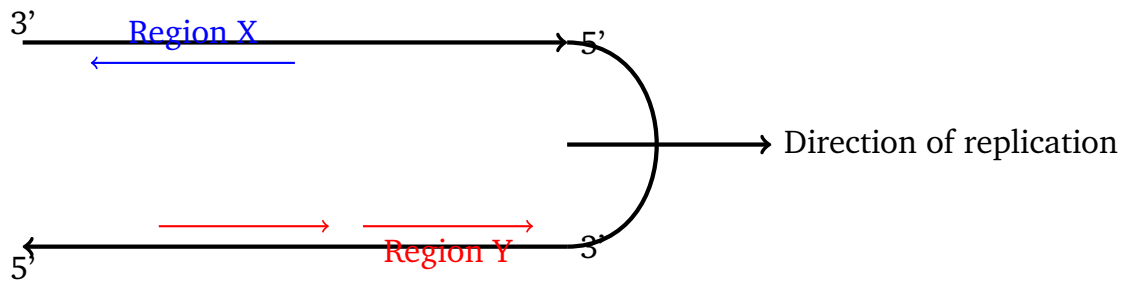
NEET PG Biochemistry Sample Paper-8

Duration: 15 Minutes **Maximum Marks: 64**

Instructions

- This paper contains **16** Multiple Choice Questions.
- Each correct answer carries **+4** mark. Incorrect answer: **−1** marks. Only **one** correct option.
- Unattempted questions carry **0** marks.
- Use of mobile phones, smartwatches, or any electronic gadgets is strictly prohibited.

Q1. A researcher is analyzing a segment of eukaryotic DNA during replication. The following schematic represents a replication fork with specific functional regions marked:



Which of the following descriptions correctly identifies the nature of synthesis occurring at Region X and Region Y?

(A) Region X represents continuous synthesis (leading strand); Region Y represents discontinuous synthesis (lagging strand).

(B) Region X represents discontinuous synthesis (lagging strand); Region Y represents continuous synthesis (leading strand).

(C) Both Region X and Region Y undergo continuous synthesis via DNA Polymerase α.

(D) Both Region X and Region Y undergo discontinuous synthesis via Okazaki fragments.

Q2. A 45-year-old chronic alcoholic presents with confusion, ataxia, and ophthalmoplegia. Laboratory evaluation reveals elevated serum pyruvate and lactate levels. A deficiency in which of the following enzymes is most likely responsible for this clinical presentation?

(A) Pyruvate kinase

(B) Pyruvate dehydrogenase complex

(C) Glucose-6-phosphate dehydrogenase

(D) Phosphofructokinase-1

Q3. A 2-week-old infant is brought to the clinic due to poor feeding, vomiting, and progressive jaundice. Urinalysis shows a strong positive reaction for reducing sugars, but a glucose oxidase dipstick test is negative. Which of the following enzymes is most likely deficient in this infant?

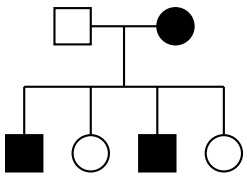
(A) Galactokinase

(B) Galactose-1-phosphate uridyltransferase

(C) Fructokinase

(D) Aldolase B

Q4. A pedigree chart of a family affected by a specific metabolic disorder is shown below:



Given that the mutant allele is extremely rare in the general population, what is the most likely mode of inheritance illustrated in this pedigree?

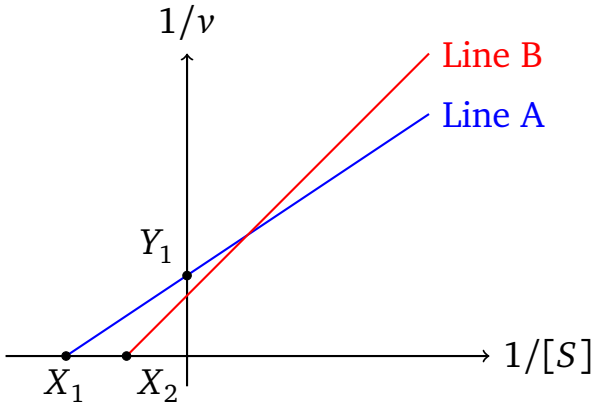
(A) Autosomal Dominant

(B) Autosomal Recessive

(C) X-linked Dominant

(D) Mitochondrial / Maternal Inheritance

Q5. An enzyme-catalyzed reaction is studied in the presence and absence of an inhibitor. The kinetic data is plotted on a Lineweaver-Burk double-reciprocal graph as shown below:



Which of the following statements correctly evaluates the kinetic parameters and the type of inhibition observed?

(A) Line B represents competitive inhibition, where V_{max} remains unchanged and K_m is increased.

(B) Line B represents non-competitive inhibition, where V_{max} is decreased and K_m remains unchanged.

(C) Line B represents uncompetitive inhibition, where both V_{max} and K_m are decreased.

(D) Line B represents competitive inhibition, where V_{max} is decreased and K_m is decreased.

Q6. A 5-year-old child presents with mental retardation, lens dislocation, and long, thin fingers (marfanoid habitus). Laboratory tests reveal markedly elevated levels of plasma methionine and urinary homocystine. Supplementation with high doses of which of the following vitamins is most likely to benefit this patient?

(A) Thiamine (Vitamin B1)

(B) Riboflavin (Vitamin B2)

(C) Pyridoxine (Vitamin B6)

(D) Cobalamin (Vitamin B12)

Q7. During a laboratory exercise, a student is asked to identify a point mutation in a specific structural gene. The schematic shows a template DNA strand and its corresponding altered sequence after a replication error:

Original:	5'-A	T	G	C	A	T-3'
Mutated:	5'-A	T	G	T	A	T-3'

If this single base substitution changes a codon specifying a hydrophilic amino acid to one specifying a hydrophobic amino acid without altering the translational reading frame, this mutation is best classified as a:

(A) Nonsense mutation

(B) Silent mutation

(C) Frameshift mutation

(D) Missense mutation

Q8. A 30-year-old female presents with severe muscle cramps and weakness during short-duration, high-intensity exercise. Ischemic forearm exercise testing demonstrates a failure of blood lactate levels to rise despite significant effort. A muscle biopsy indicates a marked accumulation of glycogen with normal structure. Which of the following glycogen metabolism enzymes is deficient?

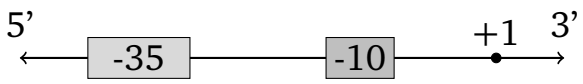
(A) Glucose-6-phosphatase

(B) Myophosphorylase

(C) Acid α-glucosidase

(D) Glycogen debranching enzyme

Q9. An investigator isolates a bacterial mutant whose RNA polymerase binds tightly to the promoter region but fails to initiate the transition from the closed complex to the open complex during transcription. The structural arrangement of the prokaryotic promoter elements is mapped below:



Which of the following subunits of prokaryotic RNA polymerase is most likely structurally defective or missing in this mutant strain?

(A) Alpha (α) subunit

(B) Beta (β) subunit

(C) Sigma (σ) factor

(D) Omega (ω) subunit

Q10. A 52-year-old man with a history of severe alcoholism and malnutrition is admitted with a bleeding tendency, swollen and bleeding gums, and petechiae around hair follicles on his lower extremities. Hydroxylation of which of the following pairs of amino acid residues is directly impaired in this patient due to his underlying vitamin deficiency?

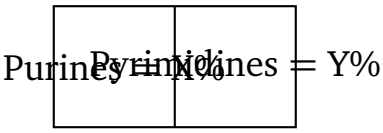
(A) Lysine and Valine

(B) Proline and Lysine

(C) Glycine and Proline

(D) Alanine and Serine

Q11. An analytical biochemistry laboratory evaluates a sample of double-stranded DNA. The structural integrity of the nitrogenous bases is verified using quantitative degradation. The proportional distribution of a subset of bases is represented in the diagram below:



According to Chargaff's rules applied to standard genomic double-stranded DNA, what must be the values of X and Y?

(A) X = 40%, Y = 60%

(B) X = 60%, Y = 40%

(C) X = 50%, Y = 50%

(D) The values depend entirely on the specific GC-content of the organism's genome.

Q12. A newborn is evaluated for hypotonia, hepatomegaly, and severe fasting hypoglycemia. Administration of epinephrine or glucagon triggers no significant rise in blood glucose levels during fasting, but a normal glycemic response is elicited shortly after a carbohydrate-rich meal. A liver biopsy reveals an accumulation of abnormal glycogen containing short, outer branches. Which enzyme is defective?

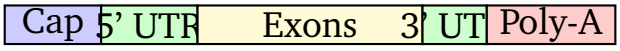
(A) Glycogen synthase

(B) α-1,6-glucosidase (Debranching enzyme)

(C) α-1,4 $\rightarrow$ α-1,4 glucan transferase

(D) Amylo-α-(1,4)$\rightarrow\alpha$-(1,6)-transglucosidase (Branching enzyme)

Q13. The processing of eukaryotic precursor mRNA involves several coordinated modifications. A schematic of a fully processed eukaryotic mature mRNA molecule is shown below:



Which of the following descriptions accurately characterizes the enzymatic synthesis or structural linkage of the 'Cap' located at the 5' terminus?

(A) A 7-methylguanosine residue attached via a standard 5' to 3' phosphodiester bond.

(B) A 7-methylguanosine residue attached via an unusual 5' to 5' triphosphate linkage.

(C) An unmethylated adenosine residue linked via a 3' to 5' ester linkage.

(D) A modified uridine tail added post-transcriptionally by poly-U polymerase.

Q14. A 3-year-old boy presents with severe developmental delay, self-mutilating behavior (lip and finger biting), and hyperuricemia. Urinalysis reveals an abundance of orange, sand-like crystals in the diapers. The metabolic pathway responsible for this condition involves a defect in a salvage mechanism. Which of the following enzymes is deficient?

(A) Adenosine deaminase (ADA)

(B) Hypoxanthine-guanine phosphoribosyltransferase (HGPRT)

(C) Xanthine oxidase

(D) Phosphoribosyl pyrophosphate (PRPP) synthetase

Q15. A 35-year-old female presents with severe episodic abdominal pain, neuropsychiatric disturbances, and a history of dark wine-colored urine when samples are left standing in light. Biochemical analysis confirms a defect in heme biosynthesis. The diagnostic pathway demonstrates an accumulation of early porphyrin precursors. Which of the following enzymes is most likely deficient in this patient?

(A) δ-Aminolevulinic acid (ALA) synthase

(B) Porphobilinogen deaminase

(C) Uroporphyrinogen decarboxylase

(D) Ferrochelatase

Q16. A couple seeks genetic counseling because the husband has a well-documented autosomal dominant disorder. The wife is genotypically normal. The husband's father was entirely unaffected by the condition, while his mother was heterozygous and severely symptomatic. What is the probability that this couple will pass the mutant allele to their first child?

(A) 0%

(B) 25%

(C) 50%

(D) 100%

Detailed Solutions

Q1.

Solution

Concept:

DNA replication is bidirectional and asymmetrical because DNA polymerases can only synthesize new strands in the 5' to 3' direction. At a replication fork, the two template strands run in opposite antiparallel directions, requiring distinct continuous and discontinuous mechanisms to replicate both strands simultaneously.

Solution:

(a) The replication fork moves from left to right as indicated by the schematic arrow. The upper template strand runs 3' to 5' from left to right, allowing continuous synthesis toward the fork.

(b) Region X is being synthesized on the upper strand in the 5' to 3' direction toward the moving replication fork, making it the leading strand, which undergoes continuous synthesis.

(c) The lower template strand runs 5' to 3' from left to right, meaning synthesis must proceed away from the replication fork to maintain the mandatory 5' to 3' polymerisation direction.

(d) Region Y shows short fragments synthesized away from the fork, which represents the lagging strand where synthesis is discontinuous, producing Okazaki fragments that are later ligated.

(e) DNA Polymerase α synthesizes the RNA-DNA primers, but elongation of the leading and lagging strands is carried out by polymerases ϵ and δ respectively.

Final Answer: Region X represents continuous synthesis (leading strand); Region Y represents discontinuous synthesis (lagging strand).

Answer: (A)

Go Back to Question 1

Q2.

Solution

Concept:

The clinical triad of confusion, ataxia, and ophthalmoplegia in a chronic alcoholic patient is diagnostic of Wernicke encephalopathy. This condition is caused by a severe deficiency of thiamine (Vitamin B1), an essential cofactor required for key enzymes in carbohydrate metabolism, particularly those involved in glucose utilization within the central nervous system.

Solution:

(a) Thiamine pyrophosphate (TPP) serves as an essential coenzyme for the pyruvate dehydrogenase (PDH) complex, α-ketoglutarate dehydrogenase, and transketolase.

(b) When thiamine is deficient, the activity of the PDH complex is severely impaired, preventing the aerobic conversion of pyruvate into acetyl-CoA for entry into the citric acid cycle.

(c) Consequently, pyruvate accumulates upstream and is shunted into the anaerobic pathway via lactate dehydrogenase, converting it into lactate and causing lactic acidosis.

(d) This metabolic block compromises ATP production in glucose-dependent tissues like the brain, leading to localized cellular dysfunction and the characteristic neurological symptoms seen in Wernicke encephalopathy.

(e) Other listed enzymes like pyruvate kinase, glucose-6-phosphate dehydrogenase, and phosphofructokinase-1 do not utilize thiamine pyrophosphate as a cofactor and are not primarily inactivated by thiamine deficiency.

Final Answer: Pyruvate dehydrogenase complex

Answer: (B)

Go Back to Question 2

Q3.

Solution

Concept:

Classic galactosemia is an autosomal recessive metabolic disorder arising from an inability to properly metabolize galactose. It presents early in infancy with life-threatening manifestations following milk ingestion, as lactose is hydrolyzed into glucose and galactose in the intestinal tract.

Solution:

(a) The combination of progressive jaundice, vomiting, and poor feeding after milk consumption points toward an inborn error of galactose or fructose metabolism.

(b) Urinalysis detects the presence of reducing sugars, which includes galactose, fructose, and lactose, giving a positive reaction in non-specific copper reduction tests like Benedict's reagent.

(c) The negative glucose oxidase dipstick test specifically excludes glucose as the source of the reducing sugar, narrowing the diagnostic possibilities to galactose or fructose.

(d) The early onset at two weeks of age specifically fits galactose exposure from milk, rather than fructose, which is only introduced later in life with solid food or juices.

(e) A deficiency of galactose-1-phosphate uridyltransferase leads to the accumulation of toxic galactose-1-phosphate in tissues, causing severe hepatic dysfunction, cataracts, and systemic injury.

Final Answer: Galactose-1-phosphate uridyltransferase

Answer: (B)

Go Back to Question 3

Q4.

Solution

Concept:

Mitochondrial inheritance, also known as maternal inheritance, applies to genes contained within the circular mitochondrial genome. Because mitochondria are transmitted to the zygote exclusively via the cytoplasm of the oocyte, the inheritance pattern follows strict maternal lines.

Solution:

(a) Analysis of Generation I shows an affected mother and an unaffected father sharing a pedigree bond.

(b) In Generation II, all four children, consisting of two sons and two daughters, inherit the metabolic condition and are fully affected.

(c) If the condition were X-linked dominant, an affected mother would have a 50% chance of passing it to any child, rather than 100

(d) If it were autosomal dominant or recessive, a 100% transmission rate to all offspring of both sexes from a rare mutant allele is highly improbable.

(e) The pedigree demonstrates that an affected female transmits the trait to all of her children, while affected males cannot pass the condition to their offspring, confirming maternal mitochondrial inheritance.

Final Answer: Mitochondrial / Maternal Inheritance

Answer: (D)

Go Back to Question 4

Q5.

Solution

Concept:

Enzyme inhibitors alter the kinetic parameters of biochemical reactions. A Lineweaver-Burk plot flips kinetic data into a double-reciprocal form, where the y-intercept matches $1/V_{max}$ and the x-intercept matches -$1/K_m$, helping to identify the exact mechanism of inhibition.

Solution:

(a) Line A shows the uninhibited enzyme reaction. Line B shows the reaction in the presence of the inhibitor, showing an altered intersection on the horizontal axis.

(b) Both Line A and Line B intersect precisely at the same point (Y_1) on the vertical y-axis, meaning that $1/V_{max}$ is unchanged, and thus V_{max} remains constant.

(c) The x-intercept for Line B (X_2) shifts closer to the origin compared to Line A (X_1), meaning the value of -$1/K_m$ becomes less negative, which indicates that K_m has increased.

(d) An increase in K_m paired with an unchanged V_{max} is the hallmark kinetic definition of competitive inhibition, where the inhibitor binds reversibly to the active site.

(e) This competitive block can be overcome by increasing substrate concentrations, which explains why the maximal velocity (V_{max}) remains fully attainable at high substrate levels.

Final Answer: Line B represents competitive inhibition, where V_{max} remains unchanged and K_m is increased.

Answer: (A)

Go Back to Question 5

Q6.

Solution

Concept:

Classical homocystinuria is an autosomal recessive disorder caused by a functional deficiency of cystathionine β-synthase. This metabolic block prevents the conversion of homocysteine into cystathionine, leading to a toxic buildup of methionine and homocysteine in blood and urine.

Solution:

(a) The patient presents with marfanoid features, ectopia lentis, developmental delay, and thromboembolic risks, which clinically separates homocystinuria from true Marfan syndrome.

(b) Cystathionine β-synthase handles the transsulfuration pathway, converting homocysteine and serine into cystathionine while using pyridoxal phosphate (Vitamin B6) as an essential cofactor.

(c) In a subset of patients, the enzyme defect stems from a reduced binding affinity for its coenzyme, pyridoxine.

(d) Administering high therapeutic doses of pyridoxine can saturate the mutant enzyme and partially or fully restore its catalytic activity, reducing systemic homocysteine levels.

(e) This vitamin supplement helps clear the metabolic bottleneck, mitigating structural complications, though some variants remain unresponsive to B6 and require dietary restriction of methionine.

Final Answer: Pyridoxine (Vitamin B6)

Answer: (C)

Go Back to Question 6

Q7.

Solution

Concept:

Point mutations are single-base substitutions within a protein-coding gene sequence. Depending on how the altered nucleotide impacts the genetic code and triplet codons, these mutations can alter structural translation in different ways.

Solution:

(a) Comparing the original and mutated strands reveals a single nucleotide substitution where a Cytosine (C) is replaced by a Thymine (T) at the fourth position.

(b) Because the overall number of bases remains unchanged, the downstream reading frame is preserved, ruling out a frameshift mutation.

(c) A nonsense mutation generates a premature stop codon, which terminates translation early and truncates the protein rather than changing one amino acid property.

(d) A silent mutation alters the nucleotide sequence without changing the identity of the encoded amino acid due to the degeneracy of the genetic code.

(e) A missense mutation alters a single codon so that it specifies a different amino acid. Changing a hydrophilic residue to a hydrophobic one alters protein structure, as seen here.

Final Answer: Missense mutation

Answer: (D)

Go Back to Question 7

Q8.

Solution

Concept:

McArdle disease (Glycogen Storage Disease Type V) is caused by a congenital deficiency of skeletal muscle glycogen phosphorylase (myophosphorylase). This enzyme breaks down glycogen into glucose-1-phosphate to fuel anaerobic glycolysis during high-intensity exercise.

Solution:

(a) The clinical picture shows exercise intolerance, muscle cramping, and weakness brought on by intense exertion, which stems from an inability to access energy stores in muscle.

(b) Ischemic forearm testing confirms that blood lactate levels fail to rise during exercise because glycogen breakdown is blocked, preventing the downstream glycolysis that generates lactate.

(c) Muscle biopsy reveals an accumulation of glycogen within muscle cells because the initial step of degradation is inactive, but the glycogen structure itself remains completely normal.

(d) Glucose-6-phosphatase deficiency (Von Gierke) affects the liver and causes fasting hypoglycemia, but does not alter muscle glycogen metabolism or cause isolated exercise cramps.

(e) Debranching enzyme defects cause Type III glycogenosis, which produces structurally abnormal glycogen with short outer branches, unlike the normal glycogen observed in this patient.

Final Answer: Myophosphorylase

Answer: (B)

Go Back to Question 8

Q9.

Solution

Concept:

Transcription initiation in prokaryotes requires the core RNA polymerase enzyme to associate with a specific regulatory protein called the sigma (σ) factor. Together, they form the holoenzyme capable of recognizing and binding to promoter regions upstream of genes.

Solution:

(a) The prokaryotic promoter contains conserved sequences located at the -35 region and the -10 region (Pribnow box) relative to the transcription start site (+1).

(b) The core RNA polymerase ($\alpha_2\beta\beta'\omega$) has a general affinity for DNA but cannot pinpoint promoters or initiate transcription independently.

(c) The sigma factor directly recognizes and binds to the -35 and -10 promoter blocks, guiding the holoenzyme to form the closed promoter complex.

(d) Crucially, the sigma factor also mediates the local unwinding of the DNA double helix at the AT-rich -10 sequence, converting the closed complex into an open complex.

(e) A mutant polymerase that binds the promoter but cannot melt the strands to form the open complex lacks functional sigma factor activity.

Final Answer: Sigma (σ) factor

Answer: (C)

Go Back to Question 9

Q10.

Solution

Concept:

Scurvy is a nutritional deficiency caused by a severe lack of ascorbic acid (Vitamin C). It typically occurs in individuals with poor dietary intake, such as chronic alcoholics, and leads to defective collagen synthesis and compromised connective tissue integrity.

Solution:

(a) The clinical manifestations include swollen, bleeding gums, petechiae, perifollicular hemorrhages, and impaired wound healing, all of which reflect capillary fragility from weak collagen support.

(b) Collagen synthesis requires post-translational modification within the endoplasmic reticulum, where specific amino acid residues are hydroxylated to stabilize the triple helix structure.

(c) Prolyl hydroxylase and lysyl hydroxylase catalyze the addition of hydroxyl groups to proline and lysine residues, creating hydroxyproline and hydroxylysine.

(d) These enzymes require Vitamin C as a reducing agent to maintain iron in its active ferrous (Fe^{2+}) state during the enzymatic cycle.

(e) Without ascorbic acid, proline and lysine residues remain unhydroxylated, preventing collagen fibers from cross-linking properly, which causes the structural degradation seen in scurvy.

Final Answer: Proline and Lysine

Answer: (B)

Go Back to Question 10

Q11.

Solution

Concept:

Chargaff's rules provide foundational principles regarding the structural composition of double-stranded DNA. These biochemical laws state that DNA from any cell of any organism must exhibit a 1 to 1 stoichiometric ratio of pyrimidine and purine bases, which is a direct consequence of specific base-pairing mechanics.

Solution:

(a) Double-stranded genomic DNA consists of two antiparallel polynucleotide strands held together by hydrogen bonds between complementary nitrogenous bases.

(b) Adenine, a purine, always pairs exclusively with Thymine, a pyrimidine, forming two hydrogen bonds. Similarly, Guanine, a purine, pairs with Cytosine, a pyrimidine, forming three hydrogen bonds.

(c) Because every single purine base on one strand is dynamically paired with a matching pyrimidine base on the opposite strand, the total count of purines equals the total count of pyrimidines.

(d) Therefore, the concentration of Adenine equals Thymine, and the concentration of Guanine equals Cytosine within any standard double-stranded molecule.

(e) This mathematical relationship dictates that exactly 50% of the total bases in the genome are purines (X) and exactly 50% are pyrimidines (Y), regardless of variations in the total GC-content between different biological organisms.

Final Answer: X = 50%, Y = 50%

Answer: (C)

Go Back to Question 11

Q12.

Solution

Concept:

Cori disease, also classified as Glycogen Storage Disease Type III, is an inherited metabolic disorder caused by a deficiency of the glycogen debranching enzyme. This enzymatic defect restricts full glycogen degradation, leading to structural abnormalities in stored glycogen and systemic metabolic manifestations.

Solution:

(a) The glycogen debranching enzyme possesses two distinct catalytic properties on a single polypeptide chain: oligo-α-1,4→α-1,4-glucan transferase and amylo-α-1,6-glucosidase.

(b) When glycogen phosphorylase removes glucose units from a glycogen molecule, it stops four residues away from a branch point, leaving a structure known as a limit dextrin.

(c) In this patient, the transferase component functions normally to shift three residues, but the α-1,6-glucosidase activity is missing, preventing the cleavage of the final single glucose residue at the branch.

(d) This structural block causes an accumulation of abnormal glycogen containing short, outer branches within hepatic and muscle tissues, causing hepatomegaly and hypotonia.

(e) Because glycogen degradation is stalled at outer branches during fasting, glucagon cannot trigger glucose release, leading to severe fasting hypoglycemia, while postprandial glucose disposal remains unimpeded.

Final Answer: α-1,6-glucosidase (Debranching enzyme)

Answer: (B)

Go Back to Question 12

Q13.

Solution

Concept:

Eukaryotic pre-mRNA undergoes extensive post-transcriptional modifications in the nucleus before it can be exported to the cytoplasm for translation. The initial modification is 5' capping, which is critical for protecting the transcript from exonuclease degradation and facilitating ribosome assembly.

Solution:

(a) The capping process begins immediately after transcription initiation, when the nascent pre-mRNA transcript reaches a length of approximately twenty to thirty nucleotides.

(b) An enzyme complex removes a phosphate from the 5' end of the transcript and then transfers a guanosine monophosphate molecule from GTP to the remaining diphosphate.

(c) This enzymatic step creates an unusual 5' to 5' triphosphate linkage between the new guanosine residue and the initial nucleotide of the mRNA molecule.

(d) Subsequently, methyltransferase enzymes transfer a methyl group from S-adenosylmethionine to the nitrogen at position 7 of the terminal guanine ring, forming a 7-methylguanosine cap.

(e) This unique 5' to 5' structural linkage cannot be recognized by standard cellular 5' exonucleases, effectively shielding the mature mRNA from premature degradation inside the cytoplasm.

Final Answer: A 7-methylguanosine residue attached via an unusual 5' to 5' triphosphate linkage.

Answer: (B)

Go Back to Question 13

Q14.

Solution

Concept:

Lesch-Nyhan syndrome is a severe X-linked recessive metabolic disorder caused by a complete deficiency of hypoxanthine-guanine phosphoribosyltransferase. This biochemical defect disrupts the purine salvage pathway, shifting the metabolic balance toward excessive de novo purine synthesis and degradation.

Solution:

(a) The purine salvage pathway normally recycles free purine bases, converting hypoxanthine into IMP and guanine into GMP using phosphoribosyl pyrophosphate as a ribose donor.

(b) When HGPRT is completely deficient, these free purine bases cannot be salvaged and are instead channeled directly into the degradation pathway, forming large amounts of uric acid.

(c) Furthermore, because PRPP is not consumed by the salvage pathway, intracellular PRPP levels rise significantly and stimulate the rate-limiting step of de novo purine synthesis.

(d) The combined increase in de novo purine production and base degradation leads to severe hyperuricemia, causing orange sodium urate crystals in diapers and uric acid nephropathy.

(e) This metabolic imbalance causes characteristic neurological features, including severe cognitive impairment, choreoathetosis, and compulsive self-mutilation behavior like biting of lips and fingers.

Final Answer: Hypoxanthine-guanine phosphoribosyltransferase (HGPRT)

Answer: (B)

Go Back to Question 14

Q15.

Solution

Concept:

Acute Intermittent Porphyria is an autosomal dominant metabolic disorder caused by a partial deficiency of porphobilinogen deaminase, an enzyme in the heme biosynthetic pathway. This partial block causes a toxic accumulation of early porphyrin intermediates during metabolic triggers.

Solution:

(a) The pathway for heme synthesis begins with the condensation of glycine and succinyl-CoA to form δ-aminolevulinic acid, which then dimerizes to form porphobilinogen.

(b) Porphobilinogen deaminase catalyzes the polymerization of four porphobilinogen molecules into a linear tetrapyrrole intermediate known as hydroxymethylbilane.

(c) A deficiency in porphobilinogen deaminase creates a pathway bottleneck, causing a massive upstream accumulation of δ-aminolevulinic acid and porphobilinogen in blood and tissues.

(d) These early precursors are neurotoxic, leading to the clinical manifestations of the disease, which include severe episodic abdominal pain, psychiatric symptoms, and peripheral neuropathies.

(e) When urine containing excess porphobilinogen is exposed to light and air, it spontaneously polymerizes into porphobilin, turning the urine a dark wine-red color.

Final Answer: Porphobilinogen deaminase

Answer: (B)

Go Back to Question 15

Q16.

Solution

Concept:

Autosomal dominant inheritance patterns dictate that a single copy of a mutant allele is sufficient to cause the phenotypic expression of a disorder. These genes are located on non-sex chromosomes, meaning transmission patterns occur independently of parental or fetal sex.

Solution:

(a) The husband is diagnosed with a well-documented autosomal dominant disorder, meaning he carries at least one copy of the dominant mutant allele in his genome.

(b) Because his father was completely unaffected by the condition, his father must be homozygous normal and could not have passed a dominant mutant allele to his son.

(c) The husband must have inherited his single mutant allele from his heterozygous, symptomatic mother, confirming that the husband is heterozygous for the disorder.

(d) The wife is genotypically normal, meaning she carries two normal recessive alleles and will exclusively contribute a normal allele to all future offspring.

(e) During meiosis in the heterozygous husband, there is a 50% chance of distributing the mutant allele into any given sperm cell, resulting in a 50% probability of transmission.

Final Answer: 50%

Answer: (C)

Go Back to Question 16

Answer Key

Q	Ans	Q	Ans	Q	Ans	Q	Ans	Q	Ans
1	A	2	B	3	B	4	D	5	A
6	C	7	D	8	B	9	C	10	B
11	C	12	B	13	B	14	B	15	B
16	C								