

Ans. - # Introduction :

Def - Gene mutation is abrupt, inheritable and quantitative as well as qualitative change in genetic material of an organism. It is change in DNA sequence which causes change in sequence of RNA and protein consequently.

• Term mutation was proposed by - Hugo de Vries in Peanut (Pisum sativum)

Bateson given term - Discontinuous variation for it
Darwin " " - Sports.

• Seth Wright first observed mutation in Ancon sheep (lamb with exceptionally short legs). It was a type of homozygous recessive mutation.

Kinds of mutation :

→ According to type of cell.

(1) Somatic mutation -

- If mutation takes place in somatic cells.
- Effect of somatic mutation will not be transferred to next generation and it will be atrophied with death of organism.

* In case of plants, it can be inherited by help of vegetative propagation.

(2) Germinal / Gametic mutation -

- Mutation in germinal cell.
- Effect of this mutation transferred to next generation.

→ According to expression.

(1) Dominant -

Recessive is mutated to form dominant gene.

• Its effect is expressed immediately.

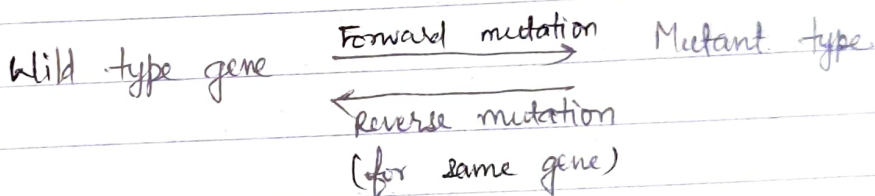
(2) Recessive -

- Dominant gene is mutated to form recessive gene.
- Effect of recessive mutation will not be expressed unless it is present in homozygous condition.
- * Most of recessive mutations are deleterious (harmful).

→ According to expression.

(1) Forward mutation -

Mutation in wild type gene.



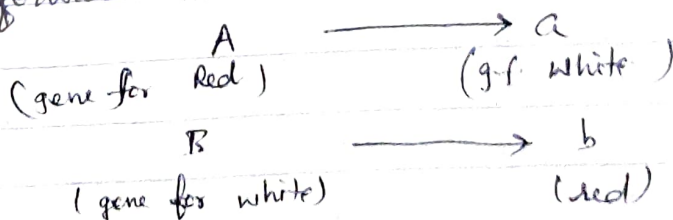
(2) Reverse mutation -

Mutant type, if again mutated in wild type, due to mutation in same gene.

* Frequency of reverse mutation is very less in compare to forward mutation.

(3) Suppression -

Mutation in different gene which suppresses effect of forward mutation.



Here mutation in gene B directly suppresses the

frequency of mutation of gene A by giving red colour.

→ According to size and quality.

(1) Point mutation.

(2) Multiple (gross) mutation.

(1) Point mutation.

• When heritable alteration occurs in very small segment of DNA i.e. in single ~~codon~~ or a pair of nucleotides.

• It is of two types.

(1) Substitution mutation -

- Due to substitution of nitrogenous bases in triplet.

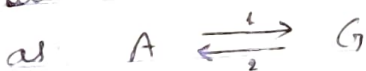
- No. of bases is not $\uparrow$ ed or $\downarrow$ ed and only a triplet codon is affected by this mutation.

Substitution mutation



Transition

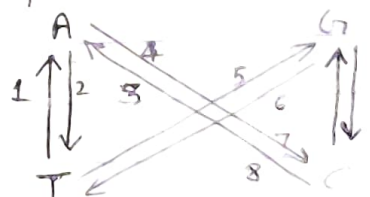
- If a purine is substituted by other purine base or a pyrimidine base is substituted by other pyrimidine base.



Four types of transition mutations are possible.

Transversion

- If purine base is substituted by pyrimidine base or a pyrimidine base is substituted by a purine base.



8 types of transversion mutations are possible.

(2) Frame shift mutation (Gibberish mutation)

- It is due to either increase or decrease in no. of bases
- Deletion or addition of bases causes shift of coding frame.

e.g. deletion

5' ... AUG ACA AGA GUA AAC ACG UUU

Then

5' ... AUG AAA GAG UGA GCA GGC UG ...

addition:

5' ... AUG GAC CAU ACG GGC CAU ... 3'

A added

then

5' ... AUH GAA CCA UAC GCC CCA G ... 3'

② Multiple mutation / Gross mutation

- When changes involve more than one nitrogenous base pair or entire gene, then it is multiple or gross mutation.
- These mutations occur due to rearrangement of genes within genome. And may be of following types.

(1) Two mutation in same functional gene can produce same phenotypic effect depending upon cis - or trans - position of gene.

(2) Rearrangement of no. of gene per chromosome.

✓

(3.) Due to movement of gene locus, new phenotypes are produced.

are produced. Movement of locus may be due to translocation (movement to non-homologous chromosome) or inversion (movement of gene in same chromosome).

→ Mis-sense mutation: Here polypeptide with different sense will be synthesized.

→ Same-sense mutation : Even after mutation, sense of polypeptide synthesis will not be changed.

→ Non-sense mutation: If due to mutation, a non-sense codon is produced in middle of coding frame.

e. g

9
5' AUG GAG CAU UGG AUG CUC ... 3' mRNA
 ↑
 replaced by A

Then,

5' - AUG GAG CAU UGA AUG CUG - 3' mRNA
 ↳ non sense codon formed

This will cause premature termination of polypeptide synthesis.

→ classification of mutation a/t origin.

- (1) Spontaneous mutation.
- (2) Induced mutation.

(1) Spontaneous mutation

These occur suddenly in nature and their origin is unknown. These are also called background mutation. These occur in *Penicillium*, bread mould, *Drosophila*, microorganisms etc.

(2) Induced mutation

These mutations are induced artificially in living organisms by exposing them to radiation or chemicals.

Mechanism of mutation