



## ACUTE INTERMITTENT PORPHYRIA

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## KEYWORDS :

## INTRODUCTION

Acute intermittent porphyria is rare autosomal dominant disease due to deficiency of hydroxymethylbilane synthase (HMB synthase). Heme synthesis process is multi step process that involves specific enzyme at every steps.

Drugs ,diet ,hormones activate disease.

AIP is most common porphyria worldwide with prevalence of AIP is approximately 5 per 1 lakh population. AIP affects women more than men ,with ratio of 1.5-2 : 1.

Most patient becomes symptomatic at age 18-40 years. Attacks before puberty or after age 40 are unusual unless major provocation

In india majority of cases reported from kumhar community in western Rajasthan.

## Clinical Presentation

21 year girl presented with abdominal pain,nausea ,vomiting since 5days and multiple episodes of seizure followed by altered sensorium since 1 day prior to admission.

No signs and symptoms of meningitis or raised ICT

No h/o fever

No h/o psychological symptoms,

No h/o drugs or toxin exposure

No h/o passage of dark colored urine.

No h/o photosensitivity rash.

On admission pulse – 130bpm, BP- 130/80

RR-24cpm ,RS- bilateral conducted sound

CVS: HSWNL

Per abdomen: Soft ,non tender, no organomegaly

CNS: GCS E1VTM1 ,no localising signs ,no focal deficit

## Investigation

Hb: 11g/dl

TC:16000 N=58 % L=27% B=7%

Platelet=2.9 lakh

Serum urea=20

Serum creatinine = 1.0mg/ dl

Serum sodium = 119 meq /L at time of admission which improved to 136 mEq/L on treatment

Serum uric acid=2.7 mg/dl

Usg abdomen ,CT brain ,EEG ,CSF analysis were normal

URINE PORPHOBILINOGEN =detected

24 hours 5 aminolevulinic acid =15.28 mg/24 hours

Diagnosis of porphyria is suspected due to history of abdominal pain , seizure . And treated with following medication

Carbohydrate load of 300 kcal

Tab .gabapentin 300mg TDS

Tab. Levepsy 500 mg TDS

After treatment patient improved properly and went home after decannulating ET tube.

On follow up no further seizure episodes.

- Acute intermittent porphyria is rare genetic disease
- Presents with acute abdominal pain ,high degree of suspicion is required to diagnose case.

- Prompt diagnosis and avoidance of precipitating factors , hemin infusion are main stay of treatment

## REFERENCES

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