**ACUTE INTERMITTENT PORPHYRIA**

**J. Siva Somana**, T. **Uma**

1 Assistant Professor, Department of Biochemistry, Government Thoothukudi Medical College, Thoothukudi.
2 Junior Consultant, Department of Biochemistry, IIOG Hospital, Chennai.

**ABSTRACT**

**BACKGROUND**

Acute intermittent porphyria is an autosomal dominant condition due to deficiency of porphobilinogen deaminase with variable clinical expression and aggravated by drugs, diet, etc.

**Clinical History**

A 17-year-old male was brought to the medical ward with history of fever and on treatment with antimalarials, porphyria got aggravated. Clinical findings: patient was unconscious with GCS-8, pupils dilated but reacting to light. Patient was investigated and the results were as follows: CBC showed anaemia and thrombocytopenia. Urinary PBG and ALA excretion are high. We hereby report an unusual case of AIP presenting with PRES, seizures, and EEG changes.

**CONCLUSION**

Both clinical profile and laboratory investigations are essential for diagnosis. An acute attack of porphyria is based primarily on clinical features even when it is biochemically proven as acute intermittent porphyria.

**KEYWORDS**

Acute Intermittent Porphyria, Variable Clinical Expression, PRES, Porphobilinogen (PBG), Delta-aminolevulinic Acid (ALA).

**HOW TO CITE THIS ARTICLE:** Somana JS, Uma T. Acute intermittent porphyria. Journal of Evolution of Research in Medical Biochemistry 2017; Vol. 3, Issue 1, Jan-June 2017; Page:5-6.

**BACKGROUND**

Acute intermittent porphyria is an autosomal dominant condition due to deficiency of porphobilinogen deaminase and the enzyme activity will be half of normal activity in all tissues where they are expressed. Variable expression due to low clinical penetrance is prominent. Affected individuals are mostly asymptomatic throughout life. Heterozygotes experience acute attacks of porphyria at incidence rates of 1 in 10 to 1 in 5 times during their lifetime.

**Case Report**

A 17-year-old male was brought to Medicine Department with complaints of fever of 5 days’ duration and was investigated for the same. Patient was treated with an antimalarial, Tab. Primaquine. One day in the ward, patient developed seizures and was shifted to ICU.

**Family History**

No significant history.

**On Examination**

Patient was unconscious, afebrile, high blood pressure (BP = 170/110 mmHg).

**CVS and RS Examination**

Normal.

**Investigations**

1. CBC: Anaemia and Thrombocytopenia.
2. Liver function test: Normal.
5. Urine PBG level was 154.4 μmol/L (reference: 0 to 0.88 μmol/L).
7. Qualitative tests for ALA was positive.
8. Hyponatraemia with Na+ = 128 mmol/L.

**Treatment**

DISCUSSION

Acute intermittent porphyria is an autosomal dominant condition due to deficiency of porphobilinogen deaminase gene that maps to human chromosome 11q23. It is the commonest acute porphyria. Acute neurovisceral attacks are potentially life-threatening without proper diagnosis, management, and elimination of triggers. Attacks include abdominal pain, psychiatric symptoms, signs of sympathetic and hypothalamic autonomic overactivity accompanied by convulsions and motor and sensory deficits at times and also precipitated by drugs inducing hepatic haem formation and metabolised by the hepatic cytochrome P-450 system. Cutaneous photosensitivity does not occur. Urine will be normal in colour, but pink discolouration occurs due to the formation of coloured oxidation products of porphobilinogen from the porphyrin precursors. Exacerbations are secondary to multiple environmental and physiological factors, including drugs, infection, hormonal changes, or fasting. AIP is characterised by markedly increased urine ALA, PBG levels and normal faecal porphyrin levels.

CONCLUSION

Our patient is a case of acute intermittent porphyria, presented as PRES because of which the patient had seizures and became unconscious, and the patient was recovering with supportive treatment. An acute attack of porphyria is based primarily on clinical features even when it is biochemically proven as acute intermittent porphyria.

REFERENCES