Mutation and Polyploidy

1. What is mutation? Describe its types and significances. (8 marks)

-Def^{n}: Sudden appearance of marked heritable variation in the nature of any organism due to change in their genotype.

Types:

i. Gene mutation.

ii. Chromosomal mutation.

i. Gene mutation:

-Any change that alters the nature of gene at its molecular level.

Types of gene mutation:

a. Frameshift mutation.

b. Substitution mutation.

c. Nonsense mutation.

a. Frameshift mutation:

- Caused by the addition or deletion of one or more nitrogen base in the sequence.

Eg: A C G A G G A C U....

Delete A

C G A G G A C U A....

b. Substitution mutation:

- Replacement of nitrogen bases by another type.

Types of substitution mutation:

a. Transition:

- Mutation arised due to substitution of purine bases by purine or pyrimidine bases by pyrimidine.

Eg: i.e. Purine (A, G), Pyrimidine (C, T)

C G T

C A T

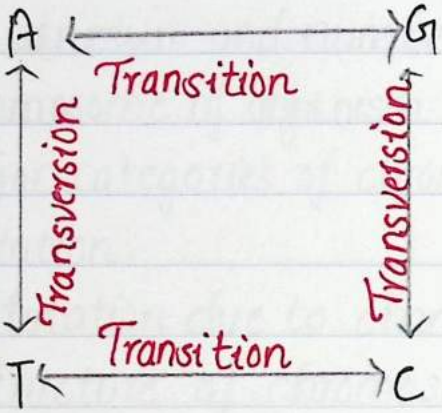
b. Transversion:

- Mutation arises when a purine base is replaced by pyrimidine base and a pyrimidine is replaced by purine base.

Eg:

A G C T

C G C T



c. Nonsense mutation:

-Arises due to change in one nitrogeneous base of sense codon. i.e. sense codon converted into non-sense codon.

Eg: CGA ⟶ UGA

UAC ⟶ UAA

UAT ⟶ UGA

ii. Chromosomal mutation:

-Sudden heritable variation in

the structure and number of chromosome of organism.

Major categories of chromosomal mutation:

A) Mutation due to change in structure of chromosome.

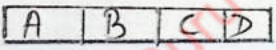
B) Mutation due to change in chromosomal number.

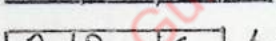
A) Mutation due to change in structure of chromosome:

Types:

a. Deletion / Deficiency
b. Duplication
c. Inversion
d. Translocation

a. Deletion / Deficiency:

- A segment of chromosome breaks off and gets lost.
- Large deficiency is detected during synapsis (Zygotene).
- Involves a break of a piece of chromosome but not reattached. 

- 2 Types: 

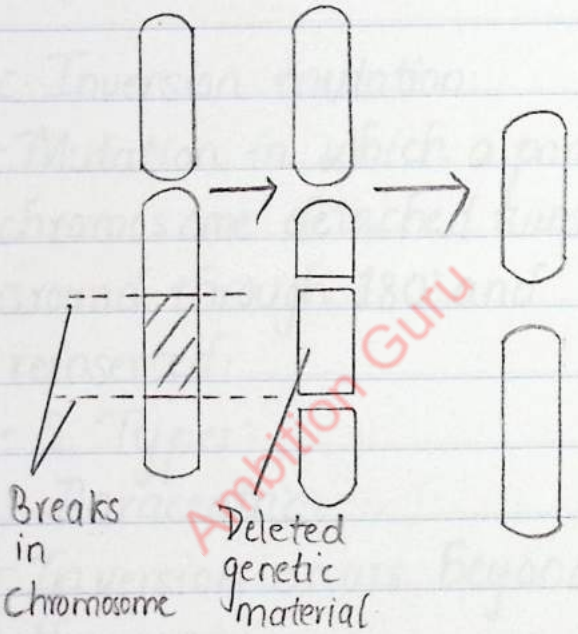
1. Terminal deficiency:

- Occurs due to single break in chromosome.
- Terminal piece of chromosome lost.
- Terminal loss is deficiency.

2. Interstitial deletion:

- Occurs when chromosome breaks

through two points.
- Middle piece is lost and remaining parts unite again.



Eg: ABCDE.FGH ⟶ ABCE.FGH

b. Duplication mutation:
- Mutation occurs when one or more gene sequence is repeated.

- Also known as gene amplification.

Eg:

ABCD.EF ⟶ ABCD.EFEF

c. Inversion mutation:

- Mutation in which a part of chromosome detached turns around through 180° and reinserted.
- 2 Types:

1. Paracentric:

- Inversion occurs beyond the centromere.

Eg:

ABCD.EFG ⟶ ABCD.EGF

2. Pericentric:

- Inversion including centromere.

Eg:
ABCDE.FGH ⟶ ABCF.EDGH

d. Translocation:
- Mutation which occurs due to shifting of a segment of one chromosome to another non-homologous chromosome.
- 2 Types:

1. Simple Translocation:
- One-way transfer of genes from one chromosome to another non-homologous chromosome.

Eg:
Before: ABCDE.FGH MNOP.QRS

After: ABCDE.F MNOP.GHQRS

2. Reciprocal Translocation:
- Mutal exchange of segment between two non-homologous chromosome.

Eg:
Before:
ABCD. EFG HIJK. LMN
After:
ABJK. EFG HICD. LMN

B. Mutation due to change in Chromosomal Number.

Types:
a. Aneuploidy
b. Euploidy

a. Aneuploidy:
- Change in chromosome number

either due to addition or deletion of one or more chromosome.
- Condition arises due to abnormal number of chromosome in gamete and zygote.

Types:

i. Monosomic (2n-1):
- Mutation which occurs due to loss of single chromosome from diploid set.
- Represented by 2n-1.
where, n = haploid number of chromosome.
Eg: Turner's syndrome (XO)

ii. Double monosomy (2n-1-1):
- Type of mutation where two missing chromosomes belong to

different pair (non-homologous).
- Represented by (2n-1-1)

iii. Nullisomic (2n-2):
- Arises due to loss of one pair of homologous chromosome from complete set.
- Found in wheat, etc.
- Represented by (2n-2).

iv. Polysomic:
- Addition of one or more chromosome.

a. Trisomic (2n+1):
- An organism having two complete genomes plus one extra chromosome.

- Result of addition of a chromosome in diploid set.
- Represented by (2n+1).

Eg: Trisomy 21 (Down syndrome)
⇒ 1 extra additional, 21st chromosome.

Double trisomy 18th trisomy (Edward's syndrome), 13th trisomy (Patau's syndrome), etc.

5. Tetrasomic (2n+2):

- An organism having two complete set of genomes plus one pair of homologous chromosome.
- Represented by (2n+2).

b. Euploidy:
- Condition of addition or loss of complete set of chromosom
- Chromosome number is exact multiple of haploid set.

Types:

i. Monoploidy/haploidy:
- One set of chromosome present.
- Eg: Ant, wasps, bacteria, few fungus and bryophytes.

ii. Polyploidy:
- Condition of addition of one or more set of chromosome in diploid organism.
- May be triploid, tetra ploids,

pentaploids, hexaploids and so on.
- Eg: flatworms, earthworms, etc.

Significance of mutation:

i) Mutations may improve an organism's survival.
ii) In agriculture, increases yielding and quality of yield products.
iii) Increase the yield of milk, egg, wool or fur in animal.
iv) In micro organisms, increases rate of fermentation, antibiotics production or drug development.

2. What is polyploidy? Describe its types with suitable example. (8 marks)

- Def^{n}: Condition of addition of one or more set of chromosome in diploid set of chromosome.
- Common in plants but rare in animals.
- For eg: Triticum aestivum, Nicotiana tobacum, Solanum tuberosum, etc.

Types:

i. Autoploidy.
ii. Alloploidy.

i. Autoploidy:
- Polyploids which originate by the multiplication of chromosome of the single species.
- Also known as autoplyploidy.
- About half of all polyploids are thought to be the result of autopolyploidy.

ii. Alloploidy:
- Polyploids which originate by combining complete chromosome sets from two or more species.
- Also this condition is known as allopolyploidy.
- Offsprings produced by such

unions are typically sterile due to uneven chromosome pairing.

- Example:

Raphano brassica

Parents: Raphanus sativum (Radish) $2n = 18$ × Brassica oleracea (Cabbage) $2n = 18$

Gametes: AA × BB

AB F1 hybrid

$2n = 18$

↓

Raphano brassica (Colchicine)

$2n = 36$ (AA BB)

Explain:

- A Russian geneticist G.D Karpenchenka produced Raphanobrassica experimentally.
- Made cross between radish and cabbage.
- Wanted to produce a plant that had cabbage leaves and Radish roots.
- Both radish and cabbage possess equal chromosome so, he was able to successfully cross them producing a hybrid with $2n = 8$.
- But, unfortunately hybrid was sterile.
- After that, treated with Colchicine and obtain fertile Raphanobrassica.