# NEET PG Biochemistry Sample Paper-7

**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 studying DNA replication in a mammalian cell line. She isolates a mutant strain that synthesis lagging strand fragments but fails to join them together, leading to accumulation of unsealed Okazaki fragments. The enzyme defective in this cell line normally catalyzes the formation of which bond?

(A) $3' \rightarrow 5'$ Phosphodiester bond using ATP

(B) $5' \rightarrow 3'$ Phosphodiester bond using GTP

(C) N-Glycosidic bond using CTP

(D) Hydrogen bonds between complementary bases

**Q2.** A 48-year-old chronic alcoholic presents with severe ataxia, confusion, and ophthalmoplegia. Laboratory evaluation reveals a marked deficiency in an enzyme that requires a cofactor derived from water-soluble vitamins. Which of the following biochemical reactions is directly impaired in this patient?

(A) Conversion of Succinate to Fumarate

(B) Conversion of $\alpha$-ketoglutarate to Succinyl-CoA

(C) Conversion of Malate to Oxaloacetate

(D) Conversion of Pyruvate to Oxaloacetate

**Q3.** A 4-year-old child presents with global developmental delay, self-mutilating behavior such as lip-biting, and hyperuricemia. The disease is inherited in an X-linked recessive pattern. The enzyme deficient in this condition is normally involved in which pathway?

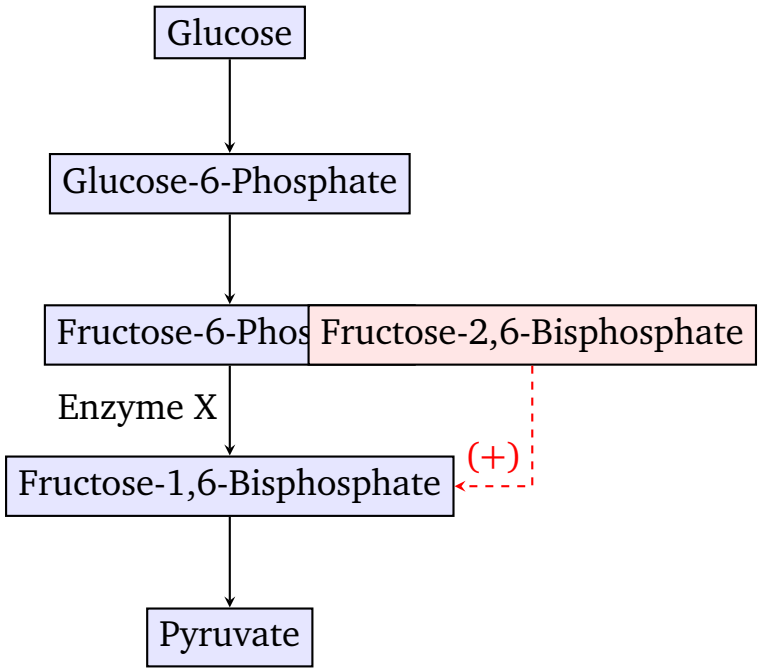
(A) De novo Purine synthesis

(B) Purine salvage pathway

(C) Pyrimidine salvage pathway

(D) De novo Pyrimidine synthesis

**Q4.** Consider the following metabolic pathway showing the regulatory intersections of carbohydrate metabolism:




An allosteric activator for the step regulated by "Enzyme X" is Fructose-2,6-bisphosphate. Which of the following hormonal states significantly increases the concentration of this intracellular regulator?

(A) High Glucagon/Insulin ratio

(B) Elevated Epinephrine during fasting

(C) High Insulin/Glucagon ratio

(D) Elevated Cortisol with prolonged starvation

**Q5.** A 35-year-old male is evaluated for dark urine that turns black upon standing, along with progressive bluish-black pigmentation of his sclera and ear cartilage. The defective enzyme in this patient is responsible for the catabolism of which of the following pairs of amino acids?

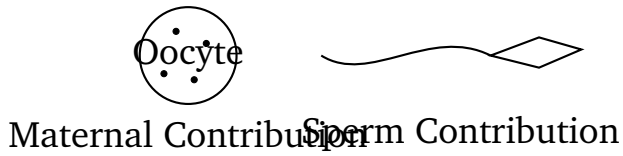
(A) Phenylalanine and Tyrosine

(B) Tryptophan and Methionine

(C) Lysine and Leucine

(D) Valine and Isoleucine

**Q6.** A pedigree analysis of a family with a history of progressive muscle weakness and cardiomyopathy reveals that an affected female transmits the disease to all of her children, whereas an affected male does not transmit the disease to any of his offspring. This transmission pattern matches the genetic architecture shown below:




Which of the following properties is most typically associated with the mutations responsible for this inheritance pattern?

(A) High rate of mitotic crossover

(B) Heteroplasmy and variable expressivity

(C) Genomic imprinting during gametogenesis

(D) Allelic exclusion in somatic tissues

**Q7.** During fatty acid synthesis, the condensation of acetyl-CoA and malonyl-CoA units occurs on the Fatty Acid Synthase multienzyme complex. Which of the following vitamins supplies the reactive prosthetic group that tethers the growing acyl chain to the Acyl Carrier Protein (ACP) domain?

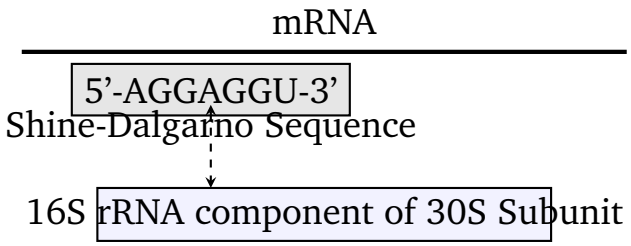
(A) Pantothenic acid

(B) Riboflavin

(C) Thiamine

(D) Niacin

**Q8.** The following schematic diagram highlights a key step in prokaryotic translation initiation:




What is the exact complementary sequence found at the 3' end of the 16S rRNA that base-pairs with the Shine-Dalgarno sequence shown above?

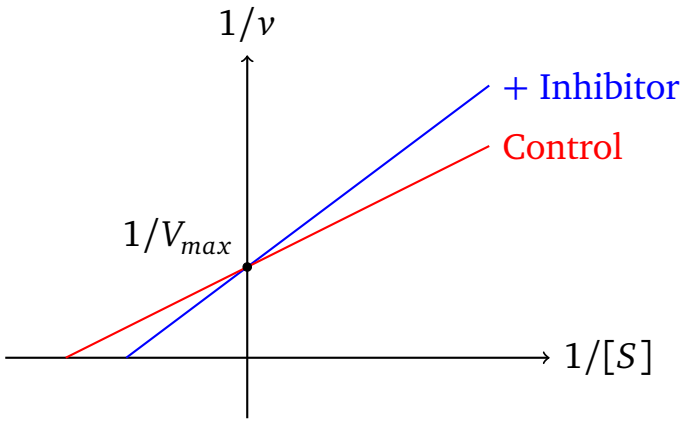
(A) 5'-UCCUCCA-3'

(B) 5'-ACCUCCU-3'

(C) 5'-AGGAGGU-3'

(D) 5'-UUGGUUG-3'

**Q9.** A Michaelis-Menten plot is constructed for an enzyme-catalyzed reaction in the presence and absence of a reversible metabolic inhibitor. The experimental data is transformed into a Lineweaver-Burk double-reciprocal plot as shown below:




Based on this diagnostic plot, which type of inhibition is occurring, and how are the kinetic parameters affected?

(A) Non-competitive inhibition; $V_{max}$ decreased, $K_m$ unchanged

(B) Competitive inhibition; $V_{max}$ unchanged, $K_m$ increased

(C) Uncompetitive inhibition; $V_{max}$ decreased, $K_m$ decreased

(D) Allosteric inhibition; $V_{max}$ increased, $K_m$ increased

**Q10.** A 2-year-old boy presents with hepatomegaly, severe fasting hypoglycemia, and lactic acidosis. Physical exam reveals a doll-like facies. Administration of epinephrine or glucagon fails to induce an increase in blood glucose levels, but causes a rapid rise in blood lactate. A liver biopsy shows significantly elevated glycogen content with normal structure. Which enzyme is missing in this child?

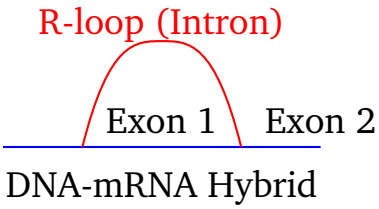
(A) Glycogen Phosphorylase

(B) Glucose-6-Phosphatase

(C) $\alpha$-1,4-Glucosidase

(D) Debranching Enzyme

**Q11.** A molecular biologist is mapping a cloned eukaryotic gene. He matches the mature cytoplasmic mRNA sequence back to the genomic locus using electronic alignment software. The structural differences discovered can be represented by the loop configuration below:




Which of the following highly conserved dinucleotide sequences define the 5' (donor) and 3' (acceptor) boundaries of the sequence that forms the looped-out region during nuclear pre-mRNA splicing?

(A) 5'-AT...GC-3'

(B) 5'-AG...GT-3'

(C) 5'-GT...AG-3'

(D) 5'-AC...TG-3'

**Q12.** A 3-month-old infant is brought to the clinic due to poor feeding, vomiting, and progressive yellowing of the skin. Urine analysis reveals the presence of reducing sugars, but a glucose oxidase dipstick test is negative. The child is diagnosed with classical galactosemia. Deficiencies in this metabolic pathway result in the toxic accumulation of which cellular metabolite?

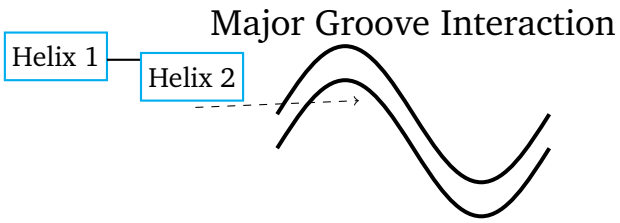
(A) Galactose-1-phosphate

(B) UDP-Galactose

(C) Glucose-1-phosphate

(D) Galactitol only

**Q13.** In a molecular genetics laboratory, a technician performs an analysis of an uncharacterized operon. She notices that a specific regulatory protein contains a structural motif consisting of an $\alpha$-helix, a short turn, and a second $\alpha$-helix that fits perfectly into the major groove of B-DNA, as modeled below:




What is the primary classification of this common DNA-binding protein motif?

(A) Leucine Zipper

(B) Zinc Finger

(C) Helix-Turn-Helix

(D) Beta-Barrel

**Q14.** A newborn displays ambiguous genitalia and severe salt-wasting crisis within the first week of life. Laboratory studies show marked hyponatremia, hyperkalemia, and elevated levels of 17-hydroxyprogesterone. The biochemical

block in this infant's adrenal gland prevents the synthesis of which steroid intermediates?

(A) 11-Deoxycorticosterone and 11-Deoxycortisol

(B) Cholesterol and Pregnenolone

(C) DHEA and Androstenedione

(D) Cortisol and Testosterone exclusively

**Q15.** A patient with a history of deep vein thrombosis is found to have significantly elevated plasma homocysteine levels. To optimize the conversion of homocysteine back to methionine, which pair of vitamins must act as essential cofactors/co-substrates for the primary salvage enzyme?

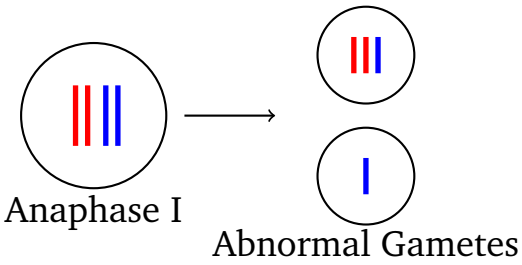
(A) Pyridoxine (B6) and Biotin (B7)

(B) Cobalamin (B12) and Methyltetrahydrofolate

(C) Thiamine (B1) and Riboflavin (B2)

(D) Ascorbic Acid (C) and Niacin (B3)

**Q16.** Karyotype analysis of a child presenting with intellectual disability, prominent epicanthal folds, low-set ears, and a single palmar crease confirms Trisomy 21. Meiotic nondisjunction is determined to be the underlying mechanism. The generation of this numerical chromosomal aberration can be tracked by analyzing the configuration of homologous chromosomes during gametogenesis:




Failure of homologous chromosomes to separate most commonly occurs during which phase of maternal oogenesis?

(A) Mitotic Anaphase of primordial germ cells

(B) Anaphase I of Meiosis I

(C) Anaphase II of Meiosis II

(D) Prophase I pachytene arrest

## Detailed Solutions

**Q1.**

### Solution

**Concept:**

DNA replication requires coordination between leading and lagging strand synthesis. While the leading strand is synthesized continuously, the lagging strand is generated discontinuously as a series of short segments called Okazaki fragments. To complete replication and restore a single, uninterrupted strand of DNA, these fragments must be joined together. The enzyme responsible for sealing these breaks is DNA ligase.

**Solution:**

(a) DNA ligase repairs nicks in the sugar-phosphate backbone by creating a bond between the adjacent fragments. This reaction requires an energy source, which is ATP in eukaryotic and mammalian cells, or NAD+ in most prokaryotes.

(b) The specific reaction involves a nucleophilic attack by the 3'-hydroxyl group of one nucleotide on the 5'-phosphate group of the next nucleotide, forming a standard $3' \rightarrow 5'$ phosphodiester bond.

(c) A $5' \rightarrow 3'$ phosphodiester bond notation does not represent the proper directional synthesis or assembly order carried out by standard polymerases and ligases, which always direct structural connectivity or elongation from the 5' end to the 3' end, establishing a $3' \rightarrow 5'$ linkage.

(d) N-glycosidic bonds anchor nitrogenous bases to the deoxyribose sugar rings and are not target substrates for replication strand-joining.

(e) Hydrogen bonds form spontaneously between complementary base pairs to hold the two strands together and do not require enzymatic ligation using nucleotide triphosphates.

**Final Answer:** $3' \rightarrow 5'$ Phosphodiester bond using ATP

**Answer: (A)** Go Back to Question 1

## Q2.

### Solution

**Concept:**

The clinical presentation of ataxia, confusion, and ophthalmoplegia defines Wernicke encephalopathy, a condition caused by a severe deficiency of thiamine (vitamin B1). Chronic alcoholism impairs thiamine absorption and storage. Thiamine pyrophosphate (TPP) serves as an essential cofactor for several key multi-enzyme complexes involved in energy metabolism, including the pyruvate dehydrogenase complex, the $\alpha$-ketoglutarate dehydrogenase complex, and transketolase.

**Solution:**

(a) The $\alpha$-ketoglutarate dehydrogenase complex in the citric acid cycle requires TPP, lipoamide, FAD, NAD+, and coenzyme A to catalyze the oxidative decarboxylation of $\alpha$-ketoglutarate.

(b) When thiamine is deficient, the lack of functional TPP directly blocks this specific step, stopping the conversion of $\alpha$-ketoglutarate to succinyl-CoA and impairing ATP production in high-energy tissues like the brain.

(c) The conversion of succinate to fumarate is catalyzed by succinate dehydrogenase, which requires FAD, not thiamine.

(d) The conversion of malate to oxaloacetate is carried out by malate dehydrogenase and uses NAD+ as an electron acceptor.

(e) The conversion of pyruvate to oxaloacetate is an irreversible carboxylation reaction catalyzed by pyruvate carboxylase, an enzyme that requires biotin (vitamin B7) as its prosthetic group rather than thiamine.

**Final Answer:** Conversion of $\alpha$-ketoglutarate to Succinyl-CoA

**Answer: (B)** Go Back to Question 2

## Q3.

### Solution

**Concept:**

The triad of severe developmental delay, self-mutilation, and hyperuricemia in a young male indicates Lesch-Nyhan syndrome, an X-linked recessive metabolic disorder. The underlying defect is a complete or near-complete deficiency of hypoxanthine-guanine phosphoribosyl-transferase (HGPRT). This enzyme plays a crucial role in purine nucleotide metabolism by recycling purine bases that are generated during normal cellular turnover.

**Solution:**

(a) The HGPRT enzyme is a central component of the purine salvage pathway, where it converts hypoxanthine back into inosine monophosphate (IMP) and guanine back into guanosine monophosphate (GMP).

(b) When HGPRT is deficient, hypoxanthine and guanine cannot be salvaged and are instead degraded into uric acid, leading to severe hyperuricemia, gout, and distinctive neurodevelopmental complications.

(c) Because purine bases cannot be recycled, the intracellular level of PRPP increases while purine nucleotide levels drop, removing feedback inhibition and driving up de novo purine synthesis secondary to the salvage defect.

(d) The de novo purine synthesis pathway builds the purine ring from simple precursors rather than recycling pre-formed bases.

(e) Pyrimidine salvage and de novo pyrimidine synthesis handle cytosine, uracil, and thymine nucleotides, which are unaffected in this specific disease.

**Final Answer:** Purine salvage pathway

**Answer: (B)**

Go Back to Question 3

## Q4.

### Solution

**Concept:**

Phosphofructokinase-1 (PFK-1), designated as Enzyme X, is the key rate-limiting enzyme of glycolysis. It is strongly activated by fructose-2,6-bisphosphate (F-2,6-BP), which signals an abundance of glucose. The level of F-2,6-BP is regulated by a bifunctional enzyme called PFK-2/FBPase-2, whose activity is directly modulated by phosphorylation in response to pancreatic hormones.

**Solution:**

(a) In the well-fed state, blood glucose levels rise, triggering a high insulin-to-glucagon ratio. Insulin activates protein phosphatase-1, which dephosphorylates the bifunctional enzyme.

(b) Dephosphorylation activates the PFK-2 domain while inhibiting the FBPase-2 domain, increasing the synthesis of F-2,6-BP, which allosterically activates PFK-1 and drives glycolysis forward.

(c) A high glucagon-to-insulin ratio, seen during fasting, activates protein kinase A to phosphorylate the bifunctional enzyme, destroying F-2,6-BP to favor gluconeogenesis.

(d) Epinephrine during fasting triggers cAMP production, which mimics the effects of glucagon by decreasing F-2,6-BP levels to shut down glycolysis in the liver.

(e) Cortisol promotes gluconeogenesis during prolonged starvation by altering gene expression rather than rapidly increasing F-2,6-BP levels via acute allosteric pathways.

**Final Answer:** High Insulin/Glucagon ratio

**Answer: (C)**

Go Back to Question 4

## Q5.

### Solution

**Concept:**

The presentation of dark urine that turns black upon standing, combined with blue-black tissue pigmentation (ochronosis), is diagnostic of alkaptonuria. This autosomal recessive disorder is caused by a deficiency of homogentisate oxidase, an enzyme in the catabolic pathway of aromatic amino acids. Without this enzyme, homogentisic acid accumulates and polymerizes into a dark pigment that deposits in connective tissues.

**Solution:**

(a) Homogentisate oxidase operates downstream in the degradation pathway where phenylalanine is converted into tyrosine, which is then metabolized into homogentisate.

(b) Consequently, a defect in homogentisate oxidase impairs the proper breakdown of both phenylalanine and tyrosine, leading to the clinical manifestations of alkaptonuria.

(c) Tryptophan is an aromatic amino acid, but it degrades via the kynurenine pathway to form nicotinamide, while methionine is a sulfur-containing amino acid metabolized via the homocysteine pathway.

(d) Lysine and leucine are purely ketogenic amino acids that break down into acetyl-CoA and acetoacetate through pathways independent of homogentisate.

(e) Valine and isoleucine are branched-chain amino acids whose catabolism relies on branched-chain $\alpha$-keto acid dehydrogenase, a pathway disrupted in maple syrup urine disease.

**Final Answer:** Phenylalanine and Tyrosine

**Answer: (A)**

Go Back to Question 5

## Q6.

### Solution

**Concept:**

The pedigree details an inheritance pattern where an affected female transmits the condition to all her children, while an affected male never transmits it. This is classic maternal or mitochondrial inheritance. Mitochondria are inherited exclusively from the mother because the oocyte contains numerous mitochondria, whereas the sperm contributes only its nuclear genome during fertilization.

**Solution:**

(a) Mitochondrial DNA mutations display heteroplasmy, a condition where a single cell contains a mixture of normal and mutated mitochondrial genomes.

(b) During cell division, mitochondria replicate and distribute randomly to daughter cells. This random segregation leads to variable expressivity, causing significant phenotypic differences among family members.

(c) Mitotic crossover is a rare somatic recombination process that does not explain the strict maternal inheritance observed in multi-generational family pedigrees.

(d) Genomic imprinting involves epigenetic gene silencing based on the parent of origin, but it typically applies to nuclear genes and does not cause a complete absence of paternal transmission to all offspring.

(e) Allelic exclusion ensures that only one allele of a gene is expressed while the other is silenced, a process seen in immunoglobulin production rather than mitochondrial transmission.

**Final Answer:** Heteroplasmy and variable expressivity

**Answer: (B)**

Go Back to Question 6

## Q7.

### Solution

**Concept:**

Fatty acid synthesis is carried out by Fatty Acid Synthase (FAS), a homodimeric multienzyme complex. During this process, intermediates are moved between different active sites while remaining covalently attached to the enzyme. This tethering is managed by the Acyl Carrier Protein (ACP) domain, which relies on a specialized prosthetic group derived from a water-soluble vitamin.

**Solution:**

(a) The ACP domain requires activation by a phosphopantetheinyl transferase, which links a 4'-phosphopantetheine group to a conserved serine residue on the protein.

(b) This 4'-phosphopantetheine prosthetic group is derived from pantothenic acid (vitamin B5) and contains a highly reactive sulfhydryl (-SH) group that forms a thioester bond with the growing fatty acid chain.

(c) Riboflavin (vitamin B2) is the precursor for FAD and FMN, cofactors used primarily in oxidation-reduction reactions like those in the electron transport chain.

(d) Thiamine (vitamin B1) forms thiamine pyrophosphate, a cofactor essential for oxidative decarboxylation and transketolase reactions.

(e) Niacin (vitamin B3) is the structural basis for NAD+ and NADP+, which act as soluble electron carriers rather than covalently bound structural tethers on the FAS complex.

**Final Answer:** Pantothenic acid

**Answer: (A)**

Go Back to Question 7

## Q8.

### Solution

**Concept:**

Translation initiation in prokaryotes requires the small ribosomal subunit (30S) to position itself correctly at the start codon of the mRNA. This precise alignment is mediated by the Shine-Dalgarno sequence, a purine-rich region located upstream of the AUG start codon. The 30S subunit contains a structural RNA component that directly pairs with this mRNA sequence.

**Solution:**

(a) The 16S rRNA component of the 30S subunit contains a complementary, pyrimidine-rich sequence at its 3' terminus that base-pairs with the Shine-Dalgarno sequence.

(b) Given the mRNA Shine-Dalgarno sequence 5'-AGGAGGU-3', the complementary antiparallel alignment requires the 16S rRNA sequence to read 3'-UCCUCCA-5', which is written as 5'-ACCUCCU-3' from its 5' to 3' end.

(c) The sequence 5'-UCCUCCA-3' is incorrect because it runs parallel to the mRNA instead of antiparallel, violating standard nucleic acid base-pairing orientation.

(d) The sequence 5'-AGGAGGU-3' is identical to the Shine-Dalgarno sequence on the mRNA and would result in electrostatic repulsion rather than complementary base-pairing.

(e) The sequence 5'-UUGGUUG-3' does not match the base-pairing pattern needed to align with the purine core of the Shine-Dalgarno region.

**Final Answer:** 5'-ACCUCCU-3'

**Answer: (B)**

Go Back to Question 8

## Q9.

### Solution

**Concept:**

Enzyme inhibitors alter the kinetic properties of enzymes, changes that can be distinguished using a Lineweaver-Burk double-reciprocal plot. This plot graphs the inverse of reaction velocity $(1/v)$ against the inverse of substrate concentration $(1/[S])$. The y-intercept represents $1/V_{max}$, while the x-intercept indicates $-1/K_m$.

**Solution:**

(a) In the provided plot, both the control curve and the inhibitor curve intersect at the exact same point on the y-axis, demonstrating that $1/V_{max}$ is unchanged and $V_{max}$ remains the same.

(b) The inhibitor curve shifts rightward along the x-axis, moving the x-intercept closer to zero. This change represents an increase in $K_m$, confirming that a higher substrate concentration is required to reach half-maximal velocity.

(c) An unchanged $V_{max}$ coupled with an increased $K_m$ is the classic hallmark of competitive inhibition, where the inhibitor competes with the substrate for the active site.

(d) Non-competitive inhibition decreases $V_{max}$ while leaving $K_m$ unchanged, which would cause the lines to intersect on the x-axis rather than the y-axis.

(e) Uncompetitive inhibition decreases both $V_{max}$ and $K_m$, creating parallel lines on a double-reciprocal plot.

**Final Answer:** Competitive inhibition; $V_{max}$ unchanged, $K_m$ increased

**Answer: (B)**

Go Back to Question 9

## Q10.

### Solution

**Concept:**

A presentation featuring fasting hypoglycemia, lactic acidosis, hepatomegaly, and a doll-like facies points to von Gierke disease (Glycogen Storage Disease Type I). This condition blocks the final step of both glycogenolysis and gluconeogenesis, preventing the liver from releasing free glucose into the blood during fasting.

**Solution:**

(a) Glucose-6-phosphatase hydrolyzes glucose-6-phosphate into free glucose in the lumen of the endoplasmic reticulum. A deficiency in this enzyme traps glucose-6-phosphate inside hepatocytes.

(b) The trapped glucose-6-phosphate enters glycolysis, leading to an overproduction of pyruvate and lactate, which causes severe lactic acidosis. It also stimulates glycogen synthesis, leading to the accumulation of normal-structured glycogen in the liver.

(c) A deficiency in glycogen phosphorylase (Hers disease) causes mild fasting hypoglycemia without significant lactic acidosis, as gluconeogenesis remains completely functional.

(d) A deficiency in $\alpha$-1,4-glucosidase (Pompe disease) leads to glycogen accumulation within lysosomes, causing severe cardiomyopathy without fasting hypoglycemia.

(e) Debranching enzyme deficiency (Cori disease) causes the accumulation of abnormal glycogen with short outer branches (limit dextrin) and milder hypoglycemia because gluconeogenesis is unaffected.

**Final Answer:** Glucose-6-Phosphatase

**Answer: (B)**

Go Back to Question 10

## Q11.

### Solution

**Concept:**

Pre-mRNA splicing is a fundamental nuclear process in eukaryotic gene expression where non-coding intervening sequences (introns) are excised and coding sequences (exons) are joined together. This highly coordinated catalytic reaction is carried out by the spliceosome, a large macromolecular complex consisting of small nuclear ribonucleoprotein particles (snRNPs). The recognition of splice sites depends on highly conserved, specific consensus nucleotide sequences located at the boundaries of all nuclear introns.

**Solution:**

(a) The vast majority of nuclear pre-mRNA introns follow the classic GT-AG rule, which describes the invariant dinucleotides found at the ends of the intron strand.

(b) The 5' splice site, also known as the donor site, always begins with a highly conserved 5'-GT-3' dinucleotide sequence on the coding DNA strand, which transcribes to 5'-GU-3' in the primary pre-mRNA transcript.

(c) The 3' splice site, also known as the acceptor site, consistently terminates with a highly conserved 5'-AG-3' dinucleotide sequence immediately preceding the downstream exon boundary.

(d) The configurations 5'-AT...GC-3', 5'-AG...GT-3', and 5'-AC...TG-3' do not represent the canonical consensus motifs that direct spliceosome assembly, and mutations altering the normal boundary sequences frequently cause aberrant splicing patterns linked to genetic diseases.

**Final Answer:** 5'-GT...AG-3'

**Answer: (C)**

Go Back to Question 11

## Q12.

### Solution

**Concept:**

Classical galactosemia is an autosomal recessive inborn error of carbohydrate metabolism caused by a severe deficiency of galactose-1-phosphate uridylyltransferase (GALT). This enzyme is a key component of the Leloir pathway, which converts dietary galactose into glucose-1-phosphate. When this pathway is blocked, galactose metabolism stalls, leading to alternative reduction pathways and the toxic buildup of upstream metabolic intermediates in multiple tissues, including the liver, brain, and kidneys.

**Solution:**

(a) A deficiency in the GALT enzyme prevents the conversion of galactose-1-phosphate and UDP-glucose into UDP-galactose and glucose-1-phosphate, directly causing an accumulation of galactose-1-phosphate within cells.

(b) The intracellular accumulation of galactose-1-phosphate is highly cytotoxic, directly causing cellular injury that manifests clinically as hepatomegaly, jaundice, liver cirrhosis, and early cataract formation.

(c) High systemic levels of galactose drive the alternative reduction of excess sugar by aldose reductase into galactitol, an osmotically active sugar alcohol that accumulates within the lens of the eye and induces cataract development.

(d) While galactitol causes osmotic damage, the primary, tissue-toxic upstream metabolite characteristic of GALT deficiency is galactose-1-phosphate, distinguishing it from milder galactokinase deficiency where galactose-1-phosphate is not formed.

**Final Answer:** Galactose-1-phosphate

**Answer: (A)**

Go Back to Question 12

## Q13.

### Solution

**Concept:**

Gene expression is regulated through specialized structural frameworks within transcription factors known as DNA-binding domains. These domains contain specific structural configurations that recognize and bind to unique nucleotide sequences along the major or minor grooves of B-DNA. This interaction allows the regulatory proteins to stabilize, recruit, or block transcription machinery, thereby controlling the transcription rate of downstream target operons or eukaryotic genes.

**Solution:**

(a) The structural motif described consists of two alpha-helices separated by a short turn, where one helix fits into the major groove of the DNA molecule to read sequence-specific information while the other stabilizes the interaction.

(b) This specific spatial layout defines the helix-turn-helix (HTH) structural domain, which is the classic DNA-binding motif identified in prokaryotic repressor proteins, such as the lac repressor, and homeodomain proteins.

(c) The leucine zipper motif features a periodic repetition of leucine residues every seven positions along an amphipathic alpha-helix, forming a dimerization interface that brings two separate DNA-binding regions together.

(d) The zinc finger motif stabilizes its structural loops using a coordinated zinc ion bound to specific cysteine and histidine residues, creating finger-like protrusions that insert into the DNA major groove.

(e) The beta-barrel structural motif is an arrangement of anti-parallel beta-strands forming a pore-like structure, which is typically found in transmembrane porins rather than sequence-specific transcription factors.

**Final Answer:** Helix-Turn-Helix

**Answer: (C)**

Go Back to Question 13

## Q14.

### Solution

**Concept:**

Congenital adrenal hyperplasia (CAH) comprises a group of autosomal recessive disorders characterized by enzyme deficiencies in the steroidogenic pathways of the adrenal cortex. The clinical findings of ambiguous genitalia in females, alongside severe salt-wasting crises, hyponatremia, hyperkalemia, and a prominent elevation of 17-hydroxyprogesterone, point directly to the most common form of this condition: 21-hydroxylase deficiency.

**Solution:**

(a) The 21-hydroxylase enzyme is responsible for converting progesterone into 11-deoxycorticosterone and converting 17-hydroxyprogesterone into 11-deoxycortisol within the adrenal cortex.

(b) A biochemical block at this step halts the production of both 11-deoxycorticosterone and 11-deoxycortisol, completely shutting down the synthesis of downstream glucocorticoids and mineralocorticoids.

(c) The deficiency of mineralocorticoids leads to aldosterone depletion, which prevents sodium reabsorption in the renal tubules, triggering severe clinical salt-wasting, hyponatremia, and hyperkalemia.

(d) Because the 21-hydroxylase pathway is blocked, accumulated steroid intermediates are shunted into the adrenal androgen pathway, driving excess testosterone production and causing ambiguous genitalia in female newborns.

(e) Cholesterol, pregnenolone, DHEA, and androstenedione are located upstream of or parallel to the 21-hydroxylase block, so their synthesis remains uninhibited.

**Final Answer:** 11-Deoxycorticosterone and 11-Deoxycortisol

**Answer: (A)**

Go Back to Question 14

## Q15.

### Solution

**Concept:**

Hyperhomocysteinemia is a significant metabolic risk factor associated with endothelial dysfunction, arterial damage, and an increased risk of deep vein thrombosis. Homocysteine sits at a critical metabolic branch point where it can either be remethylated to methionine or transsulfurated to cysteine. The primary remethylation pathway operating in most extrahepatic tissues relies on the enzyme methionine synthase to transfer a methyl group back onto homocysteine.

**Solution:**

(a) Methionine synthase catalyzes the transfer of a methyl group from N5-methyltetrahydrofolate to homocysteine, converting it into methionine while regenerating free tetrahydrofolate.

(b) This complex biochemical reaction requires methylcobalamin, an active coenzyme form of vitamin B12, to act as an intermediate methyl carrier during the enzymatic transfer.

(c) N5-methyltetrahydrofolate serves as the primary methyl donor in this pathway, meaning both vitamin B12 and folate are essential components required to lower homocysteine via remethylation.

(d) Vitamin B6 (pyridoxine) is a necessary cofactor for cystathionine beta-synthase, but it operates in the alternative transsulfuration pathway rather than the primary remethylation pathway.

(e) Biotin, thiamine, riboflavin, niacin, and ascorbic acid are involved in other metabolic pathways and do not participate as cofactors or co-substrates for methionine synthase.

**Final Answer:** Cobalamin (B12) and Methyltetrahydrofolate

**Answer: (B)**

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## Q16.

### Solution

**Concept:**

Trisomy 21, the genetic cause of Down syndrome, results from a failure of chromosomal separation during cell division, a process known as meiotic nondisjunction. This error produces gametes with an abnormal number of chromosomes. While nondisjunction can happen during spermatogenesis or oogenesis, statistical and molecular mapping studies show that the vast majority of these errors occur during maternal meiosis, with risk increasing alongside advancing maternal age.

**Solution:**

(a) Maternal oogenesis begins during embryonic development, where primary oocytes arrest in prophase I until ovulation begins at puberty, leaving chromosomes suspended in this state for decades.

(b) The primary defect causing classical Trisomy 21 is a failure of homologous chromosome pairs to separate properly during anaphase I of maternal meiosis I, sending an intact homologous pair into a single secondary oocyte.

(c) If anaphase I proceeds normally, a secondary error can occur during anaphase II where sister chromatids fail to separate, though this represents a less common cause of meiotic nondisjunction.

(d) Mitotic anaphase errors in primordial germ cells would affect the entire germline lineage, which does not align with the sporadic, age-dependent nature of standard meiotic nondisjunction events.

(e) The prolonged pachytene or dictyate arrest in prophase I sets the stage for age-related cohesion loss, but the actual failure of physical separation takes place during anaphase I.

**Final Answer:** Anaphase I of Meiosis I

**Answer: (B)**

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# Answer Key

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