CSIR NET Life Science Unit 8

Types & Causes of Mutations

- A mutation is a rapid, stable, discontinuous, and inheritable alteration that occurs in an organism as a result of a permanent change in its genotype. Mutant is the name given to the result of a mutation, which might be a genotype, a cell, a polypeptide chain, or a human. Mutations are the source of all variants since they are responsible for the emergence of new features in the race.

- Mutations can arise in a variety of cells, but most of them have little impact on individuals and go unnoticed most of the time. Somatic and germinal cells are both susceptible to mutations. Somatic mutations result in mutated buds, while germinal mutations are valuable because they can be transmitted through grafting.

- Germ mutations are crucial in terms of heredity since they are handed down to future generations. Individual gene mutation rates are quite low, but since there are so many genes, the possibility of a mutation in one or more of them is very high.

- Mutations have a variety of impacts, including detrimental, deadly, beneficial, and neutral, as well as apparent and unseen effects. The majority of mutations occur in recessive genes that are not expressed, whereas others occur in dominant genes that result in mutants.

- Mutations have been occurring in nature also and are called spontaneous mutations. When numerous physical and chemical agents are used to increase the frequency of mutations, they are called induced mutations.

Spontaneous Mutations

- Spontaneous denotes that the occurrence resulted in an unknown mutation. As a result, mutations that develop in the absence of a mutagen are called spontaneous mutations. When it comes to genetic phenomena, mutations with clearly defined effects are commonly employed, however, the majority of mutations do not fit into this category.

- The type of cell that contains a mutant allele determines the effect of the mutations. When two heterozygotes mate, a recessive mutation is usually not found until a later generation. In bacteria and haploid eukaryotes, dominance has little effect on mutation expression.

- If a dominant mutation occurs in a parent, it will be visible in any offspring who inherit it. Because most mutations are recessive in origin and do not occur on the X-chromosome, they are referred to as autosomal recessive.
- Germinal mutations occur in cells that will eventually create gametes, while somatic mutations occur in all other cells. Mutations that can be turned on and off at will are some of the most beneficial for genetic study. Conditional mutations cause phenotypic differences under one set of environmental variables known as permissive conditions.

Character	**Somatic Mutations**	**Germinal Mutations**
Type of cells involved	Somatic Cells	Germ cells
Inheritance	Inheritable	Heritable
Examples	Bud mutation; White scars in the red eye of drosophila	Short-legged ancon sheep

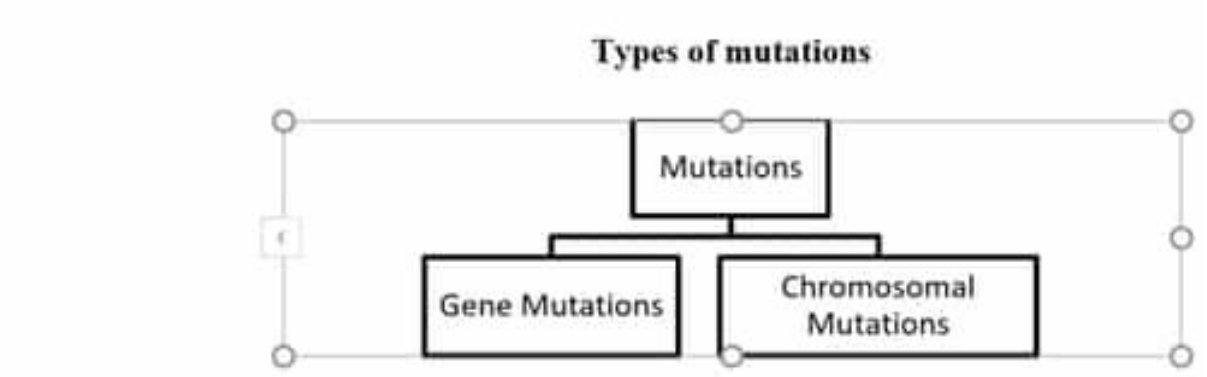
- Mutations are statistically random events, and no one can anticipate when or which cells will undergo mutations. However, because every gene mutates at a predictable rate, it is possible to assign a probability to specific mutational events. Background mutations are commonly referred to as spontaneous mutations because their origin is unknown.
- There is a certain probability that a given mutation will occur in a specific cell and a certain probability that a mutant allele of a gene would exist in a population of a certain size.
- Although the specific cause of spontaneous mutations is unknown, some experts believe that they may be caused by radiations prevalent in the atmosphere. Nitrogen bases can take on tautomeric forms on rare occasions. To form uracil residues, cytosine residues may undergo very slow spontaneous deamination.

- In the presence of defective DNA polymerase, E.coli produces a mutation rate one thousand times the normal. Sometimes spontaneous mutation may occur due to defective proofreading and repairing machinery of DNA.

Induced mutations

Mutations can be caused by a variety of factors, including ultraviolet or ionising radiation, as well as chemical mutagens. Over 2,000 crop varieties have been produced since the 1950s by inducing mutations to change genetic features at random and then selecting improved kinds among the descendants.

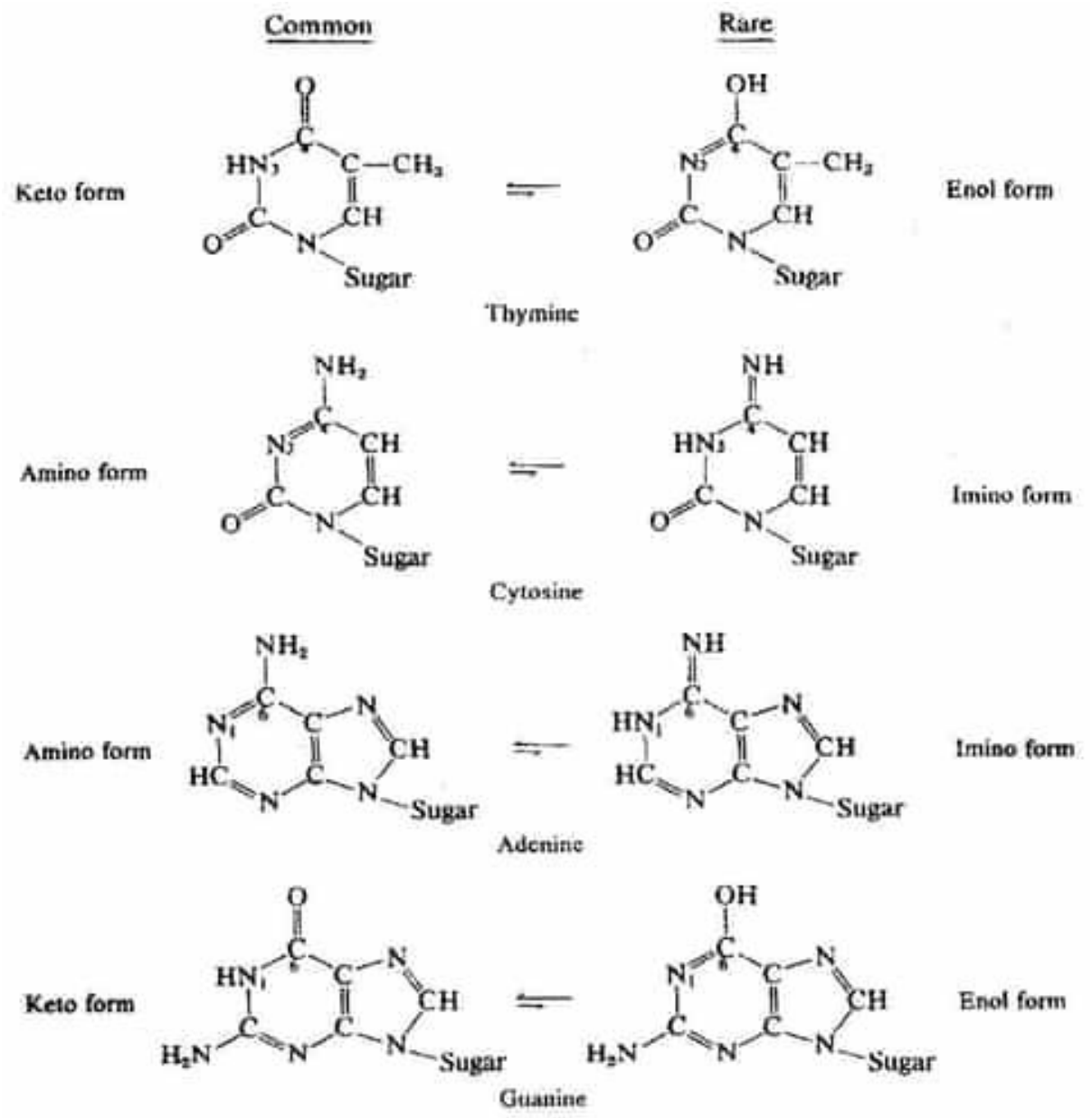
Types of mutations



Gene mutations are permanent alterations to a gene's DNA chain. Despite the fact that each gene is a potential mutation site, some genes mutate more frequently than others. Mutable genes can be discovered in both plants and mammals. Gene mutations arise naturally in bacteria at a rate of around 1 in 106 gene duplications. A muton, or the smallest component of a gene, can cause a point or gene mutation. Tautomerism and substitution are two types of modifications.

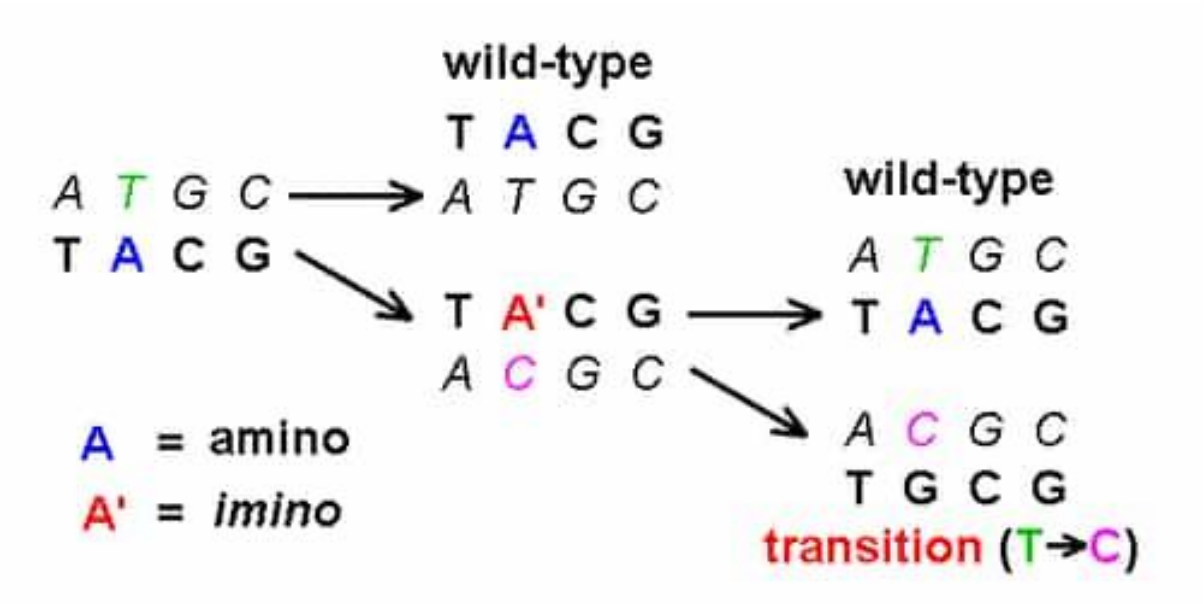
Tautomerism: Tautomerism is a phenomenon in which a single chemical molecule exists in two or more interconvertible forms with differing relative positions of one atomic nucleus, which is usually hydrogen. The two structures are known as tautomers, and the only difference between them is the number of electrons and protons. They're both in a state of dynamic equilibrium. When these chemicals react, the only thing that happens is that protons are transferred. Desmotropism is another name for tautomerism.

Tautomers include ketone-enol, enamine-imine, lactam-lactim, and others. In this process, a hydrogen atom is exchanged between two other atoms while creating a covalent connection with one of them. Tautomerism is a process that can be reversed

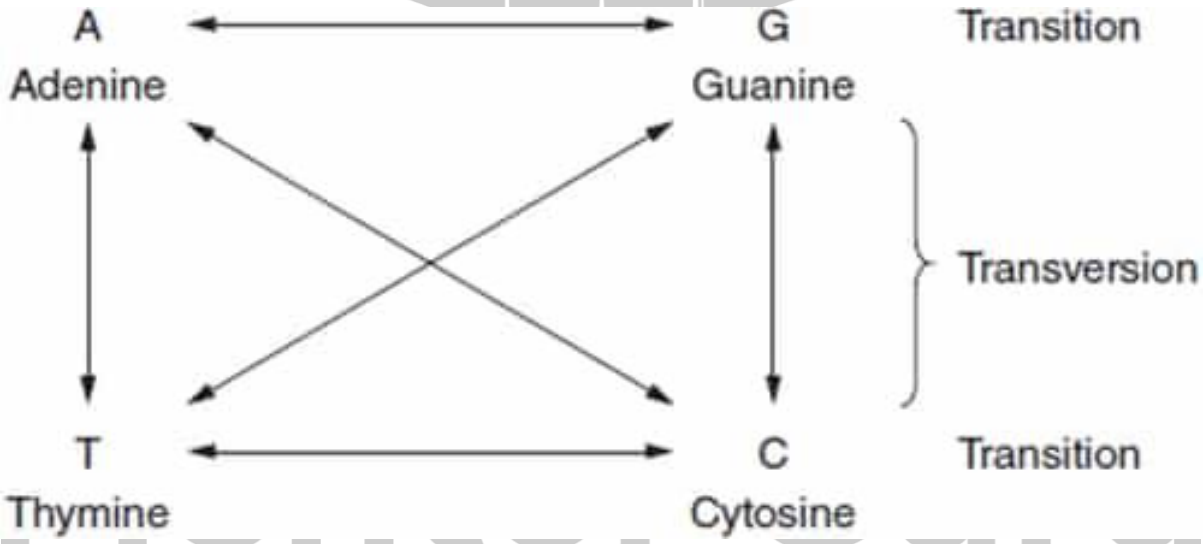


- **Substitution mutations** are those in which one or more nitrogenous base pairs are replaced with others in a gene. Transition, transversion, and frameshift are three possible subtypes.

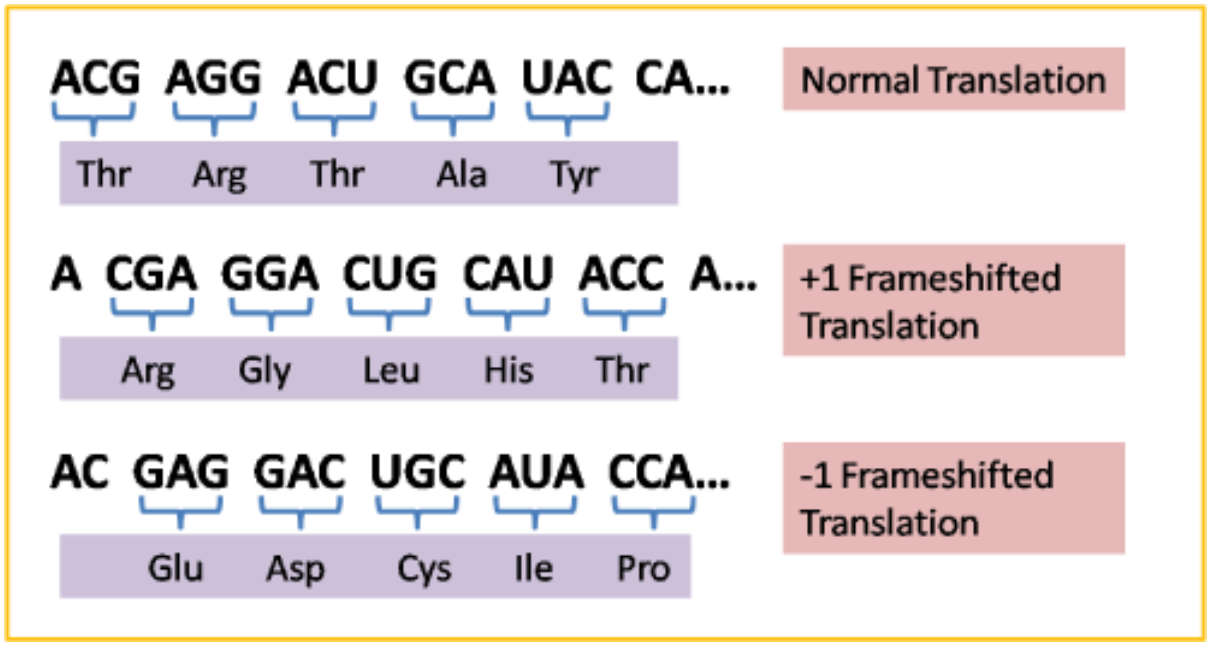
(a) Transition: There are two types of DNA substitution mutations. Transitions are interchanges of two-ring purines (A G) or one-ring pyrimidines (C T), and so involve similar-shaped bases.



(b) Transversions entail the substitution of purine for pyrimidine bases, resulting in the exchange of one-ring and two-ring structures.



(c) Frameshift mutation is a genetic mutation that occurs when a deletion or insertion in a DNA sequence causes the sequence to be read in a different way. A DNA sequence is made up of a series of smaller molecules known as nucleotides. Codons are three-nucleotide units that correspond to a certain amino acid or stop signal. DNA (or RNA) nucleotide sequences are read three nucleotides at a time. The codon sequence is read in order from the nucleotide sequence during translation to build a chain of amino acids and construct a protein. Frameshift mutations occur when one or more nucleotides are added or removed from the usual codon sequence, provided that the number of nucleotides added or removed is not a multiple of three.


ACG AGG ACU GCA UAC CA...
Thr Arg Thr Ala Tyr
Normal Translation
A CGA GGA CUG CAU ACC A...
Arg Gly Leu His Thr
+1 Frameshifted Translation
AC GAG GAC UGC AUA CCA...
Glu Asp Cys Ile Pro
-1 Frameshifted Translation