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DIAGNOSIS OF ACUTE INTERMITTENT PORPHYRIA (AIP) IN A 20 YEARS OLD BOY: A CASE STUDY

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ABSTRACT

Acute Intermittent Porphyria (AIP) is one of the acute hepatic porphyria. It is a rare inherited autosomal dominant metabolic disorder. This disorder itself causes a severe neuropathic abdominal pain (that can mimic acute surgical abdomen) accompanied by a wide range of gastrointestinal, psychiatric and neurological symptoms. However, the diagnosis of acute Porphyria can be very demanding due to overlapping features amongst the various types. We reported the case in 20 years old male patient who presented with acute abdominal pain, seizures and rapid progressive weakness. Initially his symptoms were misdiagnosed as acute appendicitis resulting in negative appendicectomy thus delaying the needed definite treatment. After one year he was diagnosed as AIP. He improved and recovered with proper line of treatment.

Key words: porphyria, vanillylmandelic acid, acute abdominal pain, porphoblinogen deaminase

INTRODUCTION

Acute Intermittent Porphyria (AIP) is a rare innate metabolic abnormality. It is autosomal dominant type of Porphyria caused by genetic mutation of the 3rd enzyme named Porphobilinogen Deaminase (PBG Deaminase) also called Hydroxymethylbilane Synthase (HMBS) in heme biosynthesis (Anderson *et al*., 2001).

Clinically, AIP are recognized to heme reduction and accretion of intermediates PBG and aminolevulinic acid (ALA), proximal to the defect (Anyaegbu *et al.,* 2012). Multiple organ systems can be affected in patients with AIP.

The clinical symptoms such as vomiting, hypertension and tachycardia are considered to be acute attacks (Puy *et al.,* 2010). Moreover abdominal pain may simulate acute surgical abdomen leading to negative laparotomy (Kumar *et al.,* 2010). Peripheral neuropathy with muscle weakness is also very common symptoms of AIP (Anderson *et al*., 2005).

The eliminating of predisposing factor is imperative in this condition. Such drugs that may enhance the risk of disease need to be avoided. Excessive amount of carbohydrate, glucose administration, or hematin infusion may be recommended for AIP recovery (Bloomer and Bonkovsky, 1989).

CASE REPORT

Twenty years old male patient came in OPD at Abbasi Shaheed Hospital, Karachi with complain of abdominal pain. On the basis of proper history, the first episode of abdominal pain was noted in one year back which was sudden in onset, generalized type of pain, pricking in nature but did not go to back side. However, it was assumed as appendicitis on behalf of clinical analysis which was operated by the surgeon. Nevertheless after 5-6 months of operation, patient felt again abdominal pain (second episode) which subsided itself.

However, the patient came in hospital with third episode of abdominal pain, moreover nausea, vomiting and epileptic fits (fits persisted 10 to 15 minutes). Patient rapidly underwent progressive weakness followed by both the upper and lower limb that persisted up to 15 days and patient got bed ridden. Beside this, patient also complained about dark colored urine and some time constipation. Patient had not any history of parasthesias, sphincter disturbances and drug intake. On the basis of general physical examination; patient has normal height with thin built, normal pulse but episodic increased blood pressure. Abdominal examination was unremarkable. Neurological examination showed features of bilateral lower motor neuron type of weakness in upper limb which was more marked distally with bilateral wrist drop (Fig. **1**). Wasting of small muscles of hand was also noted but there was no loss of sensation in both limbs. Patient was recommended for laboratory investigation and following tests were carried out as shown in Table 1.

Table 1: Laboratory Investigation

Laboratory tests	Results	Normal value/ range
Hemoglobin (Hb)	10.3gm/dL	13.5-17.5gm/dL
Erythrocyte Sedimentation Rate (ESR)	20mm/ h	0-22mm/ h
Total Leukocyte Count (TLC)	6300/mm^3	4500 -11000/mm^3
Neutrophil	68%	47-77%
Lymphocytes	25%	16-43%
Eosinophils	4%	0.3-7%
PBG and coproporphyrins	Positive in Urine	-
Vanillylmandelic acid (VMA)	Negative in Urine	-
Urea	46 mg/dL	7-20 mg/dL
Creatinine	1.5 mg/dL	0.6-1.2 mg/dL
Na^+	124 mEq/L	136-145 mEq/L
K^+	3.1 mEq/L	3.5-5 mEq/L
Mg^+	1.7 mEq/L	1.5-2 mEq/L
Ca^+	9.1 mg/dL	8.5-10.2 mg/dL
Albumin	3.9 gm/dL	3.5-5.2 mg/dL
vitamin D3	11 ng/ mL	20-50 ng/ mL

Moreover Liver Function Tests (LFTs) was normal. Arterial blood gases shows pH: 7.32; pCO_2 was less as 27.6; HCO_3 was also low as 22.

The symptomatic treatment was given to the patient along with general supportive measures. Gradually patient appeared to be improved.

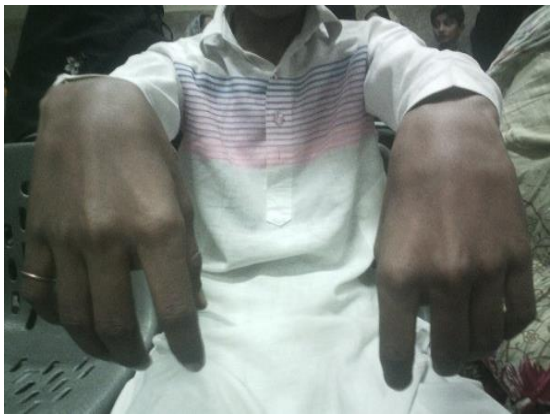

Fig.1. Bilateral wrist drop and wasting of small muscles of hand in patient with porphyria.

DISCUSSION

AIP is a dominant autosomal metabolic disease that disturbs the production of heme. It is caused by genetic mutation of the enzyme PBG deaminase (Anderson *et al*., 2001; Anyaegbu *et al*., 2012). AIP usually presents with non specific symptoms, so a good clinician and surgeon should keep a deep thoughts regarding Porphyria in differential diagnosis of acute abdomen. The disease is manifested by gastrointestinal and neuropsychiatric symptoms. The patient presents with recurrent acute abdominal pain (Bustamante *et al*., 1999) that can mimic acute surgical abdomen. Moreover fever, fast heart beat, hypertension, agitation, tremors, and over sweating are due to sympathetic excess activity. These findings were also discussed by other workers (Kauppinen, 2005; Puy *et al*., 2010; Suarez *et al*., 1997; Kumar *et al*., 2010; Anderson *et al*., 2005).

Acute attack can be very difficult to recognize because an individual sign and symptoms are non-specific, particularly in the early stages (Lowenstein *et al*., 1992). Acute attacks of AIP are often triggered by a number of factors like anticonvulsants, sulphonamides, low calorie diet, alcohol, hormones and numerous drugs (Suarez *et al*., 1997; Shah *et al.,* 2002) but contrary in present case there is no history of drug or alcohol intake.

The patient required proper management, treatment and monitoring of respiration rate, electrolytes balance, nutritional condition and moreover intravenous administration etc. Patient may need intensive care unit (ICU) in case of severity. The narcotic analgesics, chlorpromazine, or another phenothiazines agent are effective against pain, nausea and vomiting which are safe to use in acute condition of porphyrias. Short acting (SA) benzodiazepines in low doses are almost safe for nervousness and insomnia (Grandchamp *et al*., 1998). Approximately upto 350 g of carbohydrates is mandatory during attacks. If oral intake is insufficient, carbohydrates should be given intravenously. Dextrose reduces the urinary excretion of porphyrin precursors in patients with AIP (Bonkowsky *et al*., 1976).

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