CASE REPORT

A YOUNG GIRL WITH FITS

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A young girl presented with fits vomiting and epigastric pain. Investigations including CT-Scan brain, MRI brain, MRV brain and cerebrospinal fluid (CSF) examination were normal. Her urine was screened for porphobilinogen which was positive. She responded to intravenous dextrose and hypercaloric diet

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INTRODUCTION

Porphyrias are metabolic disorders caused by deficient activities of enzymes within the haeme-biosynthetic pathway. A deficiency of any of the enzymes in the biosynthetic pathway can lead to a variety of clinical symptoms.[1] In 1937, Waldenstrom in Sweden published his findings on Acute Intermittent Porphyria (AIP). By the 1960s, all known types of porphyrias had been identified. Research in the 1980s and 1990s led to the identification of the molecular defects in each type of Porphyria.[2]

AIP is an autosomal dominant disorder with a prevalence of 5 per 100 thousand people and results from deficiency of Porphobilinogen deaminase. Lack of clinical recognition often delays effective treatment.[3] Abdominal pain is the most common symptom which is usually severe and poorly localized. Positive urinary porphobilinogen, absence of fecal porphyrin and porphobilinogen deaminase mutation are diagnostic for acute intermittent porphyria.[4]

CASE REPORT

A 17 years old female presented in emergency department with 7 days history of pain epigastrium, vomiting, and fits. She also had constipation for 2 days. Vomitus was without any blood. A general practitioner had prescribed ciprofloxacin, metoclopramide, omeprazole and oral rehydration salt. She had two episodes of generalized tonic clonic fits. Fits were not associated with aura, tongue bite, urinary and fecal incontinence.

On admission she was confused, disoriented, dehydrated, with a pulse of 110 per minute and blood pressure of 85/50 mmHg. Tongue was dry and there was tenderness in epigastrium. Examination of the central nervous system revealed Glasgow Coma Scale (GCS) of 12/15, with no neck rigidity.

She was given intravenous fluids, Ceftriaxone, Acyclovir, Valproic acid, Omeprazole and Metoclopramide. Her blood complete picture, liver function tests, serum amylase, serum calcium, phosphate, ECG and ultrasound abdomen were within normal limits. Her serum urea was mildly raised (9 mmol/l). CT scan brain was normal. Analysis of CSF was also normal. On the 4th day of her admission, she had generalized tonic clonic fits. A neurologist's consultation was obtained. EEG, MRI, MRV Brain and CSF studies were repeated which were within normal limits. Tablet carbamazepine was added for fits. EEG showed diffuse cortical dysfunction. Upper GI endoscopy revealed mild antral gastropathy. Subsequently she developed vertigo and became ataxic. Carbamazepine was discontinued. Lamotrigine, Dexamethasone and Betahistine were started. Psychiatric consultation was also requested.

Though her fits were controlled but she still complained of pain epigastrium and vomiting and her general condition worsened.

On a subsequent reassessment of the case it was revealed that she had symptomatic relief after intravenous dextrose and her symptoms correlated with the onset of her menstrual cycle. One of her first cousins also suffered from a similar illness. A strong suspicion of porphyria was made and urine for porphobilinogen was requested which was reported positive. The saved urine darkened to reddish colour. Based on presence of neurovisceral symptoms, positive family history, symptomatic relief with intravenous dextrose and a positive test for porphobilinogen in urine, diagnosis of acute porphyria was made with a likelihood of Acute Intermittent Porphyria (AIP). All the drugs were stopped, 10% dextrose was given by vein and patient advised to take a carbohydrate rich diet.

She subsequently became asymptomatic and was discharged to home. She was advised to take plenty of carbohydrates. A medic alert bracelet for Porphyria was given. Urinary screening of the rest of her family was negative

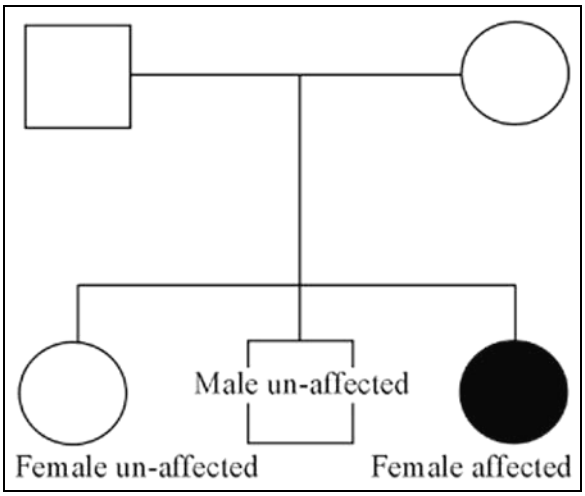

Figure-1: Family tree of the patient

DISCUSSION

Acute Intermittent Porphyria has a varied and non-specific presentation. It should be considered as a differential diagnosis in individuals who present with features of abdominal pain especially in combination with neuropsychiatric manifestations. Delay in diagnosis not only leads to unnecessary investigations but also puts patient's life at risk. High index of suspicion and vigilance is required for diagnosis. Intravenous hematin in a dose of 3–4 mg/kg is effective but haem arginate is believed to be safer.[5] Other measures like intravenous dextrose and high carbohydrate diet are also useful to reduce the severity of the attack. Before prescribing any medication for such patients it is important to be aware of the safe and unsafe drugs for this illness. DNA analysis has improved the detection of presymptomatic heterozygotes in AIP families.[6] Unfortunately, there is no curative treatment for any type of Porphyria but timely treatment and avoidance of aggravating factors and drugs prevent long term complications. Haem arginate being the treatment of choice should be made available.

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