

Concept of Genes, Split Genes, Exons, Introns and Complementation Test

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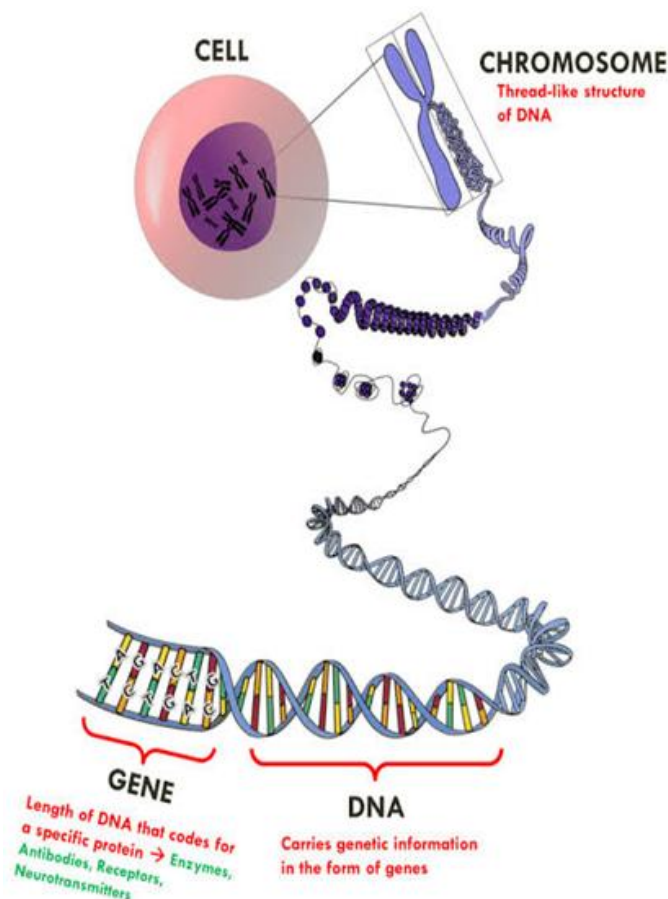
Abstract: *The heritable units which are transmitted from parents to offsprings are usually referred as Genes. So, a Gene is the fundamental biological command that has a specific role in transmission of traits or characters from one generation to the next. Due to the parallel behaviour of genes to that of chromosomes, as Genes are present on the chromosomes, everyone believed that chromosomes have definite role in inheritance, before the rediscovery of Mendel's laws. But after the discovery of DNA, it was accepted by everyone that DNA acts as a carrier of genetic information. Generally, If we need a trait to be expressed phenotypically, then the gene which is responsible for that particular trait has to express at the molecular level. So one has to understand the physical and chemical nature of the gene to understand how the traits or characters expressing phenotypically. Usually, a gene has a continuous nucleotide sequence without any interruption, which codes for an amino acid or a protein but there are some other type of genes in which the nucleotide sequence is not continuous and interrupted by non-coding sequences. So the genes which have both coding and non-coding sequences are referred as Split genes. Coding sequences are called Exons and non-coding sequences called as Introns, which are normally spliced in processing of mRNA, in a split gene. Production of a normal individual from a cross between two homozygous which are effected by same defect is referred as complementation and the test that is used to determine whether the mutation is occurred in the same gene or different genes is called as Complementation Test. Here, I am giving you a brief informative note on fine structure of gene, split genes, exons, Introns and Complementation Test.*

Keywords: Gene, Split genes, Exons, Introns, Complementation

1. History and Fine Structure of Gene

Gene refers to the basic hereditary factors/units that are usually transmitted from one generation to the next generation, which determines the expression of either parent characters in the offspring. Otherwise, it is also described as the fundamental biological command which links one

generation to the next. Mendel assumed these genes as particulate units and referred as hereditary units or factors, while concluding his results of monohybrid and dihybrid crosses. This theory was completely hypothetical and Mendel did not consider the physical and chemical nature of the gene.



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Various theories were put forth to explain the physical and chemical nature of genes and some of them were 'one gene one character' by De Vries, Specific character of an individual is determined by a specific gene, and 'presence or absence' theory was given by Bateson and Punnett, trait in a cross dominates the other trait has a dominator which is absent in the recessive trait. In 1926, Morgan gave 'particulate gene theory', in which genes arranged as beads on a string (chromosome). After the discovery of DNA, as genetic carrier material, all theories were discarded.

How a Gene is defined (Classical)

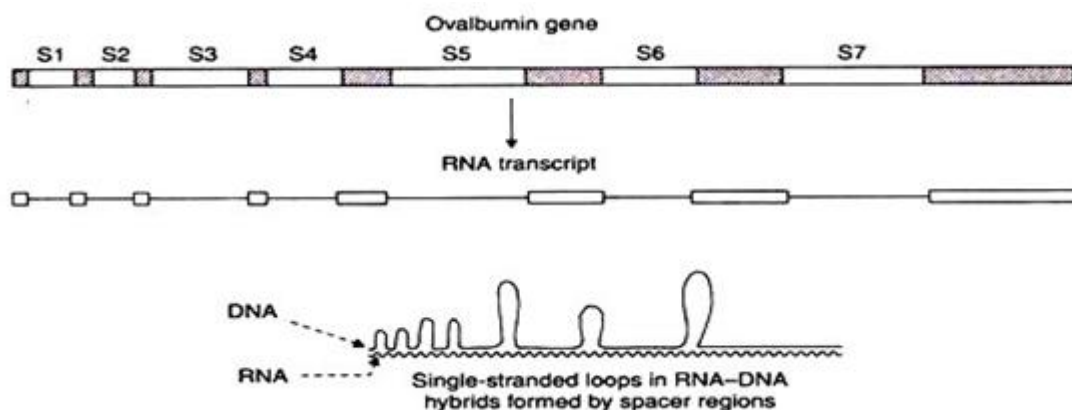
- Unit of fundamental **physiological function** that has definite locus in the chromosome and plays a specific role in expressing specific trait, like, Tall or Dwarf plant and green or yellow pods in Pea.
- Unit of segregation**, as it gets divided and switched by crossing over during meiosis.
- Unit of mutation**, as it is responsible for dissimilar phenotypic expression, on spontaneous or induced change.

How a Gene is defined (Modern)

Parallel behavior of gene with that of the chromosomes and studying underlying physical and molecular nature of gene in expressing a trait phenotypically, gene is defined as:

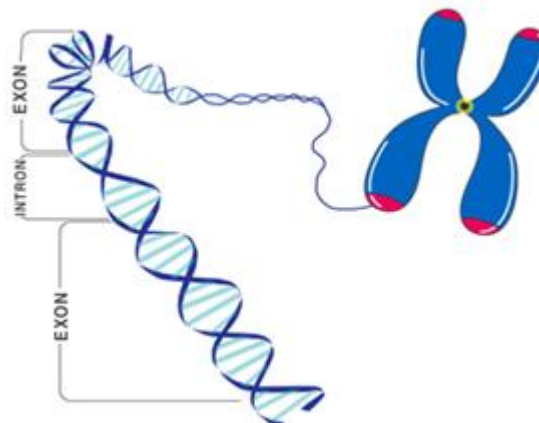
- Cistron**: Specifies a single polypeptide chain, liketerm physiological function. Term was coined by Benzer. For example, Hemoglobin requires two cistron fractions of globin protein one each for α and β chains.
- Muton**: In a cistron, there are many loci or sites where mutations can happen. Therefore, gene as unit of mutation is referred as Muton and it is the smallest length of DNA where mutational changes can happen. Term was coined by Benzer.
- Recon**: In a cistron, during meiosis recombination occurs by crossing over. Therefore, the smallest unit of DNA, where recombination can occur is referred as recon.

Split Genes



Split genes were first time reported for the gene ovalbumin of chickens, as this gene has seven interrupted genes (non - coding sequences) and half of the ovalbumin gene is excised during mRNA processing. Later reported in tRNA genes of yeast, β - globin genes of rabbits and ribosomal genes in *Drosophila*.

In terms of nucleotide sequences or fractions of polypeptide chain some genes are different from normal genes. Normally a gene consists of a continuous nucleotide sequence and those fractions or sequences code for specific protein. But, in some genes the nucleotide fractions or sequences are discontinuous and interrupted by non - coding sequences. Therefore, genes with both coding and non - coding sequences are called as Split genes or intervened genes.



Split Gene

Let us, know coding and non - coding sequences:

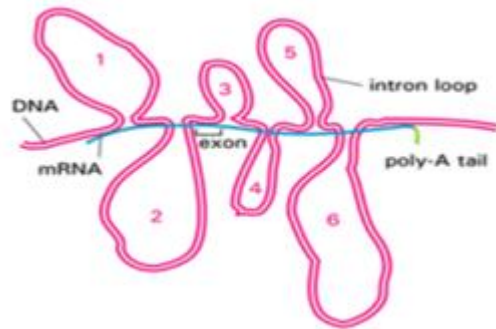
- Coding Sequence**: In a split gene, nucleotide sequences, which are capable of coding an amino acid or a protein are called as coding sequences. These coding sequences are referred as **Exons**.
- Non - Coding Sequence**: In a split gene, nucleotide sequences, which are not capable of coding an amino acid or a protein are called as non - Coding sequences. These non - coding sequences are referred as **Introns**.

During the processing of mRNA, non - coding sequences are removed as they are interrupted sequences, do not code for any protein, in a split gene. The exons are alone joined by enzyme ligase.

Determination of Non - Coding sequences

R loop technique is best employed for the determination of interrupted fractions or sequences. It involves hybridization of mRNA and DNA of the same gene, provided with ideal condition (high conc. of formide and high temperature). During the process mRNA pairs to single strand of DNA and the non - coding fractions are looped in pairing. Numbers of

interrupted sequences are determined by the number of loops formed while pairing.

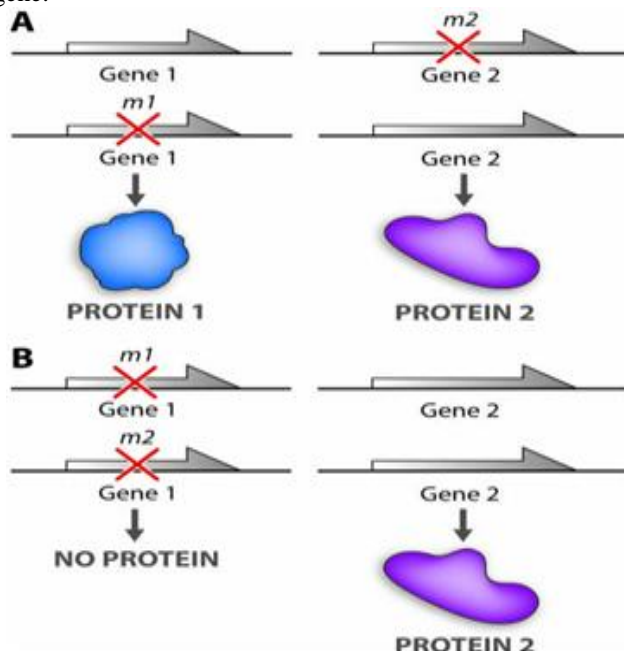


Loop formation in mRNA and DNA Duplex

Complementation Test

Let us understand the concept of complementation test with an example. Consider two homologous chromosomes carrying mutations in different genes. Say, one chromosome has mutation in gene p and other in gene Q. Then, during synthesis one chromosome produce a mutant p and normal Q gene. Other chromosome synthesis a normal p and a mutant Q gene. Therefore, we can say that the cell contains normal p and Q genes. In this phenomenon, two mutant chromosomes complement each other and functions normal. So, it is said to be the two mutations lie within the loci of different genes.

In other case, let - two chromosomes carry mutation on the same gene Q, neither chromosome will produce normal Q gene and thus, normal Q function will not be observed. So, it said to be the two mutant chromosomes do not complement each other and referred that both mutants lie in the same gene.



Complementation Test

Therefore, complementation test is done to find out whether the mutation is occurred in the same gene or in different genes.

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