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Elmer

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Bsc part 2nd paper VI, Unit - 2 (3).

Question :- Write Notes on Gene Mutation

Answer :- Origin of Gene Mutation :-

A. Spontaneous mutation - mutation occurs during normal cellular activities, primarily DNA replication and repair.

B. Induced Mutation - mutation occurs as a result of treatment with a mutagenic agent or environment; mutation rate is usually higher than background levels.

i. Ionizing radiation - α -, β -, γ - or X-rays; usually results in deletions or insertions of DNA.

ii. Non-Ionizing radiation - UV light; causes adjacent thymines on one DNA strand to bond together (thymine dimers) resulting in a structure that must be repaired in

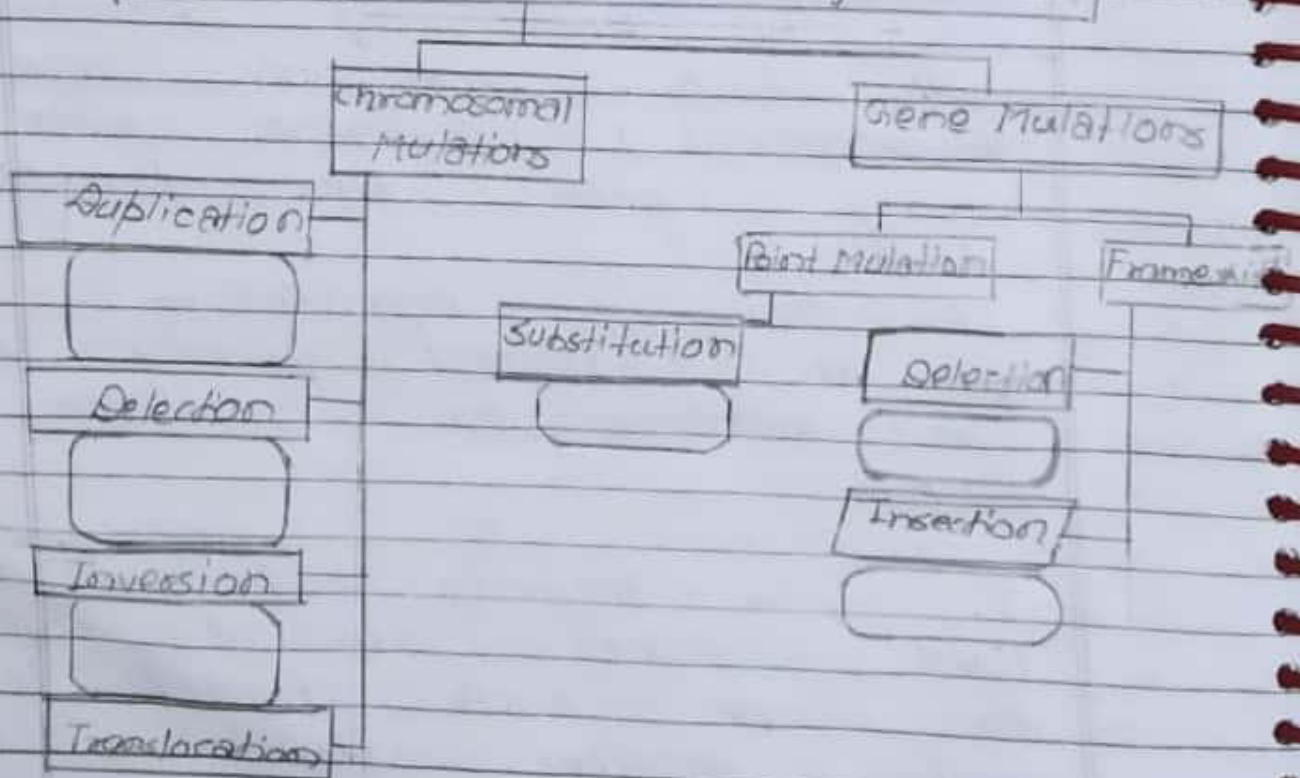
a structure that must be repaired in order for DNA replication to proceed; inefficient repair can lead to point mutations.

(iii) chemicals — chemical substances that interact with DNA to create base changes.

~~SYNOPSIS~~

Gene mutation

Types of mutation (definitions)



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Effects on Gene Mutation :

(i) Effect on protein (codons) :-

A. Silent Mutation :- change in a codon (usually in the third position) that does not change the amino acid coded for.

B. Nonsense mutation :- change in a codon from amino acid specificity to a stop codon; results in premature amino acid chain termination during translation.

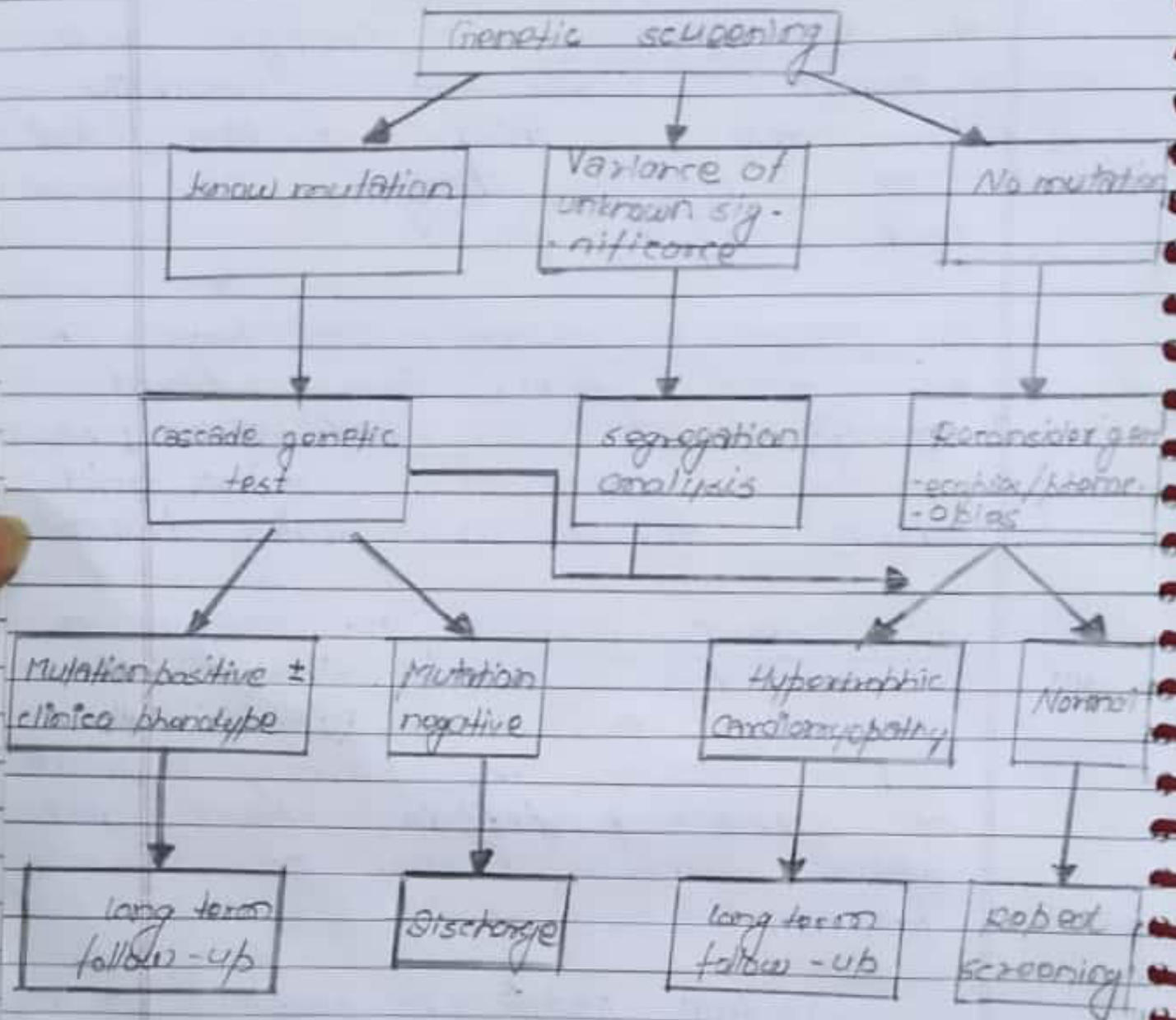
C. Missense mutation :- change in a codon that changes the specificity to a different amino acid; changes the primary sequence of the polypeptide chain and alters the function of the protein.

D. Neutral Mutation :- change in the codon such that a different amino acid is specified however the new amino acid (is specified) behaves similarly to the original one (e.g., has a similar functional group) and does not alter the function of the protein.

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Gene mutations: Understanding the significance using *in silico*.



Guidelines for genetic screening in families at risk for hypertrophic cardiomyopathy.

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Types of Gene Mutation :-

(i) Morphological Mutation :- This involves changes in morphology including colour, shape, size, etc., e.g., albino ascospores in *Neurospora*, kernel colour in corn, curly wings in *Drosophila*, and dwarfism in pea.

(ii) Lethal Mutation :- This involves genotypic changes leading to death of an individual. Example includes albino mutation resulting from chlorophyll deficiency in plants.

(iii) Biochemical Mutation :- Biochemical mutations are identified by a deficiency, so that the defect can be overcome by supplying the nutrient or any other chemical compound, for which the mutant is deficient. Such mutation has been studied in bacteria and fungi, as well as blood disorders in human.