ACUTE INTERMITTENT PORPHYRIA

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INTRODUCTION

Acute intermittent porphyria(AIP) is an autosomal dominant, severe form of metabolic disorder affecting females more than males, due to partial deficiency of the enzyme Porphobilinogen Deaminase(PBGD)[1]. This leads to excessive formation and urinary excretion of porphyrin precursors, aminolaevulinic acid and porphobilinogen[2]. The patients usually present with sudden severe abdominal pain lasting for several hours which may mimic acute abdomen with associated symptoms of vomiting, fever, diarrhea and painful micturition. There are a large number of factors which may precipitate an acute attack of acute intermittent porphyria. These include drugs, alcohol, fasting / starvation, stress, smoking, chemicals and excess of estrogens and progesterone.This case report is presented to emphasize the need for being vigilant for this rare metabolic disorder.

CASE REPORT

A 19 years soldier was referred from Combined Military Hospital Dera Ismail Khan to Military Hospital Rawalpindi with one week history of severe abdominal pain, vomiting, fever and seizures. Abdominal pain was sudden in onset, continuous, colicky, diffused in abdomen. In family history, his mother had similar complaints and was diagnosed as case of AIP. He also developed high grade, intermittent fever with no rigors and chills. On examination he was conscious, alert and oriented with vital signs of pulse 78/min, BP 135/80mmhg, R/R 23/min and temperature 99°F. Abdominal examination revealed mild tenderness in epigastrium. Rest of the systemic examination was unremarkable. Urine and blood for aminolevolunic acid (ALA) and PBG were positive. He was shifted to Medical Intensive Care Unit (ICU). A provisional diagnosis of Acute Intermittent Porphyria was made on the basis of positive family history, typical clinical features and presence of Porphobilinogen (PBG) in urine of the patient. Rest of the laboratory investigation were with in normal limits except MRI brain which showed a hyperintense area in posterior right occipital lobe (Figure).

He was treated with narcotic analgesics for abdominal pain, I/v glucose 300g/d. Intravenous hematin was not available in market. Patient was counseled in details for identification and avoidance of precipitating factor to fasten recovery and prevent future complications.

DISCUSSION

Porphyria or porphyrin is derived from Greek word "PORPHYRUS" which means "purple colour". It is classified into hepatic and erythropoietic on the basis of primary site of overproduction and accumulation of porphyrin and its precursors[3]. Symptomatic AIP is a severe form of acute hepatic porphyria, expressed after puberty, more commonly in women . It presents as abdominal pain which is the most common symptom. Other features include urinary incontinence, dysuria, fever, tachycardia, hypertension, sweating, tremor and Periphral motor neuropathy. Seizures may be seen in patients of AIP with hyponatremia due to vomiting or SIADH[4]. Psychiatric complaints like hysteria, anxiety, depression or psychosis may be seen in AIP[5].

Laboratory testing confirms or excludes the diagnosis by ALA and PBG determination both in urine and plasma . Caloric supplementation reduce excretion of ALA and PBG hence suppress the symptoms[5,6]. Precipitating factors include infections, surgery, drugs like barbiturates, sulfonamides etc, alcohol, stress,smoking, fasting/ starvation and estrogens/ progesterones. There is no definite treatment of AIP. By avoiding the precipitating factors (as discussed above) acute attacks can be prevented[10]. In case of an acute

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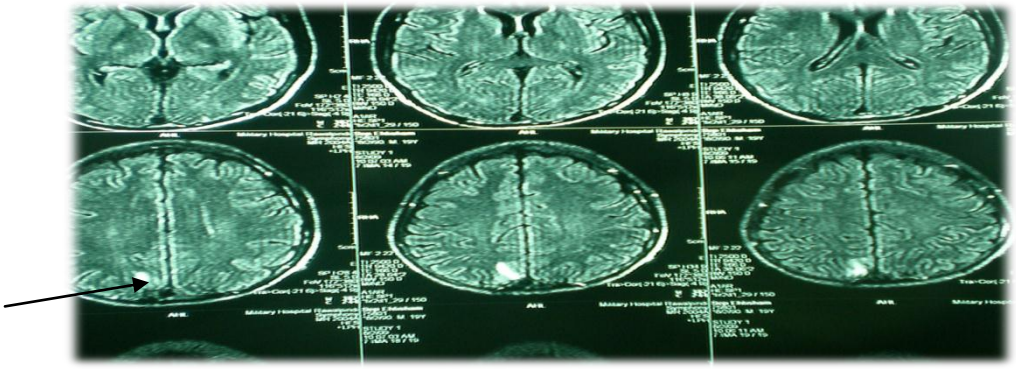

Figure: A hyperintense area with abnormal signal intensity in right posterior Occipital lobe

attack, symptomatic treatment is given. Abdominal pain is treated with opiod analgesics and vomiting with phenothiazines. Hypertension, electrolyte disturbances especially hyponatremia due to SIADH should be corrected. Intravenous 25% Dextrose 1-2 liters in 24 hours should be given. Hemin is given I/V in dose of 3-4 mg/kg over a period of 10-15 minutes once daily for four days[7].

Porphyria is a rare metabolic disease, due to its vague presentation, the cases are generally missed or not properly treated in time leading to mortality. Certain changes in life style, diet, avoidance of inducer drugs and chemicals can make the patients comfortable and productive. The highest degree of suspicion and vigilance in diagnosis and prompt treatment in time in AIP patients can produce excellent outlook.

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