

Case Based Questions

Q.No.1: Read the following and answer any four questions from (i) to (v) given below:

Thalassemia is an inherited blood disorder that causes our body to have less haemoglobin than normal due to mutations in the DNA of cells that make haemoglobin. The mutations associated with thalassemia are passed from parents to children.

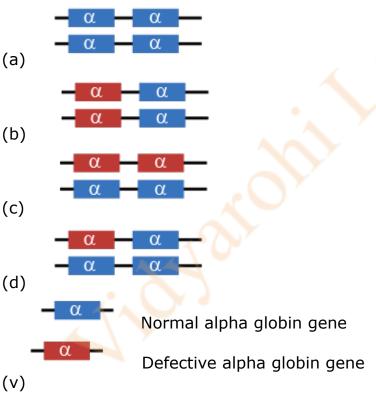
Haemoglobin molecules are made of chains called alpha and beta chains that can be affected by mutations. In thalassemia, the production of either the alpha or beta chains is reduced, resulting in either alpha-thalassemia or betathalassemia. The more mutated genes, the more severe thalassemia is. In alpha-thalassemia, the severity of thalassemia depends on the number of gene mutations an individual inherits from his parents. Four genes are involved in making the alpha haemoglobin chain and an individual gets two from each of the parents. If an individual has one mutated gene, he will have no signs or symptoms of thalassemia but the person will be a carrier of the disease. If an individual has two mutated genes, thalassemia signs and symptoms will be mild. This condition might be called alpha-thalassemia trait. If an individual has three mutated genes, thalassemia signs and symptoms will be moderate to severe. This is called haemoglobin H disease. Inheritance of four mutated genes is rare and usually results in stillbirth. This condition is called alpha thalassemia major or hydrops fetalis. Babies born with this condition often die shortly after birth or require lifelong transfusion therapy.

In beta-thalassemia, the severity of thalassemia depends on which part of the haemoglobin molecule is affected. Two genes are involved in making the beta haemoglobin chain and an individual gets one from each of the parents. If a person inherits one mutated gene, mild signs and symptoms will appear. This condition is called thalassemia minor or beta-thalassemia. If a person inherits two mutated genes, signs and symptoms will be moderate to severe. This condition is called thalassemia major, or Cooley anaemia. Babies born with two defective beta haemoglobin genes usually are healthy at birth but develop signs and symptoms within the first two years of life. A milder form, called thalassemia intermedia, also can result from two mutated genes.

- (i) Which of the following statement is not correct about thalassemia disease?
- (a) Thalassemia is an autosomal-linked recessive disease.
- (b) Thalassemia is caused due to qualitative problem of haemoglobin.
- (c) Alpha-thalassemia is controlled by two closely linked genes present on chromosome 16 of each parent.
- (d) The mutation or deletion in genes for alpha and beta globin chain results in

alpha and beta thalassemia, respectively.

- (ii) Cooley anaemia results when
- (a) four alpha haemoglobin genes are mutated.
- (b) one beta-haemoglobin gene is mutated.
- (c) two beta-haemoglobin genes are mutated.
- (d) three-alpha haemoglobin genes are mutated.
- (iii) Which of the following statement is incorrect about alpha- and betathalassemia?
- (a) In alpha thalassemia, the haemoglobin does not produce enough alpha protein.
- (b) The severity of thalassemia depends on how many genes are mutated.
- (c) An individual inherits four genes for alpha chain of haemoglobin from each parent.
- (d) Beta-thalassemia occurs when a person inherits one defective globin gene for beta chain.
- (iv) The alpha-thalassaemia trait has been associated with protection against severe malaria. Which of the following genotype will confer protection against malaria?



Assertion: Hydrops fetalis is a condition in alpha-thalassemia.

Reason: It results due to inheritance of all four defective genes for alpha globin chain.

- (a) Both assertion and reason are true, and the reason is the correct explanation of the assertion.
- (b) Both assertion and reason are true, but the reason is not the correct explanation of the assertion.
- (c) Assertion is true but reason is false.
- (d) Both assertion and reason are false.

Q.No.2: Read the following and answer any four questions from (i) to (v) given below.

Packaging of DNA Helix

Each diploid human cell contains genome of approximately 6 billion base pairs of DNA packaged into 23 pairs of chromosomes. Because each base pair is around 0.34 nanometers long, each diploid cell therefore contains about 2 meters of DNA. How is this chromosomal DNA compacted into the microscopic space of the eukaryotic nucleus? In eukaryotes, DNA is wrapped around an octamer of proteins known as histones to form structures called nucleosomes. H1, H2A, H2B, H3 and H4 are five major types of histones involved eukaryotic DNA packaging. Each nucleosome is linked to the next one with the help of a linker DNA. This is also known as the "beads on a string" structure.

- (i) The histone octamer of a nucleosome is formed of
- (a) two molecules each of H1, H2A, H2B and H3.
- (b) two molecules each of H2A, H2B, H3 and H4.
- (c) two molecules each of H2A, H2B and H3 and one molecule each of H1 and H3.
- (d) one molecule each of H2A, H2B and two molecules each of H1, H3 and H4.
- (ii) Histones are rich in
- (a) acidic amino acids.
- (b) polar amino acids.
- (c) basic amino acids.
- (d) non-polar amino acids.
- (iii) DNA is wrapped tightly around histone octamer due to interaction of
- (a) negative charges on DNA with positive charges on histone proteins.
- (b) negative charges of histones with negative charges on DNA.
- (c) positive charges on DNA with negative charges on histone proteins.
- (d) positive charges on DNA with positive charges on histone proteins.
- (iv) What does "bead" represent in "beads on string structure"?
- (a) DNA
- (b) Histone octamer
- (c) Linker DNA
- (d) Nucleosome

(v)

Assertion: Chromatin appears as euchromatin and heterochromatin in a nucleus.

Reason: Euchromatin stains lightly and heterochromatin stains darkly.

- (a) Both assertion and reason are true, and the reason is the correct explanation of the assertion.
- (b) Both assertion and reason are true, but the reason is not the correct explanation of the assertion.
- (c) Assertion is true but reason is false.
- (d) Both assertion and reason are false.

Q.No.3: Read the following and answer any four questions from (i) to (v) given below.

The life cycle of HIV

HIV cannot reproduce on its own. HIV attaches itself to a type of white blood cell that keep us healthy by fighting off infections and diseases. The virus then fuses with it and takes control of the cell's DNA, makes copies of itself inside the cell, and finally releases more HIV into the blood. HIV will continue to multiply and spread throughout the body – a process called the HIV lifecycle. In this way, HIV weakens the body's natural defenses and over time severely damages the immune system.

- (i) Read the following statements about HIV and select the option with correct statements.
- A. HIV is a member of group of viruses called retroviruses.
- B. HIV infects white blood cells called T-helper cells.
- C. AIDS caused by HIV is a congenital disease.
- (a) A, C
- (b) B, C
- (c) A, B
- (d) A, B, C
- (ii) HIV can enter our body:
- (a) by sharing utensils.
- (b) from mosquito bite.
- (c) by sharing toilets.
- (d) from an infected mother to child during breast-feeding.
- (iii) What happens after the fusion of HIV with an immune cell?
- (a) Viral DNA is transcribed into viral RNA using host's transcription machinery.
- (b) Viral RNA is transcribed into DNA using viral reverse transcriptase.
- (c) Viral genome is integrated into host's genome using viral enzyme, integrase.
- (d) Viral RNA is used to make multiple copies of viral RNA using host's machinery.
- (iv) The test performed to detect HIV infection is:
- (a) RFLP
- (b) ELISA
- (c) Southern blot
- (d) Western blot
- (v) Treatment of AIDS involves
- (a) vaccination that destroys the virus.
- (b) drugs that target different stages of the lifecycle of HIV.
- (c) transfusion of immune cells into the body of an infected person.
- (d) administration of substances such as a-interferon to activate immune system.

Q.No.4:

In Mendel's experiments on garden pea plants, each gene exists in two alleles which are inherited as dominant and recessive. Mendel's results were groundbreaking partly because they contradicted the historical view that offspring always exhibit a blend of parents' traits. In some cases, for example, inheritance of flower colour in the snapdragon, *Antirrhinum majus*, however, alleles are not always fully dominant or recessive to one another. The phenotype of a heterozygous organism, thus, does not resemble either of the two parents instead is intermediate between the phenotypes of its homozygous parents. In snapdragon, a cross between a homozygous parent with white flowers and a homozygous parent with red flowers will produce offspring with pink flowers. It implies that neither of the alleles is completely dominant over the other in such cases.

- (i) The phenomenon in which an offspring exhibits a phenotype intermediate between the phenotypes of its homozygous parents due to absence of complete dominance of either of the alleles is called
- (a) Codominance.
- (b) Pleiotropy.
- (c) Incomplete dominance.
- (d) Epistasis.
- (ii) Suppose the two genes controlling the red and white flower colours in snapdragon are R and r, respectively. What would be the phenotypic ratio of a cross between a pink-flowered snapdragon and white-flowered snapdragon?
- (a) 2 Pink : 1 white (b) 2 Pink : 2 White (c) 2 Pink : 2 Red
- (d) 1 Red: 2 Pink: 1 White
- (iii) Which of the followings is the results of a cross between two pink-flowered snapdragons?
- (a) Phenotypic ratio 3:1; genotypic ratio 1:2:1
- (b) Phenotypic ratio 3:1; genotypic ratio 3:1
- (c) Phenotypic ratio 1:2:1; genotypic ratio 3:1
- (d) Phenotypic ratio 1:2:1; genotypic ratio 1:2:1
- (iv) Which of the following result of the cross follows the same pattern of inheritance as exhibited by flower colour in snapdragons?
- (a) The hybrid cattle of a cross between cattle with black coat and cattle with white coat have roan coat colour, where black and white patches appear separately.
- (b) The hybrid Andalusian fowl of a cross between a black Andalusian fowl and a white Andalusian fowl has blue fur colour.
- (c) On crossing a squash plant with white fruit with another squash plant with green fruit, a hybrid squash plant with white fruit is obtained.
- (d) A hybrid corn of a cross between a corn with yellow kernels and a corn with white kernel has a nearly even mix of yellow and white kernels.
- (v) When two dogs with wavy coat are crossed, offspring dogs with curly coat, straight coat and wavy coat resulted. Select the option that shows the genotype of a dog with wavy coat if alleles controlling the coat in dogs are K^{C} and K^{+} .
- (a) K^CK^C
- (b) K⁺K^C
- (c) K⁺K⁺
- (d) K+K+ KCKC