"Acute abdomen with respiratory failure:- The Porphyric Jigsaw"

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Abstract

Acute intermittent porphyria is one of the metabolic disorder which can create lot of confusion while managing a case because of it's rapid changing manifestation and involvement of different systems. We present of a 45-year-old man a case of acute intermittent porphyria who presented with acute abdominal pain and later developed areflexic paralysis in all four limbs and respiratory failure.

Key words:- Acute intermittent porphyria, acute abdominal pain.

Case Report

A 45-year-old non alcoholic man went for a marriage ceremony and had an alcohol binge there and was later admitted to the hospital under surgery with acute abdominal pain. Patient's vitals were stable with Pulse of 114/min, Respiratory rate of 22/min and Blood Pressure of 136/90 mm of Hg. Abdominal examination did not reveal any significant finding and no guarding rigidity or local tenderness was observed. X-ray abdomen was normal which neither showed any gas under diaphragm nor any air fluid level.USG abdomen was also normal and patient was treated conservatively in surgery department. Next day patient started complaining of dyspnea and weakness in all 4 limbs. Patient was tachypneic and but his single breath count was normal. X-ray chest did not demonstrate any abnormality. Arterial blood gas analysis (ABG) was suggestive of hypercapnic respiratory failure with PCO_2 of 65 and PO_2 of 55.Neurological examination revealed areflexic flaccid paralysis. He was transferred to medical intensive care unit for intubation and ventilatory support. Serum potassium levels were normal (4.1meq/L). Tentative diagnosis of Acute intermittent porphyria was entertained and urinary porphyrin concentration was measured which came out to be elevated. To rule out Gullian Barre Syndrome (GBS) analysis of cerebrospinal fluid obtained by lumbar puncture was done which revealed normal total protein. Electromyogram and nerve conduction studies did not demonstrate signs of demyelination. Patient was treated symptomatically for pain relief, antibiotic cover was given and high carbohydrate diet (300mg/dl) was provided following which he recovered after 17 days and was discharged later.

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Introduction

Porphyrias are a group rare inherited or acquired disorders of certain enzymes in the heme bio-synthetic pathway. They are broadly classified as acute (hepatic) porphyrias and cutaneous (erythropoietic) porphyrias, based on the site of the overproduction and accumulation of the porphyrins (or their chemical precursors). They manifest with either neurological complications or skin problems (or occasionally both)[1].

Discussion

Acute intermittent porphyria (AIP) is a rare autosomal dominant disorder caused by a deficiency of porphobilinogen (PBG) deaminase. As compensation for the diminished PBG activity, activity of aminolevulinate dehydratase, which is under control of negative feedback from heme is increased[2,3].

Patients with AIP have acute episodes of neurovisceral problems consisting of autonomic neuropathies (eg, constipation, colicky abdominal pain, vomiting, and hypertension), peripheral neuropathy, seizures, delirium, coma and depression with accompanied increase in excretion of porphyrin precursors. The patients of AIP are known to present with acute abdominal pain (85%–95%) and peripheral neuropathy

with muscle weakness (42%–68%). Less common and less well-known is respiratory failure (9%–20%). The exact underlying mechanisms of these signs and symptoms are unknown[2,3,4].

Table 1:- Signs & Symptoms of AIP in decreasing frequency[4]

Sr. No	Signs & Symptoms
1	Abdominal Pain
2	Tachycardia
3	Dark urine
4	Peripheral motor neuropathy
5	Constipation
6	Nausea & vomiting
7	Mental changes
8	Hypertension
9	Absent reflexes
10	Back Pain
11	Sensory neuropathy
12	Postural hypotension
13	Convulsion
14	Chest pain
15	Coma

The precipitating factors for AIP are mostly inadequate nutrition, infection, alcohol, chemicals, stress, smoking, estrogen, and drugs. Certain drugs can exacerbate AIP by inducing the hepatic hemoprotein cytochrome P450, a situation that leads to a depletion of the free heme pool, resulting in induction of aminolevulinate dehydratase[2,3].In our patient alcohol intake might have acted as a precipitating factor leading to the consequences.

Management mainly consist of treating the precipitating cause , symptomatic relief for pain , a high-carbohydrate diet (300 g/dl) and if no response is seen then intravenous hemin (3 mg/kg) administration daily have produced favourable results. Recovery depends on the degree of neuronal damage and usually is rapid if therapy is started early. Liver transplant is a high risk procedure and should not be considered as an established treatment for acute porphyrias[3].

Figure 1:-
Urine collected (Bottle 1) which changed to reddish brown on standing for 15 min (Bottle 2)

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