the genetic message carried by mRNA from the DNA is concerted in the form of a polypeptide chain having a specific; sequence . of amino acids. In addition to mRNA, the other entities involved in translation are ribosomes, amino acids and tRNA's. Ribosomes are the ribonucleoprotein particles that provide the site for protein synthesis. It may be noted that the prokaryotic and eukaryotic ribosomes are different in terms of their proteins and also the RNA. In protein synthesis involved in the translation process, twenty naturally occurring amino acids participate. In translation, a tRNA is specifically linked to an amino acid, a process called charging. This is brought about by an enzyme (called aminoacyl tRNA synthetase) which recognises only one amino acid.

Molecular Basis of Mutation: In DNA, a specific sequence of bases forms the genetic code for a gene. Mutations in the genetic code can be by a) Substitution - in which one base is replaced by another base like for e.g., the substitution of purine to purine or pyrimidine to pyrimidiiie (a substitution called transition) and purine to pyrimidine or pyrimidin to purine substitution (called transversion). b) Frame shift Mutation - in which there is addition or deletion of a few bases. Since a specific base sequence within a gene is. crucial for it to express a particular phenotype, any change in the base sequence may change the codon and thus lead to mutation or altered expression. Base substitutions can also create mismatches. However, each living cdi possesses repair mechanisms to rectify such mistakes. Only when these mechanisms fail or are defective, that mutations take place.

Regulation of Gene Expression : We know that the genetic information stored in a gene is expressed through the process of protein synthesis. Studies have shown that while some proteins may be produced **in** five to,ten molecules, Others may be produced in as many as 1,00,000 copies per cell. This suggests that gene expression is regulated.

Inducible Control as a Method of Gene Expression: Francois Jacob and Jacques Monod suggested that a group of genes are expressed and regulated together as a unit. This unit was termed by them as the operon. The RNA produced by the operon is polycistronic mRNA

Repressibte control As a Method of Gene Expression: Jacob and Monod suggested that an operon can be repressed hence stopping all its transcriptional activities. This is not only a form of gene regulation but is also an example of negative control.

Gene expression is extensively regulated based on the environment. In multicellular organisms, regulations can take place at the tissue level. That is, different genes are expressed in different urgans. Hcwevei, some genes ore expressed in all cells. These are called house-keepmg genes.

GENETIC BASIS OF INHERITANCE

Genetics owes its development to Mendei's experiments on :the garden pea (_ Pisum Sattuum). Based on his experiments, Mendel postulanted four principles of inheritance. These are : .

1. Each genetic character is controlled by a .pair of alleles (the allomorphic pair),
2. When two dissimilar alleles are present in a single individual, only one can express itself, called the dominant gene and the one that fails to express itself is called the recessive gene.
3. When any individual produces gametes, the alleles seggregate and each gamete receives oniy one member of the pair of alleles. This is'called the Principle of Segregation or Purity of Gametes. It may be noted that the paired condition is restored by random fusion of gametes during fertilisation.
4. If two pairs of contrasting traits are inherited independently, then according to the Principles of Independent Assortment of Factors, when two independent events occur simultaneously, the combined probability of two outcomes is equal to the product of their individual probabilities of occurrence.

Genetic Terminology:

1. **Allele :** Each gene consists of an allele pair. The alleles represent two alternative forms of a gene.
2. **Phenotype :** The observable morphological appearance of an individual is called the phenotype. The phenotype of an individual is determined by different combinations of alleles.
3. **Genotype :** This is the genetic constitution of an individual representing a single character or a set of characters.
4. **Homozygous Alleles :** When the two alleles of a gene are similar, they are said to be in homozygous combination.
5. **Heterozygous Alleles :** When the two alleles in a gene are dissimilar, they are in heterozygous combination.

6. **Dominant Alleles :** An allele .that influences the appearance of the phenotype even in the presence of an alternative allele is a dominant allele.

7. **Recessive Allele:** An allele that influences the appearance of the phenotype only in the K presence of another identical allele is a recessive allele.

Developments in Gcnetics After Mendel : Three botanists viz Hugo devries, Carl Correns and # Erich Tshermak-Seysenegg carried out hybridisation experiments similar to those of Mendel. This led to the rediscovery of Mendel's principles and also led to the discovery of new principles in genetics. These new concepts are :

1 **Incomplete Dominance :** Ot the two alleles of a gene, one is dominant over the other and hence does not allow this recessive allele to express itself in the heterozygote. However, the recessive allele may sometimes express itself in a heterozygote. This leads to tb« development of a different phenotype. When the recessive allele also expresses itself in the heterozygote, it is called partial dominance or incomplete dominance. For e.g., in plants like Fcur-o-Clock (Mirabilis Jalapa) 'f red flowered plants are crossed with white flowered ones, the first generation offspring plants produce pink flowers (i.e., a combination of red and white).

2. **Codominance:** When both the alleles of a gene express themselves in the offspring, it is called codominance.

3. **Polygenic Traits :** When the phenotype shows no distinct alternative forms (like for e.g., either dark-skinned humans or white-skinned humans), it is a ease of polygenic traits, i.e., all the different variations of the phenotype can be produced. The polygenic traits are generally controlled by three or more genes where the phenotype reflects the contribution of each allele. Inheritance of polygenic traits reflects the principle of continuous variation of traits.

4. **Pleiotropy :** When one gene controls several phenotypes, it is called a pleiotropic gene and the phenomenon Is called pleiotropy.