CASE REPORT

Acute Intermittent Porphyria with Transient Cortical Blindness

JAVEED IQBAL BHAT, UMAR AMIN QUREESHI AND MUSHTAQ AHMAD BHAT

From the Department of Pediatrics, Sher-I- Kashmir Institute of Medical Science, Soura, Srinagar, J&K, India.

Correspondence to:
Dr Mushtaq Bhat,
Associate Professor,
Pediatrics, SKIMS, Soura,
Srinagar 190 019,
Jammu & Kashmir, India.
Mbhat47@rediffmail.com
Manuscript received: May 25, 2009;
Initial review: June 29, 2009;
Accepted: August 11, 2009.

Acute intermittent porphyria is a hereditary disorder characterized by deficient activity of the enzyme porphobilinogen deaminase. It manifests with occasional neurovisceral crises due to overproduction of porphyrin precursors. We report a 12 year old male child with acute intermittent porphyria, who presented with encephalopathy and transient blindness of cerebral origin.

Key words: *Blindness, Hypertensive encephalopathy, Porphyria.*

Acute intermittent porphyria (AIP) manifests with occasional neurovisceral crises due to overproduction of porphyrin precursors such as aminolaevulinic acid (ALA)(1). We report a rare presentation of acute intermittent porphyria as hypertensive encephalo-pathy and transient cortical blindness.

CASE REPORT

A 12 year old male child was admitted with history of chronic intermittent abdominal pain since two months and two day history of hallucinations and deterio-ration of the consciousness. On examination he was afebrile, heart rate was 120 beats/minute and blood pressure was 170/110 mm Hg. General examination was unremarkable, the abdomen was not tender, and there was no hepatosplenomegaly. He was confused and disoriented in time and space. There were no other neurological signs except for bilateral papilledema. Routine baseline workup including hemogram, blood culture, liver function, kidney function, lumbar puncture, chest *X*-ray, ECG, and EEG were normal. Electrolyte screening showed mild hyponatremia (serum sodium 126 mmol/L). Ultrasonography of abdomen was unremarkable and serum lead levels were within normal limits. Sudden onset of bilateral blindness occurred three days after admission. Computed tomography of the brain showed hypodense lesions in both occipital lobes. In view of chronic abdominal pain and CNS symptoms, porphobilinogen (PBG) levels were obtained. Urinary porphobilinogen levels were substantially increased, (20mg/day). There was no history of any prior drug intake, the family members were screened for the disorder and none among them was found to be positive. Patient was managed in intensive care unit. Blood pressure was initially controlled with IV hypertensives and subsequently patient was put on oral calcium channel blockers. During the course of hospitalization his hypertension resolved. Patient regained consiousness and on neurological examination, no eye field defect was found on day 10^{th} of hospitalization. Patient was discharged after 20 days with complete recovery of symptoms, and as of 2 months from discharge patient's blood pressure is under control and has not experienced any new symptoms.

DISCUSSION

Peripheral neuropathy is the commonest neurological manifestation of acute intermittent

porphyria (AIP)(2). Muscle weakness often begins proximally in the legs but may involve the arms or the legs distally. Acute intermittent porphyria presenting as acute cortical blindness is rare. Kupferschmidt, *et al*.(3) reported two cases with acute intermittent porphyria presenting as cortical blindness, both were adult patients. We report a 12 year old child who presented with encephalopathy and subsequently developed cortical blindness during hospitalization. Cortical blindness in acute intermittent porphyria may be as a consequence of arterial vasospasm. The pathogenesis is not fully understood, it may occur as a result of depletion of nitric oxide synthase levels, a known vasodilatory enzyme in CNS(4). Depletion of nitric oxide synthase may be associated with unopposed vasoconstriction leading to focal hypodense lesions. Nitric oxide synthase, an important enzyme for synthesis of NO, is a cytochrome P-450 type hemoprotein whose activity may be reduced in situations of decreased heme production such as acute intermittent porphyria(5). However, our patient also presented with hypertensive encephalopathy as evidenced by blood pressure of 170/110, encephalopathy and bilateral papilledema. Transient cortical blindness with bilateral occipital hypodensities can also occur in hypertensive encephalopathy(6). It has been suggested that the bioccipital distribution of the lesions in patients with hypertensive encephalopathy may be due to the distribution of adrenergic receptors. In hypertension, stimulation of the perivascular sympathetic nerves results in increased vascular resistance, which thus protects the brain(7). In the vertebrobasilar system, however, breakthrough of autoregulation occurs because of sparse sympathetic innervation. Thus, the occipital lobes can be preferentially affected. Whether transient blindness in our patient was a consequence of acute intermittent porphyria *per se* or secondary to hypertensive encephalopathy could not be ascertained.

Contributors: MB prepared the manuscript and supervised case management. JIB and UAQ diagnosed and managed the case and helped in manuscript writing. All authors approved the final paper.

Funding: None.

Competing interests: None stated.

REFERENCES

1. Kauppinen R. Porphyrias. Lancet 2005; 365: 241-252.
2. Stein JA, Tschudy DP. Acute intermittent porphyria: a clinical and biochemical study of 46 patients. Medicine 1970; 49: 1-16.
3. Kupferschmidt H, Bont A, Schnorf H, Landis T, Walter E, Peter J, *et al*. Transient cortical blindness and bioccipital brain lesions in two patients with acute intermittent porphyria. Ann Intern Med 1995; 123: 598-600.
4. Lowenstein CJ, Snyder SH. Nitric oxide, a novel biologic messenger. Cell 1992; 70: 705-707.
5. White KA, Marletta MA. Nitric oxide synthase is a cytochrome P-450 type hemoprotein. Biochemistry 1992; 31: 6627-6631.
6. Marra TR, Shah M, Mikus MA. Transient cortical blindness due to hypertensive encephalopathy. Magnetic resonance imaging correlation. J Clin Neuroophthalmol 1993; 13: 35-37.
7. Beausang-Linder M, Bill A. Cerebral circulation in acute arterial hypertension-protective effects of sympathetic nervous activity. Acta Physiol Scand 1981; 111:193-199.