Mod 8

Q. 1. Which of the following type of recombination does not require homologous sequences and is utilized by mobile genetic elements that move about chromosomes?

- a. Mutagenic recombination.
- **b**. Site-specific recombination.
- **c**. Replicative recombination.
- d. General recombination.

Ans. 1: c. Replicative recombination.

Transfer of information from one DNA molecule to another. Recombination may be reciprocal, in which case the products are equivalent to breakage of the two DNA molecules and rejoining of the broken ends to form new molecules. Replicative recombination generates a new copy of a segment of DNA. Many transposable elements use a process of replicative recombination to generate a new copy of the transposable element at a new location.

Q. 2. Which one of the following types of mutation is most likely to lead to premature termination of translation?

- a. Insertion of a single base into DNA.
- **b.** Deletion of three bases from DNA.
- **c.** Deletion of an entire gene.
- **d.** Single base change in a promoter.

Ans. 2: a. Insertion of a single base into DNA.

Insertion of one base will alter the reading frame, and usually leads to a stop codon eventually. Deletion of three bases does not alter the reading frame; deletion of the entire gene will result, obviously, in a complete lack of gene product from that allele; a base change in a promoter might increase or decrease levels of transcription; and an exon skip might or might not alter the reading frame, depending on the reading frame of the adjacent exons.

Q. 3. Which among the following leads to frame shift mutation?

- a. Deamination of cytosine to uracil
- b. Formation of thymine dimers
- c. Both a) and b)
- d. None of the above

Ans. 3: d. None of the above

A frameshift mutation is a genetic mutation caused by a deletion or insertion in a DNA sequence that shifts the way the sequence is read. 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. For instance, if just one nucleotide is deleted from the sequence, then all of the codons including and after the mutation will have a disrupted reading frame. This can result in the incorporation of many incorrect amino acids into the protein.

Q. 4. In splice-site mutations, what sequence always defines the 5' splice site (donor site)?

- a. AT
- b. CG
- c. AG
- d. GT

Ans. 4: d. GT

A mutation that alters or abolishes the specific sequence denoting the site at which the splicing of an intron takes place. Such mutations result in one or more introns remaining in the mature messenger RNA and can disrupt the generation of the protein product.

Q. 5. Sex-linked traits more common in males than in females. Why is it so?

a. A recessive allele on the X chromosome will always produce the trait in a male.

- **b.** Any allele on the Y chromosome will be codominant with the matching allele on the X chromosome.
- **c.** All alleles on the X chromosome are dominant.
- **d.** Both a) and b)

Ans. 5: c. All alleles on the X chromosome are dominant.

Males are hemizygous for sex linked characters. They possess only a single allele for sex linked character on X chromosome. Since there is no dominant allele on Y chromosome to mask the activity of this recessive allele, thus the sex linked recessive characters are expressed in males even in the presence of a single copy of gene (hemizygous).

However, female would require two recessive alleles on both of her X chromosomes to exhibit the character.

Q. 6. Polygenic inheritance occurs in

- a. human eye colour and sickle cell anaemia
- **b.** hair pigment in mouse and tongue rolling in humans
- c. human height and eye color in human
- d. ABO blood group in human

Ans. 6: c. human height.

The term "polygenic inheritance" is used to refer to the inheritance of quantitative traits, traits which are influenced by multiple genes, not just one. One easily understood example of polygenic inheritance is height. People are not just short or tall; they have a variety of heights which run along a spectrum. Furthermore, height is also influenced by environment; someone born with tall genes could become short due to malnutrition or illness, for example, while someone born with short genes could become tall through genetic therapy. Basic genetics obviously would not be enough to explain the wide diversity of human heights, but polygenic inheritance shows how multiple genes in combination with a person's environment can influence someone's phenotype or physical appearance.

Q. 7. Patients with Xeroderma pigmentosa avoid going to sunlight. Why?

- **a.** The UV wavelengths do irrepairable damage to DNA.
- **b.** Sunlight inhibits any residual DNA repair activity of the cell.
- **c.** These patients lack pigmentation to protect them from burning.
- **d.** Suniight inhibits DNA polymerases.

Ans. 7: a. The UV wavelengths do irrepairable damage to DNA.

Xeroderma pigmentosum is a rare genetic condition where DNA cannot be repaired following damage caused by UV light. XP patients exhibit photosensitivity, so must avoid bright visible lighting. In addition, XP patients are susceptible to other agents (carcinogens) that may lead to DNA damage, and are advised to avoid these substances.

Q. 8. How does the dosage compensation is achieved in *Drosophila*?

- a. X-chromosome in males in inactivated.
- b. The activity of the single X-chromosome in males is up regulated.
- c. The activity of the two X-chromosomes in females is down regulated.
- d. The activity of the autosomes in males is up regulated.

Ans. 8: b. The activity of the single X-chromosome in males is up regulated.

Dosage compensation is an epigenetic mechanism that normalizes gene expression from unequal copy numbers of sex chromosomes. Different organisms have evolved alternative molecular solutions to this task. In *Drosophila melanogaster*, transcription of the single male X chromosome is upregulated by two fold in a process orchestrated by the dosage compensation complex. Despite this conceptual simplicity, dosage compensation involves multiple coordinated steps to recognize and activate the entire X chromosome.

Q. 9. What is the sequence of the genes on the chromosome, if the cross-over percentage between linked genes J and M is 20%, J and L is 35%, J and N is 70%, L and K is 15%, M and N is 50% and M and L is 15%?

- a. J, N, M, L, K
- b. J, M, L, N, K.
- c. J, M. L, K, N
- d. M, J, L, K, N

Ans. 9: c. J, M, L, K, N

1% recombination = Imap unit. Hence, from the percent recombination we can easily find out the sequence of genes. As the percentage recombination between J and N is highest following which there is M and N and depending upon the percentage all the other genes can be located.

Mod 7 - Gr C

- Q. 10. Green gland, used for excretion in an organism having heart for circulation, has several ganglia and tactile organs on its body and its larval form is very different than its adult form. What is the most appropriate mode of respiration?
 - A. Use of extensive tracheal system for the exchange of oxygen and carbon dioxide.
 - **B.** Gaseous exchange over thinner areas of cuticle or by gills.
 - **C.** An efficient tracheal system that delivers oxygen directly to the tissues.
 - **D.** A double transport system, where the circulating fluid contains a dissolved respiratory pigment.

Which among the above mentioned mode is correct?

- a. A and C
- **b.** Only D
- c. Only B
- d. B and D

Ans. 10: c. Only B

Although a small organism can get enough oxygen by diffusion through the surface, this is usually not true for larger organisms. Of course, any shape deviating from the sphere has a larger surface and the diffusion distances are also reduced. However, in most cases this does not suffice. Often these organs also have a thinner cuticle than other parts of the body, thus facilitating gas exchange. If the respiratory surface is turned out, forming an evagination, the resulting organ is usually called a gill.

- Q. 11. ACTH secretion was increased and as a result glucocorticoid concentration was elevated in blood, under stressful condition. Which among the following changes is/are most likely take place in this condition?
 - A. Decrease in the number of circulating eosinophils and basophils.
 - B. Reduced release of IL2.
 - C. Potentiated inflammatory response to tissue injury.
 - **D.** Increased mitotic activity of lymphocytes in lymph nodes.
- a. B and C
- **b.** A and B
- c. B and D
- d. Cand D

Ans. 11: b. A and B

Glucocorticoid excess, either because of endogenous production or exogenous administration, results in a CBC pattern known as the stress leukogram, characterized by mature neutrophilia (*i.e* increased concentration of segmented neutrophils), lymphopenia and especially in dogs, monocytosis. Eosinopenia and basopenia is another feature of the stress leukogram, although in many situations this is inapparent because the normal reference values for eosinophils and basophils are so low.

- Q. 12. Arrange the sequence through which the light passes via the four refracting media of the eye.
- 1. Vitreous body
- 2. Lens
- 3. Aqueous humor
- 4. Cornea
 - **a.** 1,2.3,4
 - **b.** 4, 1,2,3
 - **c.** 4, 3, 2, 1
 - **d.** 2, 3,4,1

Ans. 12: e. 4, 3,2, 1

The refractive media of eye is composed of four parts of eye, the cornea which is the anterior 1/6 of the external tunic (fibrous

tunic) of the eyeball. The aqueous humor is second part of the refractive media of eye, it is a colourless, transparent and watery fluid, it fills the chambers of the eye and is formed by active transport and diffusion from the capillaries of the ciliary process from which it enters the posterior chamber then it passes into the anterior chamber through pupil and escapes from the iridocorneal angle into the sinus vinus sclerae through the space of iridocorneal angle and finally drains in the opthalamic veins via anterior ciliary veins. The third part of the refractive media of the eyeball is the lens which is a adjustable, transparent and elastic, convex body lacking blood vessels and nerves and the convexity of its anterior surface is less than the of it's posterior surface. The last one of the refractive media of eye is the vitreous body. The vitreous body is a colourless, transparent and jelly-like body which is fills the posterior fourth-fifths of the eyeball. It is also a condensed superficially to form a transparent envelope, the vitreous membrane.

Q. 13. In a normal human being, AIDS is caused by the infection of HIV. This virus undergoes spontaneous mutation, thus it is very difficult to cure a person with HIV infection. What is the cause of such spontaneous mutations occuring in this virus?

- a. Replicating enzyme for AIDS virus makes large number of mistakes.
- b. High drug resistance due to high accuracy rate and high rate of virus production.
- c. High drug resistance due to low accuracy rate and low rate of virus production.
- d. Replicating enzyme for AIDS virus makes small number of mistakes.

Ans. 13:a. Replicating enzyme for AIDS virus makes large number of mistakes

A major challenge to the effective treatment of AIDS is the unique and alarming speed with which HIV becomes resistant to every drug deployed against it, including inhibitors of viral reverse transcriptase and HIV protease. The origins of drug resistance in HIV lie in the high rate of viral replication and the ease with which spontaneous mutations arise in its RNA genome. As a single-stranded RNA virus, HIV lacks a proofreading mechanism to eliminate sequence errors resulting from the low fidelity of HIV reverse transcriptase. As a result, mutations occur with high frequency.

Q. 14. Bile salts are highly lipophylic in nature, hence before facilitating lipid absorbtion they could be rapidly absorbed through the small intestine wall. This can be prevented by

- **a.** addition of hydroxyl and carboxyl acid groups to steroid nucleus of cholesterol.
- b. combining the bile acids with phospholipids to make it less easily absorbed.
- c. conjugating bile acids to glycine or taurine so that they ionize readily.
- **d.** binding the bile salts to the cholesterol to keep them from being absorbed.

Ans. 14: c. conjugating bile acids to glycine or taurine so that they ionize readily.

Addition of organic acids influenced the ionization efficiency of whole bile acids. Use of a stronger acid reduced the peak intensity of unconjugated and glycine- conjugated bile acids, while the use of TFA, the strongest acid tested, improved the intensity of taurine conjugates. The hydroxyl group at the C-12 alpha position of cholic acid and deoxycholic acid easily underwent intra-molecular hydrogen bonding with the side chain carboxyl group, accelerating the ionization efficiency. This intra-molecular hydrogen bond may also affect the formation of product ions in low energy-CID. The addition of ammonium ions to the spray liquid influenced the ionization of all bile acids, specifically enhancing the ionization efficiency of unconjugated bile acids.

Q. 15. A person went to a doctor, with an elevated serum free calcium. What could be the best conclusion, if the parathyroid hormone level (PTH) is in the normal range?

- **a.** The PTH is normal, therefore the problem does not lie in the parathyroid gland.
- **b.** The person must have excessive sensitivity to PTH, since normal levels are stimulating excessive calcium mobilization from bone.

- c. The PTH should be low if the parathyroid were functioning normally, thus the problem does lie in the parathyroid gland.
- **d.** Doctor cannot be sure what is going on; he need to perform a parathyroid scan.

Ans. 15: c. The PTH should be low if the parathyroid were functioning normally, thus the problem does lie in the parathyroid gland.

Primary hyper-parathyroidism is a disorder of the parathyroid glands, also called parathyroids. "Primary" means this disorder originates in the parathyroid glands. In primary hyper-parathyroidism, one or more of the parathyroid glands are overactive. As a result, the gland releases too much parathyroid hormone (PTH). The disorder includes the problems that occur in the rest of the body as a result of too much PTH—for example, loss of calcium from bones. The parathyroid glands produce PTH, a hormone that helps maintain the correct balance of calcium in the body. PTH regulates the level of calcium in the blood, release of calcium from bone, absorption of calcium in the small intestine and excretion of calcium in the urine. When the level of calcium in the blood falls too low, normal parathyroid glands release just enough PTH to restore the blood calcium level.

Q. 16. Spermatogenesis occur only during adulthood, whereas the level of FSII during infancy and adulthood remain the same. And the mRNA levels coding for FSH receptor are also found to be the same in testis of both age groups. For more clarified investigation

- **a.** culture testicular cells and add LH to see testosterone production.
- **b.** culture testicular cells and add testosterone to see comparative rise in FSH mRNA.
- **c.** culture testicular cells and add FSH to see comparative rise in cAMP production.
- d. add both LH and FSH to testicular cells to see the rise in cAMP production.

Ans. 16: d. add both LH and FSH to testicular cells and evaluate cAMP production.

LH binds to Leydig's cells and increases cAMP which increases protein secretion and the side-chain cleavage of cholesterol, as well as other likely steps, to increase steroidogenesis and the production of testosterone and other androgens. Regulated by steroid feedback the Leydig's cells produce the testicular steroids, lie between the seminiferous tubules and assist in the transportation of steroids in the blood, lymph and seminiferous tubules. FSH binds to the Sertoli cells of the seminiferous tubules, increases cAMP and protein synthesis androgen binding in the tubules, etc. regulated by inhibin produced by the sertoli cells. Sertoli cells secrete proteins that are important to spermatogenesis and have been called the director cells of spermatogenesis. They comprise the blood-testis barrier.

Q. 17. What is the sequence of events that causes decompression sickness in a person that dives deep under water and resurfaces rapidly?

- a. Increase in pressure —> N2 dissolves in blood —» Rapid fall in pressure —> N2 bubbles in blood
- b. Increase in pressure —> Pressure on ear oscicles —> Loss of orientation —> Imbalance
- c. Fall in pressure —>Loss of 02 from blood —>Deoxygenation —> Fatigue
- d. Increased pressure Enhanced blood pressure -» Headache —» Loss of memory'

Ans. 17: a. Increase in pressure —> N₂ dissolves in blood —» Rapid fall in pressure —» N₂ bubbles in blood

When a diver descends in the water the hydrostatic pressure and therefore the ambient pressure, rises. Because breathing gas is supplied at the same ambient pressure as the surrounding water, some of this gas dissolves into the diver's blood from where it is transferred by the blood to other tissues. On ascent, the ambient pressure is reduced, the inert gases dissolved in the tissues are then at a higher concentration than the equilibrium state and start to diffuse out again, possibly forming bubbles, which may lead to decompression sickness, a possibly debilitating or life- threatening condition. A mismanaged decompression usually results from reducing the ambient pressure too quickly, allowing the dissolved inert gases such as nitrogen or helium to form bubbles in the blood and tissues in a manner similar to the fizzing of a carbonated beverage when opened. These bubbles may block arterial blood supply to tissues or cause tissue damage. If the decompression is effective, the asymptomatic venous microbubbles present after most dives are eliminated from the diver's body in the alveolar capillary beds of the lungs. If they are not given enough time or more bubbles are created than can be eliminated safely, the bubbles grow in size and number causing the symptoms and injuries of decompression sickness.

Mod 8

- Q. 10. A chromosomal abnormality, Philadelphia chromosome is associated with chronic myelogenous leukemia. What could be the correct definition of Philadelphia chromosome?
- a. Translocated chromosomes 9 from 22
- **b.** Translocated chromosomes 22 from 9
- **c.** Base substitution in chromosome 22
- **d.** Base substitution in chromosome 9

Ans. 10: b. Translocated chromosomes 22 from 9

Philadelphia chromosome (Ph) is the chromoso—; abnormality that causes chronic myeloid leukemia (C\L abbreviated as the Ph chromosome. The Ph chromosome an abnormally short chromosome 22 that is one of the v chromosomes involved in a translocation (an exchange material) with chromosome 9. This translocation takes pi-., in a single bone marrow cell and through the procese clonal expansion (the production of many cells from this ore mutant cell), it gives rise to the leukemia.

- Q. 11. One mechanism of dosage compensation involves X-inactivation; which ensures that male (XY) and female (XX) mammals contain the same concentration of gene products encoded by the X chromosome. What is the other mechanism for dosage compensation found in insects?
- **a.** Doubling of X chromosome expression in males, than found in females.
- **b.** Doubling of X chromosomes expression in females, than found in males.
- **c.** Reduction in Y chromosomes expression in females, than in males.
- **d.** Halfing the X chromosome expression in female, than in males.
- e. Ans. 11: d. Halfing the X chromosome expression in female, than in males.

The evolution of sex chromosomes has resulted in numerous species in which females inherit two X chromosomes but males have a single X, thus requiring dosage compensation. MSL (Male-specific lethal) complex increases transcription on the single X chromosome of *Drosophila* males to equalize expression of X-linked genes between the sexes. The biochemical mechanisms used for dosage compensation must function over a wide dynamic range of transcription levels and differential expression patterns.

- Q. 12. A trihybrid cross is made between two yeast, both with genotype E/e, F/f, G/g. What proportion of the offsprings will be of the genoty pe e/e, f/f and g/g?
- a. 1/32
- **b.** 1/3
- c. 1/64
- d. None of the above

Ans. 12: c. 1/64

Frequency of e/e= 1/4, f/f = 1/4, g/g = 1/4

Therefore, probability of offsprings to be e/e, f/f and g/g will be

 $1/4 \times 1/4 \times 1/4 = 1/64$

- Q. 13. In a cross involving polygenic inheritance where three gene pairs control plant height, the shortest and tallest plants are 6 cm and 24 cm, respectively. What height should all FI display if homozygous 12- cm and 24-cm plants are crossed, assuming their environments are the same?
- a. 20 cm
- b. 43 cm

- c. 6 cm
- d. 18 cm

Ans. 13: d. 18 cm

FI offspring will all have the same genotype (1/2 of genome from each parent). Therefore, the genes coding for the extreme phenotypes are present in equal numbers. This results in a phenotype which is approximately halfway between the two extremes.

- Q. 14. Gradual deterioration of the muscles are the consequences of pseudohypertrophic muscular dystrophy, that is an inherited disorder. It is seen almost exclusively in baby born to apparently normal parents and usually results in death in the early teens. What type of disease is this?
- a. Autosomal dominant
- **b.** Autosomal recessive
- c. Sex linked recessive
- d. Sex linked dominant

Ans. 14: c. Sex linked recessive

The most common and severe type of muscular dystrophy is called Duchenne Muscular Dystrophy (DMD). This muscle- wasting disorder, which affects boys almost exclusively, typically has its onset between the ages of two and five and progresses rapidly. DMD is sometimes referred to as pseudohypertrophic muscular dystrophy because it characteristically results in a seeming enlargement of the calf muscles, which look abnormally big because fat and connective tissue have replaced degenerating muscle fibres. Like the other muscular dystrophies, DMD is inherited - it is a genetic condition. Unlike most of the other dystrophies, it is transmitted by an altered gene on the X chromosome in an "X-linked" (or "sex-linked") recessive pattern of inheritance. When a disorder is transmitted in this way only males are affected females, who rarely show any symptoms, may be carriers of the defective gene and pass on the disease to their sons and indirectly, to their grandsons through daughters who are carriers. The same inheritance pattern is seen in hemophilia and colour blindness.

- Q. 15, A stop codon has been created within a coding sequence, in case of 'non-sense' mutation those results in termination of translation because there is no corresponding tRNA to recognize them. However, tRNA molecules are themselves coded by genes, which are of course susceptible to mutation. Hence, it is possible to change an existing tRNA gene in such a way that it will recognize one of the stop codons rather than (or as well as) the codon it normally recognizes. What is such a phenomenon called in which the effect of a mutation can be negated by a second, unrelated mutation?
 - a. Bach mutation
 - **b.** Complementation
 - c. Suppression mutation
 - **d.** Epistatis

Ans. 15: c. Suppression mutation

A suppressor mutation is a second mutation that alleviates or reverts the phenotypic effects of an alread\existing mutation. Genetic suppression therefore restores the phenotype seen prior to the original background mutation. Suppressor mutations are useful for identifying new genetic sites which affect a biological process of interest. They also provide evidence between functionally interacting molecules and intersecting biological pathways.

- 16. During an experiment, an investigator found that the cell line used are RecA -/-. What could be the probable finding which led him to such observation?
- (a) Loss of recombination
- (b) Showing aberrant all morphology
- (c) Cells developed phagocytic properly
- (d) Cells were aggregating

- 16.Sol.(a) Usually in case of linked gene or characters, the production of new combination of genes or characters not present in parent of a cross or generation is called recombination. It involves crossing over. Since the cell line is homozygous it means= recombination is not found in these. In presence of recombination new progency cell will be formed, *i.e.*, they will be heterozygous type.
 - 17. In *E. coli*, four *Hfr* strains donate the genetic markers shown in the order shown: Strain 1: F L K O Z Strain 2: C Y A Z O Strain 3: P D W C Y Strain 4: P F L K O

All the $H\!f\!r$ strains are derived from the same F^+ strain. What is the order of these markers on the circular chromosome of the original $E.\ coli\ F^+$ strain?

- (a) KOZPDFLWCYAK
- (b) FLKOZPDWCYAF
- (c) FLKOZAYCWDPF
- (d) FLKCYAZOWDPF

17. Sol. (c) When the chromosome of F^+ cell integrates with F plasmid it is called high frequency recombination (Hfr) cell. The time at which a particular gene enters a recipient is related to the portion of genes on the chromosome. A map can be obtained from the time of entry of each gene.

Strain 1 — FLKOZ

Strain 2 — OZAYC

Strain 3 — YCWDP

Strain 4 — PFLKO