

## INTRODUCTION

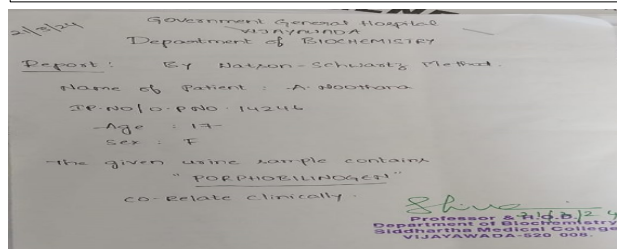
Acute intermittent porphyria (AIP) is an inherited metabolic disease due to a deficiency of the hydroxymethylbilane synthase in the heme biosynthesis. AIP can cause severe neurological symptoms involving the central, autonomic, and peripheral nervous system. Due to their relative rarity and their chameleon-like presentation, delayed diagnosis and misdiagnosis are common.

## CASE REPORT

A 17 year old female born out of non consanguineous parentage p/w seizures of GTCS semiology and abdominal pain of 1 day duration. She is a k/c/o seizure disorder of 2years duration( on brevetiracetam 50mg BD) and seizures episodes were associated with hyponatremia( treated with 3% NaCl in out side hospital). She also had recurrent episodes of abdominal pain with vomitings of 2 months duration. There is history of psychiatric disturbances in the form of paranormal behaviour of 1 month duration. There is no h/o fever, altered sensorium, loss of weight or appetite. No family h/o similar complaints. Her neurologic and systemic examination were found to be normal

## INVESTIGATIONS

Routine investigations including S.Electrolytes were done, hyponatremia with S.NA=117meq/l was noticed, and evaluated for the causes of hyponatremia. S.Osmolality=233mosm/kg, U.Osmolality:310mosm/kg, U.NA=24 meq/l found to be euvoletic hyponatremia, then thyroid profile and S.Cortisol were done and found to be normal and diagnosed as SIADH. In view of neurologic and abdominal symptoms, urine porphobilinogen was sent and it was positive-80 mg/24hrs. MRI Brain, CSF analysis, CT Abdomen, NCS and EEG were normal. Urine color was normal. Genetic analysis revealed heterozygous mutation in HMBS gene indicative of AIP.



<b>MedGenome Labs Ltd.</b> <small>1st Floor, Narayana Medical Building, Narayana Health City, #250/A, Saranavaram, Houti Road, Bangalore - 560 095, India. Tel : +91 8860 87 18888 / 8860. Web: www.medgenome.com</small>		 <b>MEDGENOME</b> <small>Genetic Testing   Sequencing   Microarray</small>				
<b>DNA TEST REPORT - MEDGENOME LABS</b>						
Full Name / Ref No:	<b>A. NUTANA</b>	Order ID/Sample ID:	<b>888023/8424839</b>			
Gender:	Female	Sample Type:	Blood			
Date of Birth / Age:	18 years	Date of Sample Collection:	30 <sup>th</sup> March 2024			
Referring Clinician:	Dr. Evangelin Blessey G.	Date of Sample Receipt:	31 <sup>st</sup> March 2024			
Test Requested:	Genetic Testing - Sequencing	Date of Report:	31 <sup>st</sup> March 2024			
<b>CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY</b> Ms. A. Nutana, presented with clinical indications of hyponatremia secondary to SIADH and acute intermittent porphyria, idiopathic generalized seizures, generalized tonic-clonic seizures, abdominal pain. Her complete blood picture with peripheral smear showed normocytic hypochromic RBC and neutrophilic leukocytosis. Her CT abdomen showed distended gall bladder and a few small stones in distal kidneys. Ms. A. Nutana has been evaluated for pathogenic variations.						
<b>RESULTS</b> PATHOLOGIC VARIANT CALIBRATED OF THE REPORTED PHENOTYPE WAS DETECTED						
<b>SNV(s)/INDEL</b>						
Gene(s) Transcribed	Location	Variants	Significance	Disease (OMIM)	Inheritance	Classification
HMBS (4)	1p34.3	c.517C>T (p.Arg177Trp)	Heterozygous	Acute Intermittent Porphyria	Autosomal dominant	Pathogenic

## RESULTS & DISCUSSION

Porphyrias are a group of autosomal dominant inherited disorders and the prevalence of acute porphyrias is 5 cases per 100,000 persons. The clinical expression of porphyrias is also exceptionally variable, as 90% of heterozygotes remain asymptomatic throughout life. In those that are clinically symptomatic, acute attacks are usually more common in females and manifest after puberty. The classic triad of AIP includes recurrent abdominal pain, neurologic dysfunction and psychiatric disturbances and our patient had all the 3 symptoms. Abdominal pain is the most common and earliest symptom of an attack, occurring in over 90% of patients, other autonomic manifestations like tachycardia, labile hypertension are prevalent in many cases but are absent in our case. Only a few patients progress to more ominous motor neuropathy( absent in our patient) or CNS involvement. Usually, peripheral neuropathy is considered the most common neurological manifestation of AIP and epilepsy occurs in up to 20% of AIP patients. Patients with variegate porphyrias and hereditary coproporphyria develop cutaneous photosensitivity. Precipitating factors include menstruation, alcohol, fasting, certain drugs. Early recognition, supportive and intravenous hematin therapy, and withdrawal of the precipitating factors are the key steps in the management of acute attacks of AIP.

## CONCLUSION

This “little imitator” is often missed or wrongly diagnosed because of its heterogeneous symptoms. Our case is unique in that it has seizures with hyponatremia as the initial manifestation rather than abdominal pain, and our case has all the features of triad. AIP should always be suspected in patients with unexplained abdominal pain and seizures.

**REFERENCES;** 1). Gerischer, L.M., Scheibe, F., Nümann, A., Köhnlein, M., Stölzel, U., & Meisel, A. (2021). Acute porphyrias-a neurological perspective. *Brain and Behavior*, 11.  
 2) Bissell, D.M., Anderson, K.E., & Bonkovsky, H.L. (2017). Porphyria. *New England Journal of Medicine*, 377(9), 862–872.