NEET PG Biochemistry Sample Paper-5

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 45-year-old chronic alcoholic presents with severe abdominal pain radiating to the back, lactic acidosis, and profound hypoglycemia. Biochemical evaluation reveals a massively elevated cytosolic $NADH/NAD^+$ ratio in hepatocytes. Which of the following metabolic conversions is directly inhibited by this altered redox state, contributing heavily to the failure of gluconeogenesis?

(A) Lactate to Pyruvate

(B) Glyceraldehyde 3-phosphate to 1,3-Bisphosphoglycerate

(C) Malate to Oxaloacetate in mitochondria

(D) Pyruvate to Acetyl-CoA

Q2. A newborn is evaluated for failure to thrive, persistent metabolic acidosis, and a distinct musty odor. A localized block in the branched-chain α-keto acid dehydrogenase (BCKDH) complex is diagnosed. The functional assembly of this macromolecular complex relies on a structural core. Which subunit forms the symmetric structural transacylase core catalyst of this specific complex?

(A) E1 α subunit

(B) E1 β subunit

(C) E2 subunit

(D) E3 subunit

Q3. A laboratory synthesizes a novel non-hydrolyzable analog of Palmitoyl-CoA where the sulfur atom is replaced by a methylene (CH_2) group. When added to an isolated mitochondrial matrix assay containing active carnitine palmitoyltransferase II (CPT-II), which specific process or intermediate formation is completely halted?

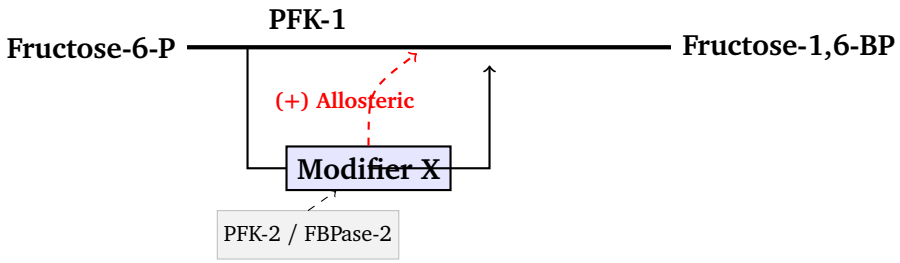
(A) Translocation of Carnitine through the inner membrane

(B) Regeneration of intra-mitochondrial Free Carnitine

(C) Intermembrane condensation of Acyl-Carnitine

(D) Binding of Malonyl-CoA to CPT-I

Q4. An experimental oncology drug candidate alters the regulatory kinetics of glycolysis in hepatoma cells. A continuous-flow metabolic flux analysis map outlines the precise allosteric integration loop of Phosphofructokinase-1 (PFK-1) below. Identify the exact regulatory modifier node labeled as **Modifier X** which bypasses ATP inhibition:



(A) Fructose-2,6-Bisphosphate

(B) Glucose-1,6-Bisphosphate

(C) Cyclic AMP (cAMP)

(D) Citrate

Q5. During prolonged starvation, the brain adapts to utilize ketone bodies. A key mitochondrial matrix enzyme is required to activate acetoacetate into acetoacetyl-CoA by transferring a coenzyme A moiety from a TCA cycle intermediate. This crucial enzyme, which is notably absent in the liver, is:

(A) Thiolase

(B) HMG-CoA Lyase

(C) β-Hydroxybutyrate Dehydrogenase

(D) Succinyl-CoA:3-Ketoacid CoA Transferase

Q6. A patient presents with severe exercise-induced muscle cramps and myoglobinuria. Muscle biopsy shows massive glycogen accumulation with normal structure. Biochemical testing demonstrates complete lack of a glycolytic enzyme that is regulated by reversible phosphorylation. Which statement correctly characterizes this enzyme in its most active conformation?

(A) Phosphorylated glycogen synthase (b form)

(B) Dephosphorylated glycogen phosphorylase (b form)

(C) Phosphorylated glycogen phosphorylase (a form)

(D) Dephosphorylated phosphofructokinase-1

Q7. Researchers studying mammalian DNA replication isolate a mutant cell line where Okazaki fragments are synthesized normally, but show an inability to remove the RNA primers at the $5'$ end of the lagging strand fragments. Which eukaryotic enzymatic assembly or specialized endonuclease is primary defective in these cells?

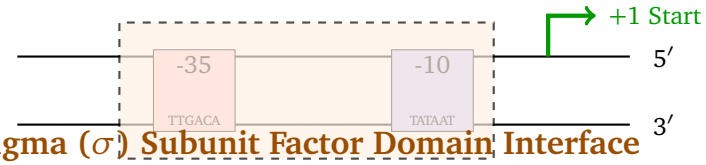
(A) DNA Polymerase γ

(B) Flap Endonuclease 1 (FEN1)

(C) DNA Ligase IV

(D) Topoisomerase II α

Q8. A molecular biology diagnostic suite maps the structural interactions inside a prokaryotic transcription initiation open ternary complex. Review the topology diagram below showing the specific interaction points of the RNA Polymerase Holoenzyme with the core promoter element:



Which specific sub-region or domain of the bacterial sigma (σ^{70}) factor directly binds and stabilizes the melted, single-stranded nucleotide state within the crucial −10 Pribnow box sequence region?

(A) Domain Region 1.1

(B) Domain Region 2.4 / 2.3

(C) Domain Region 3.0

(D) Domain Region 4.2

Q9. A patient with systemic lupus erythematosus (SLE) produces autoantibodies against Small Nuclear Ribonucleoproteins (snRNPs). These complexes are critical components of the spliceosome. During pre-mRNA splicing, which precise molecular event is driven directly by the base-pairing interaction of the U1 snRNA?

(A) Recognition and binding of the 5′ splice site junction

(B) Attack of the branch point Adenosine on the 5′ splice site

(C) Binding to the poly-pyrimidine tract upstream of the 3′ site

(D) Ligation of the upstream and downstream exons

Q10. An in vitro translation assay is prepared using synthetic mRNA containing repeating 5′-AUG-3′ codons. A novel antibiotic is introduced that selectively prevents the entry of aminoacyl-tRNA molecules into the aminoacyl (A) site of the ribosomal structure by blocking Elongation Factor Tu (EF-Tu) GTP hydrolysis. Which molecule will remain trapped in the ribosomal peptidyl (P) site?

(A) Peptidyl-tRNA

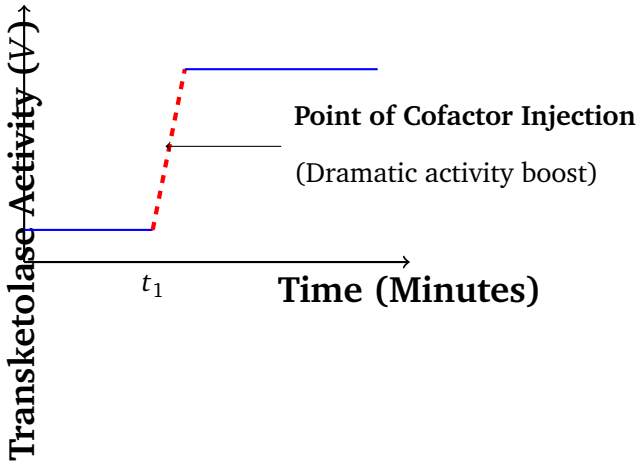
(B) Uncharged tRNA

(C) Formylmethionyl-tRNA (or Methionyl-tRNA)

(D) Releasing Factor 1 (RF-1)

Q11. The enzymatic activity profile of a pure modern line of human erythrocyte transketolase is mapped continuously during an analytical incubation

challenge. The line profile tracking below displays the reaction velocity variations observed after adding an active cofactor derived from a water-soluble B-complex vitamin:



A clinical deficiency in the structural precursor of this critical injected organic cofactor causes which classic presentation combination?

(A) Dermatitis, Dementia, Diarrhea

(B) Ophthalmoplegia, Ataxia, Confusion

(C) Megaloblastic anemia with subacute spinal cord degeneration

(D) Cheilosis, glossitis, and localized corneal vascularization

Q12. An investigator performs enzyme kinetics modeling on a newly isolated bacterial transpeptidase. In the presence of a competitive active-site inhibitor, how are the calculated Michaelis constant (K_m) and the maximum reaction velocity (V_{max}) values altered compared to the uninhibited controls?

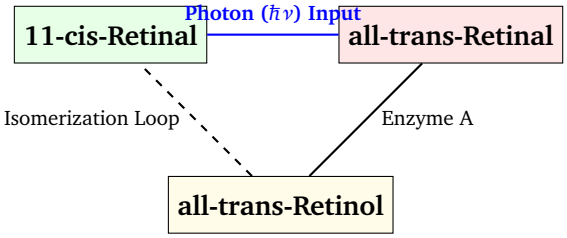
(A) K_m decreases; V_{max} remains completely unchanged

(B) K_m increases; V_{max} remains completely unchanged

(C) K_m remains unchanged; V_{max} decreases markedly

(D) Both K_m and V_{max} decrease proportionally

Q13. The multi-step mechanical scheme below showcases the structural isomerization coordinates of the prosthetic retinal chromophore within the rhodopsin visual cycle network:



Which specific enzyme is responsible for converting the unstable **all-trans-Retinal** molecule back into its functional storage reduction compound form labeled as **all-trans-Retinol** within the outer segment disk spaces?

(A) Retinal Reductase (RDH)

(B) Lecithin-Retinol Acyltransferase (LRAT)

(C) RPE65 Isomerohydrolase

(D) Retinyl Ester Hydrolase

Q14. A pedigree chart of a large multi-generational family reveals an unusual pattern of inheritance. Affected fathers pass a profound skeletal deformity trait to all of their daughters, but to none of their sons. Affected heterozygous mothers pass the phenotype to roughly 50% of both their sons and daughters. What is the most likely mode of genetic transmission?

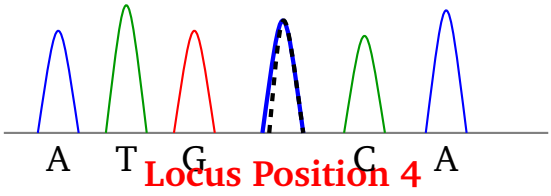
(A) Autosomal Dominant with sex-linked penetrance

(B) X-linked Recessive

(C) X-linked Dominant

(D) Mitochondrial Maternal Cytoplasmic Inheritance

Q15. A clinical genomics facility runs a high-throughput automated Sanger sequencing fragment separation assay. The fluorescence output trace from a capillary electrophoresis run corresponding to a specific targeted heterozygous hot-spot genomic mutation locus is shown below:



During standard Sanger dideoxy DNA sequencing reaction assembly, what specific chemical property of the added dideoxynucleoside triphosphates (ddNTPs) forces the absolute termination of further primer extension cascades?

(A) Absence of a 2′-hydroxyl group (—OH) on the ribose ring structure

(B) Presence of an blocking azide chemical group at the 5′-phosphate node

(C) Absence of a 3′-hydroxyl group (—OH) on the ribose ring structure

(D) Methylation at the N-7 position of the purine nitrogenous bases

Q16. A child with short stature, intellectual disability, and a history of severe infantile hypotonia exhibits an microdeletion on the paternal chromosome 15q11-q13 locus. If the exact same structural microdeletion zone were inherited instead from the biological mother, a totally distinct phenotypic syndrome (Angelman Syndrome) would occur. This distinct outcome is regulated by which mechanism?

(A) Genomic Imprinting via differential DNA methylation

(B) Alternative exon splicing variations

(C) Variable trinucleotide repeat expansion loops

(D) Dosage compensation via X-chromosome inactivation

Detailed Solutions

Q1.

Solution

Concept: Metabolism of high amounts of ethanol by hepatic alcohol dehydrogenase and aldehyde dehydrogenase utilizes NAD^+ and generates massive amounts of cytosolic NADH. This significantly increases the cytosolic NADH/NAD^+ ratio, shifting the equilibrium of reversible, NAD^+/NADH-dependent dehydrogenase reactions backward to suppress gluconeogenesis.

Solution:

Let's trace the biochemical impact of an elevated cytosolic NADH/NAD^+ ratio on gluconeogenic substrates:

(a) Pyruvate must be generated from lactate to enter gluconeogenesis. However, the high cytosolic NADH drives the lactate dehydrogenase reaction in reverse:

$$\text{Pyruvate} + \text{NADH} + H^+ \rightarrow \text{Lactate} + NAD^+$$

This **directly inhibits the conversion of Lactate to Pyruvate**, trapping lactate in the cytosol, causing severe lactic acidosis, and depleting the primary substrate required for gluconeogenesis.

(b) Similarly, the conversion of malate to oxaloacetate in the cytosol is inhibited because it requires NAD^+, further starving the gluconeogenic pathway.

(c) The reaction from glyceraldehyde 3-phosphate to 1,3-bisphosphoglycerate is a glycolytic step requiring NAD^+, while the conversion of pyruvate to acetyl-CoA is an irreversible mitochondrial matrix reaction catalyzed by PDH, which is not a component of gluconeogenesis.

Final Answer: Lactate to Pyruvate

Answer: (A)

Go Back to Question 1

Q2.

Solution

Concept: The branched-chain α-keto acid dehydrogenase (BCKDH) complex is a multi-subunit mitochondrial enzyme network that metabolizes the branched-chain amino acids leucine, isoleucine, and valine. Its structural organization mirrors other α-keto acid dehydrogenase complexes like pyruvate dehydrogenase (PDH) and α-ketoglutarate dehydrogenase (α-KGDH).

Solution:

Let's analyze the structural subunit organization of these macromolecular complexes:

(a) **E_1 subunit (α and β chains):** Acts as a thiamine pyrophosphate (TPP)-dependent decarboxylase that carries out the initial decarboxylation of the α-keto acid.

(b) **E_2 subunit (Dihydrolipooyl transacylase):** Forms the highly symmetric, multi-mer **structural transacylase core catalyst** of the complex. It contains covalently bound lipoic acid residues that accept the acyl group from E_1 and transfer it directly to Coenzyme A (CoA).

(c) **E_3 subunit (Dihydrolipooyl dehydrogenase):** Uses flavin adenine dinucleotide (FAD) and NAD^+ to regenerate the oxidized lipoamide arm on the E_2 core and is shared among all three α-keto acid dehydrogenase complexes.

Final Answer: E2 subunit

Answer: (C)

Go Back to Question 2

Q3.

Solution

Concept: The carnitine shuttle system transports long-chain fatty acids across the inner mitochondrial membrane for β-oxidation. Carnitine palmitoyltransferase II (CPT-II) is located on the matrix side of the inner mitochondrial membrane and catalyzes the terminal step of this shuttle.

Solution:

Let's evaluate the normal enzymatic reaction of CPT-II and the impact of the non-hydrolyzable analog:

(a) In the mitochondrial matrix, CPT-II normally receives acyl-carnitine and transfers the fatty acyl chain back onto intra-mitochondrial Coenzyme A (CoASH), yielding palmitoyl-CoA and free carnitine:

$$\text{Palmitoyl-carnitine} + \text{CoASH} \xrightarrow{\text{CPT-II}} \text{Palmitoyl-CoA} + \text{Free Carnitine}$$

(b) The laboratory synthesized a non-hydrolyzable analog of the product, Palmitoyl-CoA, where the thioester sulfur atom is replaced by a non-reactive methylene ($-CH_2-$) group.

(c) This structural modification prevents chemical cleavage or reverse substitution at the acyl bond, locking the enzyme complex. As a direct consequence, the catalytic cycle stalls, and the **regeneration of intra-mitochondrial Free Carnitine** is completely halted.

Final Answer: Regeneration of intra-mitochondrial Free Carnitine

Answer: (B)

Go Back to Question 3

Q4.

Solution

Concept: Phosphofructokinase-1 (PFK-1) is the primary rate-limiting and committed enzyme of glycolysis. It is tightly regulated by the energy charge of the cell, being allosterically inhibited by high levels of ATP and citrate, and activated by AMP and specific hexose bisphosphates.

Solution:

Let's identify the regulatory modifier pathway shown in the dynamic metabolic flow map:

(a) The bifunctional enzyme PFK-2/FBPase-2 controls glycolysis flux by synthesizing and degrading a powerful local side-shunt signaling molecule.

(b) When the PFK-2 domain is active, it phosphorylates fructose-6-phosphate to produce **Fructose-2,6-Bisphosphate (Modifier X)**.

(c) Fructose-2,6-bisphosphate binds to PFK-1 as its most potent **allosteric activator** (+). This binding shifts the conformational state of PFK-1 to increase its affinity for fructose-6-phosphate and effectively **bypasses the physiological inhibitory feedback** caused by high concentrations of ATP.

Final Answer: Fructose-2,6-Bisphosphate

Answer: (A)

Go Back to Question 4

Q5.

Solution

Concept: Ketone bodies synthesized by hepatocytes during prolonged starvation serve as a critical water-soluble fuel source for extrahepatic tissues such as the brain. The liver lacks the ability to consume its own synthesized ketone bodies due to the intentional absence of a specific activation enzyme.

Solution:

Let's analyze the catabolic breakdown pathway of acetoacetate in peripheral target tissues:

(a) To enter the Citric Acid Cycle, acetoacetate must first be activated into acetoacetyl-CoA within the mitochondrial matrix.

(b) This activation is achieved by transferring a CoA molecule from succinyl-CoA via the enzyme **Succinyl-CoA:3-Ketoacid CoA Transferase** (commonly known as thiophorase):

$$\text{Acetoacetate} + \text{Succinyl-CoA} \rightarrow \text{Acetoacetyl-CoA} + \text{Succinate}$$

(c) Because hepatocytes completely lack thiophorase expression, the liver cannot oxidize acetoacetate, guaranteeing that ketone bodies are exported to the bloodstream to meet the energetic demands of extrahepatic tissues like the cerebral cortex.

Final Answer: Succinyl-CoA:3-Ketoacid CoA Transferase

Answer: (D)

Go Back to Question 5

Q6.

Solution

Concept: McArdle disease (Glycogen Storage Disease Type V) is caused by a genetic deficiency in muscle glycogen phosphorylase (myophosphorylase). This key regulatory enzyme breaks down internal skeletal muscle glycogen stores into glucose-1-phosphate during high-intensity anaerobic exercise.

Solution:

Let's evaluate the conformational regulation of glycogen phosphorylase via covalent modification:

(a) Glycogen phosphorylase exists in two interconvertible structural states: a less active, dephosphorylated '*b*' form, and a highly active, **phosphorylated '*a*' form**.

(b) During muscle activity or epinephrine stimulation, phosphorylase kinase is activated and transfers a phosphate group onto specific serine residues of glycogen phosphorylase.

(c) This covalent modification locks the enzyme into its **phosphorylated glycogen phosphorylase (*a* form)** conformation, which is highly active and independent of local AMP allosteric concentrations, accelerating glycogenolysis to fuel anaerobic work.

Final Answer: Phosphorylated glycogen phosphorylase (*a* form)

Answer: (C)

Go Back to Question 6

Q7.

Solution

Concept: During eukaryotic DNA replication, lagging strand synthesis is discontinuous and produces short Okazaki fragments. Each fragment begins with an RNA primer synthesized by the DNA polymerase α-primase complex, which must be excised before the DNA segments can be joined into a continuous strand.

Solution:

Let's look at the processing steps and enzymes involved in removing eukaryotic RNA primers:

(a) When DNA polymerase δ encounters the $5'$ end of an adjacent Okazaki fragment, it displaces the RNA primer and a short segment of DNA, creating a single-stranded "$5'$ flap" structure.

(b) This specific structural flap is recognized and cleaved near its base by **Flap Endonuclease 1 (FEN1)**, a specialized structure-specific endonuclease.

(c) A defect or mutation in **FEN1** prevents the excision of these RNA primers from the lagging strand, stalling maturation and preventing DNA ligase from sealing the genomic backbone. In prokaryotes, this primer removal function is handled instead by the $5' \rightarrow 3'$ exonuclease activity of DNA Polymerase I.

Final Answer: Flap Endonuclease 1 (FEN1)

Answer: (B)

Go Back to Question 7

Q8.

Solution

Concept: In prokaryotes, transcription initiation requires the core RNA polymerase to bind a specialized sigma (σ) factor to form the functional holoenzyme. The standard sigma factor (σ^{70}) contains specific structural domains that recognize and bind core promoter sequences upstream of the transcription start site (+1).

Solution:

Let's analyze the structural interactions of the σ^{70} factor domains with the promoter:

(a) **Domain Region 4.2:** Forms a helix-turn-helix motif that binds directly to the −35 core promoter consensus sequence (TTGACA), anchoring the polymerase holoenzyme to the DNA.

(b) **Domain Region 2.4 / 2.3:** Interacts directly with the −10 **Pribnow box sequence region (TATAAT)**.

(c) Specifically, alpha-helical structures within **Domain Region 2.3 and 2.4** contain aromatic amino acid residues that flip out and intercalate into the DNA strands. This interaction stabilizes the single-stranded template state during local melting, driving the transition from a closed promoter complex to an open transcription bubble.

Final Answer: Domain Region 2.4 / 2.3

Answer: (B)

Go Back to Question 8

Q9.

Solution

Concept: Pre-mRNA splicing is mediated by the spliceosome, a macromolecular complex composed of small nuclear ribonucleoproteins (snRNPs: U1, U2, U4, U5, and U6) and auxiliary proteins. The precision of intron removal depends on complementary base-pairing between the small nuclear RNA (snRNA) components and conserved sequences on the pre-mRNA transcript.

Solution:

Let's trace the initial sequence of events during spliceosome assembly:

(a) The splicing cycle begins when the **U1 snRNP** binds to the pre-mRNA molecule. Specifically, the 5′ terminus of the U1 snRNA contains a highly conserved sequence that forms complementary base pairs with the 5′ **splice site junction** (the conserved GU consensus sequence at the 5′ intron-exon boundary).

(b) This primary base-pairing interaction targets the spliceosome to the intron border, defining the precise alignment for subsequent steps.

(c) Following U1 binding, U2 snRNP binds to the internal branch point sequence (containing the reactive adenosine residue), while the U4/U6·U5 tri-snRNP complex joins later to catalyze the transesterification reactions.

Final Answer: Recognition and binding of the 5′ splice site junction

Answer: (A)

Go Back to Question 9

Q10.

Solution

Concept: Translation elongation is a cyclical process coordinated by elongation factors. Elongation Factor Tu (EF-Tu) binds an aminoacyl-tRNA and a molecule of GTP, delivering this ternary complex directly into the vacant ribosomal Aminoacyl (A) site.

Solution:

Let's analyze the impact of blocking EF-Tu GTP hydrolysis on translation initiation and elongation:

(a) During initiation on the synthetic repeating AUG template, the initiator **formylmethionyl-tRNA (or methionyl-tRNA)** binds directly into the ribosomal **Peptidyl (P) site**. At this stage, the Aminoacyl (A) site remains vacant, waiting for the next matching aminoacyl-tRNA.

(b) The introduced antibiotic blocks EF-Tu GTP hydrolysis, which is required for EF-Tu to release the aminoacyl-tRNA and exit the ribosome. As a result, no new aminoacyl-tRNA molecules can successfully enter or bind the open A site.

(c) Because no incoming tRNA enters the A site to accept the peptide chain via peptidyl transferase activity, the initiation complex stalls. The original **formylmethionyl-tRNA (or methionyl-tRNA)** remains trapped in the **P site**.

Final Answer: Formylmethionyl-tRNA (or Methionyl-tRNA)

Answer: (C)

Go Back to Question 10

Q11.

Solution

Concept: Transketolase is a key enzyme in the non-oxidative branch of the pentose phosphate pathway, transferring two-carbon ketol groups between sugar phosphates. It requires thiamine pyrophosphate (TPP), an active coenzyme derived from the water-soluble B-complex vitamin thiamine (Vitamin B_1), as an essential structural cofactor.

Solution:

Let's link the analytical activity profile to its corresponding clinical presentation:

(a) The graph shows that baseline transketolase activity is low but spikes dramatically ($> 15\%$) at t_1 upon adding the active cofactor. This significant increase in enzyme activity upon TPP addition confirms a clinical deficiency in its precursor, **thiamine (Vitamin B_1)**.

(b) A severe deficiency in thiamine alters glucose utilization in high-metabolic tissues, resulting in **Wernicke-Korsakoff syndrome**. This neurological condition is clinically characterized by the classic triad of **ophthalmoplegia** (paralysis of ocular muscles), **ataxia** (loss of motor coordination), and global **confusion**.

(c) Dermatitis, dementia, and diarrhea describe pellagra (Niacin/B_3 deficiency); megaloblastic anemia with subacute spinal cord degeneration describes Vitamin B_{12} deficiency; and cheilosis and corneal vascularization indicate riboflavin (B_2) deficiency.

Final Answer: Ophthalmoplegia, Ataxia, Confusion

Answer: (B)

Go Back to Question 11

Q12.

Solution

Concept: Competitive inhibitors possess a structural resemblance to the native substrate and compete directly for binding at the active site of the free enzyme. This type of inhibition can be overcome by increasing the substrate concentration.

Solution:

Let's analyze how a competitive inhibitor affects Michaelis-Menten kinetic parameters:

(a) **Maximum Velocity (V_{max}):** Because high concentrations of substrate can displace the inhibitor from the active site, the enzyme can still achieve its maximum catalytic rate at saturation. Thus, V_{max} **remains completely unchanged**.

(b) **Michaelis Constant (K_m):** Because the inhibitor competes with the substrate for the active site, a higher concentration of substrate is required to reach half of the maximum velocity ($1/2V_{max}$). This means the apparent K_m **increases** (αK_m).

Final Answer: K_m increases; V_{max} remains completely unchanged

Answer: (B)

Go Back to Question 12

Q13.

Solution

Concept: The visual cycle involves the continuous regeneration of the light-sensitive chromophore 11-cis-retinal. When a photon ($\hbar\nu$) strikes rhodopsin in the rod outer segments, 11-cis-retinal undergoes photoisomerization to the unstable, active all-trans-retinal conformation, triggering the visual phototransduction cascade.

Solution:

Let's trace the enzymatic reduction step shown in the visual cycle network:

(a) Following its photoisomerization, all-trans-retinal dissociates from the opsin apoprotein into the outer segment disk space.

(b) To be recycled, it must first be reduced into a stable storage form. This specific conversion of all-trans-retinal to all-trans-retinol (Enzyme A) is catalyzed by **Retinal Reductase**, also known as **all-trans-Retinol Dehydrogenase (RDH)**, utilizing NADPH as an electron donor.

(c) The resulting all-trans-retinol then travels to the retinal pigment epithelium (RPE), where it is esterified by LRAT and converted back into 11-cis-retinal by the isomerohydrolase RPE65.

Final Answer: Retinal Reductase (RDH)

Answer: (A)

Go Back to Question 13

Q14.

Solution

Concept: X-linked dominant inheritance is a mode of genetic transmission where a single mutant allele on the X chromosome is sufficient to express the clinical phenotype in both males and females.

Solution:

Let's analyze the transmission lines described in the pedigree profile:

(a) **Affected Fathers:** A father passes his single Y chromosome to all of his sons and his single X chromosome to all of his daughters. Because affected fathers pass the trait to **all of their daughters but none of their sons**, the mutant allele must reside exclusively on the X chromosome. This rules out autosomal transmission.

(b) **Affected Mothers:** A heterozygous mother (X^MX) passes either her mutant X or her normal X chromosome to her offspring with equal probability. This explains why she transmits the phenotype to **approximately** 50% **of both her sons and daughters**.

(c) This inheritance pattern fulfills the criteria for an **X-linked Dominant** trait. X-linked recessive transmission is incorrect because an affected father cannot have normal sons while passing a recessive trait to all daughters without the mother being a carrier.

Final Answer: X-linked Dominant

Answer: (C)

Go Back to Question 14

Q15.

Solution

Concept: Sanger sequencing utilizes a mixture of standard deoxynucleoside triphosphates (dNTPs) and modified dideoxynucleoside triphosphates (ddNTPs) to determine DNA sequences. The incorporation of a ddNTP into a growing DNA strand stops further elongation.

Solution:

Let's analyze the chemical structure of ddNTPs that leads to chain termination:

(a) Standard dNTPs possess a hydroxyl group (—OH) on the $3'$ carbon of the ribose ring, which is required to form a phosphodiester bond with the incoming $5'$-phosphate group of the next nucleotide.

(b) Dideoxynucleoside triphosphates (ddNTPs) are modified to lack this specific $3'$**-hydroxyl group (—OH) on the ribose ring structure** (possessing a hydrogen atom instead at both the $2'$ and $3'$ positions).

(c) When DNA polymerase incorporates a ddNTP, the **absence of the** $3'$**-OH group** prevents the formation of the next phosphodiester bond, terminating primer extension and generating fragments of varying lengths that can be separated by capillary electrophoresis.

Final Answer: Absence of a $3'$-hydroxyl group ($-$OH) on the ribose ring structure

Answer: (C)

Go Back to Question 15

Q16.

Solution

Concept: Genomic imprinting is an epigenetic mechanism where genes are expressed in a parent-of-origin-specific manner. This allele-specific expression is regulated by epigenetic modifications established during gametogenesis.

Solution:

Let's analyze the differential presentation of the 15q11-q13 microdeletion:

(a) The 15q11-q13 chromosomal locus contains genes that are differentially silenced depending on whether they are inherited from the mother or the father. This tissue-specific silencing is maintained by **Genomic Imprinting via differential DNA methylation**.

(b) In normal individuals, the *Prader-Willi* gene (SNRPN) is active only on the paternal chromosome, while the *Angelman* gene (UBE3A) is active only on the maternal chromosome.

(c) If a child inherits the microdeletion from the **father**, they lack the functional paternal alleles, resulting in Prader-Willi Syndrome. If the exact same deletion is inherited from the **mother**, the functional maternal copies are lost, resulting in Angelman Syndrome.

Final Answer: Genomic Imprinting via differential DNA methylation

Answer: (A)

Go Back to Question 16

Answer Key

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