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CASE REPORT

Acute Intermittent Porphyria: Report of a Case

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Acute intermittent porphyria (AIP) is an inherited metabolic error in which delta-aminolevulinic acid (ALA) and porphobilinogen (PBG) are formed excessively in the liver. It results from a partial deficiency of PBG deaminase. Clinically, it is characterized by gastrointestinal symptoms and involvement of the nervous system. Here, we report a 24-year-old female who experienced her first attack of acute porphyria characterized by severe abdominal pain, hypertension, tachycardia, dark-brownish urine, seizure, proximal muscle weakness, and hyponatremia. The hyponatremia resulted from the syndrome of inappropriate secretion of anti-diuretic hormone (SIADH) according to the studies of urine and blood. Positive Watson-Schwartz test and increased level of urinary coproporphyrins were found during acute attack. Nine months later, in the asymtomatic stage, the Watson-Schwartz test was still positive. All these findings are consistent with the diagnosis of AIP. Fluid restriction and propranolol (Inderal) 120 mg daily were given and the patient recovered gradually. Because AIP is inherited in an autosomal dominant fashion and successive case reports have been presented recently, we think the prevalence of AIP in Taiwan may not be very low. Any patient presented with unexplained abdominal pain, dark urine, and neuropsychiatric symptoms should perform a Watson-Schwartz test for screening.

Key words: *acute intermittent porphyria (AIP), Watson-Schwartz test, syndrome of inappropriate secretion of anti-diuretic hormone (SIADH).*

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Porphyrias are diseases resulting from hereditary or acquired enzymatic deficiencies of the biosynthetic pathway of heme (Figure 1). In 1954, Schmid *et al* [1] classified the porphyrias as either erythropoietic or hepatic, based on the site of overproduction of the heme precursors.

The acute hepatic porphyrias, which include acute intermittent porphyria (AIP), hereditary coproporphyria, and variegate porphyria, were characterized by abdominal pain, tachycardia, hypertension, neuropathy , seizure, the syndrome of inappropriate secretion of anti-diuretic hormone (SIADH), and photosensitivity [2]. They are inherited in an autosomal dominant fashion. The basic genetic defect in AIP, hereditary coproporphyria, and variegate porphyria are a deficiency of porphobillinogen (PBG) deaminase, coproporphyrinogen oxidase, and protoporphyrinogen oxidase (or a type of defect in ferrochelatase), respectively. Owing to their various clinical manifestations, acute hepatic porphyrias are easily misdiagnosed. We hereby describe a case of AIP and review the literature thoroughly to correlate the reported clinical features of the patient.

CASE REPORT

This 24-year-old female was admitted on March 3, 1985, because of severe abdominal pain, a generalized seizure and dark urine.

She had been troubled with emotional instability for 2 years. In addition, abdominal pain, dark-brownish urine, constipation, and a nasal voice were noted occasionally about 4 months prior to this admission. On March 3, 1985, a sudden onset of

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