[MBBS 0124] **JANUARY 2024** **Sub. Code : 6056**

M.B.B.S. DEGREE EXAMINATION
(For the candidates admitted from the Academic Year 2019-2020)

FIRST YEAR – SUPPLEMENTARY (CBME)
PAPER II – BIOCHEMISTRY

Q.P. Code: 526056

Time: Three hours **Maximum : 100 Marks (80 Theory + 20MCQs)**

Answer All Questions

I. Essay: **(2 x 15 = 30)**

1. A 32-year-old woman presents with vomiting and diarrhoea. Evaluation revealed acidosis. ABG analysis revealed that she presents with normal anion gap metabolic acidosis.
   a) What are plasma and urinary buffers?
   b) Which is the major plasma buffer? Why?
   c) What is anion gap?
   d) What is the clinical significance of anion gap?
   e) Why does diarrhoea present with acidosis?
   f) Why is the anion gap normal in this condition?

2. A known case of Decompensated Liver Disease (DCLD) presents with sleep disturbances and confusion. He is diagnosed with hepatic encephalopathy due to hyperammonemia.
   a) Why does DCLD cause hyperammonemia?
   b) Why does hyperammonemia cause encephalopathy?
   c) What are the non-toxic forms of ammonia?
   d) What are the causes of primary hyperammonemia?
   e) Describe in detail the biochemical steps in the formation of Ammonia.

II. Write notes on: **(10 x 5 = 50)**

1. A 65 year old man presents with back pain. On examination he is found to have multiple lytic lesions in the spine. A serum protein electrophoresis (EPP) was performed, which revealed a diagnosis of Multiple Myeloma.
   a) What is the pH of the buffer used for serum protein electrophoresis? Why?
   b) What is the band observed in serum protein electrophoresis in Multiple Myeloma? Describe.
   c) Describe the EPP pattern observed in Nephrotic syndrome.

2. The proband was a 40-year-old male was diagnosed with poorly differentiated adenocarcinoma. A family counseling revealed that the proband had five family members with colorectal cancer diagnosed before 45 years of age. As Hereditary Non Polyposis Colon Cancer (HNPCC) is suspected, all family members were counselled that this caused by a defect of DNA repair and that all family members older than 25 years should undergo regular colonoscopic examination.
   a) What are the errors that can happen in a DNA?
   b) Which is the most common error in a DNA?
   c) Which repair defect causes HNPCC?
   d) Describe in detail about the repair mechanism causative of HNPCC.

3. STATEMENT 1 : More than one codon can code for a single aminoacid.
   STATEMENT 2 : More than one aminoacids can be coded by a single codon.
   a) Which statement is true?
   b) Mention the property of genetic code, that is applicable to the true statement.
   c) What is wobble phenomenon? Explain with an example.
   d) Mention all the properties of genetic code.

4. A 11 year old boy is diagnosed with Type 1 diabetes. He was prescribed Human Biosynthetic Insulin (BHI).
   a) Mention the technology used for the synthesis of BHI.
   b) Mention the tools required for the technology.
   c) What is cDNA? How is it synthesized?

5. A 17 year old girl is diagnosed with iron deficiency anemia. She is a vegetarian and hence her nutritionist suggests her mom that her greens be given the tangy flavor using lime instead of tomato.
   a) How is heme iron absorbed along the apical side of intestine?
   b) How is non heme iron absorbed along the apical side of intestine?
   c) What is the rationale behind adding lime to greens?
   d) Describe in detail the basolateral side transport of iron.

6. A 56-year-old man, a known hypertensive presented to his family doctor with loss of appetite, weight loss, generalized weakness and lethargy of six months duration. A blood sample was taken for analysis.

   Serum : Sodium 130 mmol/L
   Potassium 5.2 mmol/L
   Bicarbonate 16 mmol/L
   Urea 258 mg/dL
   Creatinine 7.1 mg/dL
   Calcium 7.2 mg/dL
   Phosphate 8.6 mg /dL
   Albumin 2.8g /dL
   Alkaline phosphatase 205 U/L

   A diagnosis of chronic kidney disease was made
   a) Interpret serum calcium and substantiate the change observed in chronic kidney disease.
   b) Interpret and reason out potassium level alteration observed in the condition.
   c) What is the expected change in Parathormone levels in this patient? Why?

7. A 6 year old boy presents with periodic aggressive behavior. His urinary ALA is elevated. On examination, he is icteric. A mild hepatomegaly is observed. Blood examination revealed massive elevation of AFP. HPLC and TMS examination revealed elevation of succinylacetone. A diagnosis of Type I Tyrosinemia is made.
   a) What is the most probable enzyme defect?
   b) Why does he present with elevation of ALA and neuropsychiatric manifestation?
   c) Mention other Tyrosine metabolism disorders and mention the respective enzyme defects.

8. A 55-year-old person was diagnosed with Type 2 diabetes. The diabetologist asked him to get HbA1C estimated. His HbA1C was reported with a special mention that there was an abnormal migration of the hemoglobin in electrophoresis. His SpO2 was normal.
    a) What type of mutation is this?
    b) Classify mutation based on effects of mutation on nucleotide sequence, aminoacid sequence and function of protein.
    c) Give suitable examples.

9. An 8 months old male child was referred with severe transfusion dependent anemia. Complete Blood Count (CBC) showed microcytic hypochromic anemia. In view of clinical suspicion of Thalassemia major, HPLC for Hb Variant detection was performed, which revealed, HbF-23% (very high).
    a) What are the differences between Fetal hemoglobin (HbF) and adult hemoglobin (HbA)?
    b) Draw the Oxyhemoglobin Dissociation curve of HbA and HbF in a single graph.
    c) Why does fetal hemoglobin shift the oxyhemoglobin dissociation curve?

10. A 72-year-old woman from a nursing home presents to the emergency department with a change in her mental state over the past few hours. She has a history of hypertension and is on diuretics. Her serum sodium was 110mEq/L and serum Osmolality was 278mmol/L. On physical examination, she has decreased skin turgor, orthostatic hypotension and disorientation to time, place and person without focal neurologic deficits.
    a) What is the normal serum Sodium and Serum Osmolality?
    b) Interpret and comment on her serum sodium levels and serum Osmolatity.
    c) What are the types of Hyponatremia and give examples?
    d) Which types of Hyponatremia has the 72 year old woman presented with?

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