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RESEARCH ARTICLE

ACUTE INTERMITTENT PORPHYRIA IN MALE

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Abstract

Acute intermittent porphyria presenting with short duration of gastrointestinal symptoms followed by rapidly progressive fulminant neurological syndrome during first attack is relatively uncommon. It is a neurological emergency and mimics many other psychiatric and medical disorders and can be fatal if it remains undiagnosed and untreated. So high clinical suspicion and early diagnosis and management of acute attack and prevention of further attacks are very important. We report a case of AIP with abdominal pain and convulsion to highlight this fact.

Case report:-

A 20 year old male presented to the emergency room with complaints of not passing stools since 1 week, 6 episodes of non-bilious vomiting's, with food and water as contents since last 2 days and increasing severity of abdominal pain since the last 2 days. History revealed no premorbidities, no similar episodes and no episode of seizures. Systemic examination decreased bowel sounds. He was kept under observation suspecting sub-acute intestinal obstruction.

2 days later patient developed seizures. Blood and urine samples were collected. His blood reports revealed low serum sodium, which was corrected slowly with fluid restriction and hypertonic saline. Blood investigations done to evaluate the cause which favoured SIADH. Urine in Urine bag was dark coloured on standing. After recovery patient developed mild psychiatric symptoms in the form of mood disturbance. MRI brain and CT abdomen were normal.

Based on the varied presentation and darkening of urine colour and suspicion of AIP. Urine samples were sent, which revealed increased total urinary porphyrins, very high levels of porphyrobilinogen and increased 5-aminolevulinic acid.
All of these point towards a diagnosis of ACUTE INTERMITTENT PORPHYRIA.

Discussion:-

Acute intermittent porphyria (AIP) is an autosomal dominant metabolic disorder that has varied manifestations. In AIP there is an abnormality in the haem biosynthetic pathway due to the deficiency of uroporphobilinogen I synthetase (porphobilinogen deaminase), which leads to excessive production of porphyrin precursors. The clinical features of AIP declare themselves at any stage from puberty onwards but mostly in the second and third decade of life. Penetrance of the gene responsible is incomplete, so that the disease often exists in a latent form and a family history may not be forthcoming.

The prevalence of AIP in the general adult population ranges between 1 and 8 per lakh. Clinical symptoms are rare before puberty and the diagnosis of AIP is suggested by a triad of symptoms (abdominal pain, neuropathy and mental changes) and porphobilinogen in the urine.

The severe constipation and episode of abdominal pain, hypertension and tachycardia seen during an acute attack of porphyria may be because of an autonomic neuropathy.

Convulsion can occur at any time during acute illness but acute symptomatic seizures are a well-recognized feature of porphyria in relapse and subsequent use of enzyme-inducing antiepileptic drugs can cause a worsening of condition and new symptoms, such as acute neuropathy and respiratory paralysis. Further, it is less well documented that these manifestations can occur as a rapid progressive course during the first attack of AIP.

Mental disorders accompany attacks in 24% to 80% of cases and psychiatric symptoms can dominate the picture. Psychiatric manifestations include hysteria, anxiety, depression, phobias, psychosis, organic disorders, agitation, delirium and altered consciousness ranging from somnolence to coma, and the most common neuropsychiatric manifestation reported are delirium and depression.

Heme arginate (IV) is the treatment of choice and very effective if given early in the course (within one-two days) of illness and leads to biochemical reemission followed by clinical improvement in one to two weeks but it is less effective if treatment is delayed. Limitations for using heme arginate are that it is not universally available, very costly and can cause severe coagulopathy and anaphylactic reactions sometimes.

Conclusion:-

High clinical suspicion and early diagnosis and effective management of acute attacks is very important in reducing mortality and morbidity. Acute intermittent porphyria should always be considered as differential diagnosis in case of abdominal pain with neuropsychiatric manifestation, whether family history of AIP is present or not. Finally, once the diagnosis has been confirmed, the family members must be screened for the PBGD enzyme level and/or mutation analysis to diagnose asymptomatic carriers.

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