

Department of Biochemistry

ECE Bio Paper Cases and Interpretation of associated Laboratory Findings (Vitamins, Minerals, Heme and Porphyrin Metabolism)

For class to be held on 30.03.2020 (2-5 pm)

Dr Rafat Ahmed, All Residents

Instructions

Students are required to go through the cases/questions and prepare a written assignment, to be submitted when classes resume.

Vitamin metabolism

Question 1

Case1

A 46 year old woman was observed by her husband to be not eating at all. She was admitted with dehydration and disorientation. Her physical condition improved following rehydration. She however seemed mentally disturbed. She claimed to be much younger than she really was. She craved for alcohol. She said she never gave birth to a child and that her daughter was actually her sister's child. She was unable to recognise any of her colleagues. Neuroimaging revealed cerebellar atrophy.

The probable diagnosis is

- | | |
|-------------------------|------------------------------|
| a. Alzheimer's dementia | b. Wernicke's encephalopathy |
| c. Korsakoff's syndrome | d. Viral Dementia |

Post answering the case the following questions prevail:-

1. What is Wernicke's encephalopathy? (caused by, common causes)
2. Why does alcoholism cause thiamine deficiency?
3. Why should thiamine deficiency present with encephalopathy?
4. What are the precipitating factors of Wernicke's encephalopathy in a chronic alcoholic?
5. What are the features of Wernicke's encephalopathy?
6. What are the differences between Wernicke's encephalopathy and Korsakoff's syndrome?

Case 2

A 38 year old vegetarian female presented to her doctor with fatigue and tingling/ numbness in her extremities (bilateral). The symptoms were gradually getting worse over the last year. She reported frequent episodes of diarrhoea and weight loss. On examination she was pale with tachycardia. Her tongue was beefy red. Neurological examination revealed numbness in all extremities with decreased vibration sense. The CBC demonstrated megaloblastic anaemia.

1. What is the most likely diagnosis?
2. What is the most underlying problem for the patient?
3. What are the two most common causes of megaloblastic anaemia and how would u differentiate between the two based on patient history and examination.
4. If this case was of folic acid deficiency what would be the cause of macrocytosis?

Case 3

A term infant was born and did well with breast feeding. Two days later the mother called frantically because baby was bleeding from the area of umbilical cord and nostrils. Injection vitamin K was given immediately to stop the bleeding.

1. Mention the deficiency
2. Mechanism of action of vitamin K
3. Why do newborn infants have low vitamin K?

Case 4

A 6 year old child was brought to the hospital with complaints of slow growth and pain in bones. On examination he was anaemic, had frontal bossing, bowing of legs and swelling of costochondral junction. Lab results were serum calcium – 8.2 mg/dl, serum phosphorus – 2.8 mg/dl and serum ALP – 720 U/L

1. What is the most likely diagnosis?
2. What are the various causes of the deficiency
3. Describe sources with activation, biochemical functions, deficiency manifestations (write like LQ)

Question 2

Explain why

- a) Pellagra like symptoms are seen in carcinoid syndrome.
- b) Microcytic hypochromic anaemia is seen in Vitamin C deficiency
- c) Pyridoxine is given as treatment in hyper-homocystinemia
- d) Pregnant women are administered Folic acid Prophylaxis

Mineral metabolism

Question 1

Case 1

A 10 year old girl presented with excessive tiredness, poor appetite, inability to concentrate and tingling sensation. On examination there was pallor. Lab examination revealed decrease in haemoglobin, ferritin, MCV. TIBC (Total Iron binding Capacity) was increased.

1. What is the likely diagnosis
2. Biochemical basis of all the findings
3. Describe the absorption, biochemical functions and deficiency manifestations (deficiency& excess) of the deficient Mineral

Case 2

A 44 year old woman presented to the clinic with complaints of vague abdominal discomfort, weakness and fatigue and bone pain. She had several Urinary Tract infections and several episodes of kidney stones. Her physical examination was within normal limits. The patient had normal CBC, elevated calcium and low phosphorus.

1. What is the most likely diagnosis?
2. Biochemical basis for the same.
3. What are the normal levels in blood(of the above mentioned minerals)
4. Describe mechanism by which the homeostasis of the minerals is maintained

Haemoglobin metabolism

Question 1

Case 1

A 21 year old healthy male college student went to celebrate his birthday with some friends at a bar. His friends convinced him to have his first beer since he was 21. After consuming beer he began to experience intense worsening abdominal pain that was non specific in location and described it to be like cramps. Nausea and vomiting then ensued and he was then taken to emergency room. Upon arrival to ER he was found to be anxious with hallucinations. He was noted to be hypertensive, tachycardic, and diaphoretic. Peripheral neuropathy was noticed on examination. Initial lab tests revealed normal CBC, drug screen, EtOH level. Serum and urine aminolevulinic acid and porphobilinogen were both found to be elevated.

1. What is the likely diagnosis?
2. What is the biochemical basis?
3. What is porphyria? Classify different types of porphyrias. Give an account of acute intermittent porphyria.

Question 2

Write in Brief

- a) Physiological Jaundice of Newborn
- b) Crigler Najjar Syndrome