

ANSWER THE FOLLOWING QUESTIONS

1- Choose the correct phrase:-

(54 marks)

a) Point mutation involves

(Deletion – insertion – duplication – change in single pair)

b) When an individual has only one of a particular type of chromosome it's described as

(monosomy - disomy - trisomy - tetrasomy)

c) Transition type of gene mutation is caused when

- GC is replaced by TA
- CG is replaced by GC
- AT is replaced by CG
- AT is replaced by GC

d) chromosomes have arms of unequal length with the centromere located near one end of the chromosome.

(Submetacentric - Acrocentric - Telocentric - Metacentric)

e) "Polyploid" is a term that refers to

- having multiple sex cell available for fertilization.
- having 2 sets (2N) of chromosomes.
- having 3 or more sets of chromosomes (3N, 4N, etc)
- having many expressions of the same gene.
- None of these

f) A point mutation that changes a codon specifying an amino acid into a stop codon is called mutation.

(missense - nonsense - frameshift - deletion)

g) Under the electron microscope, unfolded chromatin resembles "beads on a string." What do the "beads" represent?

- nucleosomes
- ribosomes
- beadosomes
- molecules of DNA polymerase
- molecules of RNA polymerase

h) A display of the pairs of chromosomes is called a

(pedigree chart - karyotype - caricature - chromosome map)

i) Which refers to the loss of a complete chromosome? (inversion - translocation - deletion - duplication - monosomy)

j) A trisomy occurs when an individual has three of

- any type of chromosomes
- each kind of chromosomes
- any type of autosomes
- sex chromosomes

k) A mutation in a codon leads to the substitution of one amino acid with another is called mutation.

(Nonsense - missense - frame shift - promoter - operator)

 I) Which refers to the addition of an extra segment of a chromosome? (inversion - translocation - deletion - duplication - monosomy)

<u>2-Write short notes on each of the following:</u> (36 marks)

- Mutagenic effect of ethyl methanesulphonate (EMS)
- Ames test
- Function of the secondary constriction
- Replicative segregation
- Frame shift mutation
- Tautomerization

3- Explain maternal inheritance of leaf-color phenotypes in *Mirabilis jalapa* (four O'clock) variegated plants. (4 marks)

4- Compare between:

(12 marks)

- Autopolyploidy and allopolyploidy.
- Transition and transversion mutations.

5) Which type of organisms is more affected by mutagens: prokaryotes or eukaryotes? Why? (6 marks)

6) What is the name of the process which occurs when a segment of chromosome is turned around 180°? (6 marks)

With my best wishes

Tr. Mahmoud Abderraouf



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<u>2-Write short notes on each of the following:</u> (36 marks)

 Mutagenic effect of ethyl methanesulphonate (EMS)
 <u>Ethyl methanesulphonate (EMS) is an alkylating agents, i.e. a</u> chemical which add an alkyl group (CnH2n+1) to another molecule. Alkylation of a base may change the normal base pairing. So, EMS converts guanine to 7-ethylguanine which pairs with thymine. The mispairing will lead to mutation. • Ames test

The Ames test is a bacterial reverse mutation assay specifically designed to detect a wide range of chemical substances that can produce gene mutations. The Ames test uses amino aciddependent strains of *Salmonella typhimurium*. In the absence of an external histidine source, cells cannot grow and form colonies. Only those bacteria that revert to histidine independence (his+) are able to form colonies. The number of spontaneously induced revertant colonies per plate is relatively constant. However, when a mutagen is added to the plate, the number of revertant colonies per plate is increased, usually in a dose-related manner.

The bacteria are spread on an agar medium plate with rat liver extract as a metabolic activation system and small amount of histidine. This small amount of histidine in the growth medium allows the bacteria to grow for an initial time and have the opportunity to mutate. When the histidine is depleted only bacteria that have mutated to gain the ability to produce its own histidine will survive.

- Function of the secondary constriction <u>Formation of the nucleolus, hence the secondary constriction is</u> <u>usually known as the "nucleolus organizer" or "nucleolus</u> <u>organizer region" (NOR).</u>
- Replicative segregation <u>Heteroplasmic alleles can shift in percentage during both</u> <u>mitotic and meiotic cell division, leading to a potentially</u> <u>continuous array of bioenergetic defects, a process known as</u> <u>replicative segregation.</u>
- Frame shift mutation
 Frameshift mutations arise when the normal sequence of codons is disrupted by the insertion or deletion of one or more nucleotides, provided that the number of nucleotides added or removed is not a multiple of three. This can result in the incorporation of many incorrect amino acids into the protein. In contrast, if three nucleotides are inserted or deleted, there

will be no shift in the codon reading frame; however, there will be either one extra or one missing amino acid in the final protein. Therefore, frameshift mutations result in abnormal protein products with an incorrect amino acid sequence that can be either longer or shorter than the normal protein.

• Tautomerization

Tautomerism is the ability of a molecule to exist in more than one chemical form. Many tautomers are formed by migration of a hydrogen atom, accompanied by a switch of a single bond and neighboring double bond. In cyclic structures, H- atoms can moved from one atom to another or from ring to another. This movement is called tautomeric shift. In DNA, H- atoms usually prefer specific atomic location in purine and pyrimidine bases. The N- atoms attached to C, G and A are in the amino form (- NH2). The O- atom in G and T are in Keto form (= O). Keto (C=O) enol (C-OH). Amino- (NH2) imino (=NH)



Normal base pairing in DNA is A-T and G-C. The tautomers forms are capable of unusual base pairing like T-G and C-A leading to genetic mutations by pairing incorrectly with complementary bases.

3- Explain maternal inheritance of leaf-color phenotypes in *Mirabilis jalapa* (four O'clock) variegated plants. (6 marks)

The explanation for this unusual pattern of inheritance is that the genes concerned are located in the chloroplasts within the cytoplasm, not in the nucleus, and are therefore transmitted only through the female parent. In eukaryote organisms the zygote normally receives the bulk of its cytoplasm from the egg cell and the male gamete contributes little more than a nucleus. Any genes contained in the cell organelles of the cytoplasm will therefore show maternal inheritance. The leaf variegation is due to two kinds of chloroplasts: normal green ones and defective ones lacking in chlorophyll pigment. Chloroplasts are genetically autonomous (i.e. self-determining) and have their own system of heredity in the form of chloroplast 'chromosomes'. These are small circular naked DNA molecules which carry genes controlling *some* aspects of chloroplast structure and function. A mutation in one of these genes, which affects the synthesis of chlorophyll as in *Mirabilis*, will therefore follow the chloroplast in its transmission and will not be inherited in the same way as a nuclear gene.

4- Compare between:

(12 marks)

• Autopolyploidy and allopolyploidy. <u>Autopolyploidy Type of polyploidy in which there are more</u> <u>than two sets of homologous chromosomes.</u>

Allopolyploid A polyploid formed by hybridization between two different species.

• Transition and transversion mutations.

A transition mutation is a base-pair-substitution point mutation that substitutes a purine for the other purine or a pyrimidine for the other pyrimidine, so that the purine/pyrimidine axis of the DNA molecule is maintained.

<u>A transversion mutation is a mutation that substitutes a purine</u> <u>for a pyrimidine or a pyrimidine for a purine, inverting the</u> <u>purine/pyrimidine axis of the DNA molecule.</u> 5) Which type of organisms is more affected by mutagens: prokaryotes or eukaryotes? Why? (6 marks)

Prokaryotes are more affected by mutagens because they have only one copy of each gene in its genome (haploid), while eukaryotes are diploid, i.e. have two copies (alleles) of each gene. So, if one copy is mutated, there is another copy (wild type or dominant allele) which resists the negative effect of the mutation.

6) What is the name of the process which occurs when a segment of chromosome is turned around 180°? (6 marks)

Inversion.

Dr. Mahmoud Abderraouf