Pediatric Neurology 51 (2014) 457–460

Contents lists available at ScienceDirect

Pediatric Neurology

journal homepage: www.elsevier.com/locate/pnu

Clinical Observations

Posterior Reversible Encephalopathy Syndrome in Acute Intermittent Porphyria

Bi Zhao MD [a], QianQian Wei MD [a], YunHan Wang MD [b], YongPing Chen MD [a], HuiFang Shang MD [a,*]

[a] *Department of Neurology, West China Hospital, Sichuan University, Chengdu, Sichuan Province, China*
[b] *Department of Radiology, Chengdu First People's Hospital, Chengdu, Sichuan Province, China*

ABSTRACT

BACKGROUND: Acute intermittent porphyria is an inherited disease that is rarely diagnosed in prepubertal children. It can affect the autonomic, peripheral, and central nervous system. Posterior reversible encephalopathy syndrome is a clinicoradiological entity characterized by headache, seizures, altered consciousness, and visual disorder associated with potentially reversible neuroradiological abnormalities predominantly in the parieto-occipital lobes. We report a child with acute intermittent porphyria who presented with radiological manifestations suggestive of posterior reversible encephalopathy syndrome. **PATIENT:** A 9-year-old girl underwent an appendectomy after developing abdominal pain. She subsequently developed bilateral visual disturbance, confusion, seizures, hypertension, tachycardia, nausea, vomiting, constipation, dark tea-colored urine, and recurrent abdominal pain. **RESULTS:** Initial brain magnetic resonance imaging revealed hyperintense gyriform lesions on T_2-weighted images and hypointense to isointense lesions on T_1-weighted images in both parieto-occipital lobes with mild enhancement. The diagnosis of acute intermittent porphyria was confirmed by increased urinary excretion of porphyrin precursors. Her clinical signs gradually improved after intravenous high-dose glucose treatment and symptomatic therapies. A repeat magnetic resonance imaging confirmed complete resolution of the parieto-occipital lesions, suggesting with posterior reversible encephalopathy syndrome. **CONCLUSIONS:** The association of abdominal pain, mental status changes, and autonomic dysfunction should arouse the suspicion of acute intermittent porphyria. Acute intermittent porphyria can be associated with posterior reversible encephalopathy syndrome.

Keywords: acute intermittent porphyria, posterior reversible encephalopathy syndrome, abdominal pain, magnetic resonance imaging

Pediatr Neurol 2014; 51: 457-460

Introduction

Acute intermittent porphyria (AIP) is an autosomal dominant metabolic disease characterized by deficient activity of porphobilinogen (PBG) deaminase, a necessary enzyme in the heme biosynthetic pathway.[1] AIP can affect the autonomic, peripheral, and central nervous system (CNS) because of heme depletion and accumulation of heme intermediates PBG and δ-aminolevulinic acid (ALA).[1,2]

Posterior reversible encephalopathy syndrome (PRES) is a clinicoradiological entity characterized by headache, altered mental status, seizures, and cortical blindness associated with a potentially reversible imaging pattern, predominantly white matter, and gray matter abnormalities of the parieto-occipital lobes. In most patients, abnormalities present acutely or subacutely in the setting of accelerated hypertension, eclampsia or pre-eclampsia, renal failure, autoimmune disease, immunosuppressive treatment, or cancer chemotherapy.[3,4] However, PRES has rarely been recognized in patients with AIP, especially in children.[4-12]

B.Z. and Q.Q.W. were equally responsible for the work described in this article as first authors.

Article History:
Received February 9, 2014; Accepted in final form May 15, 2014

* Communications should be addressed to: Dr. Shang; Department of Neurology; West China Hospital; Sichuan University; No 37; Guo Xue Road; Chengdu, Sichuan 610041, China.
E-mail address: hfshang2002@126.com

0887-8994/$ - see front matter © 2014 Elsevier Inc. All rights reserved.
http://dx.doi.org/10.1016/j.pediatrneurol.2014.05.016

We describe a 9-year-old girl with AIP, who presented with reversible cerebral lesions consistent with PRES on magnetic resonance imaging (MRI).

Patient Description

A 9-year-old girl had an appendectomy after developing severe generalized abdominal pain without rebound tenderness and muscle guarding; the pathologic examination revealed no inflammation of the appendix. Sudden-onset bilateral visual disturbance occurred 2 days after the surgery. On the third postoperative day, she became confused and had two generalized tonic-clonic seizures for which she received phenytoin. On that same day, MRI of the brain performed at a local hospital demonstrated hyperintense gyriform lesions on T_2-weighted images and hypointense to isointense lesions on T_1-weighted images in both parieto-occipital lobes with mild contrast enhancement (Fig 1). She was then transferred to our hospital for further investigation.

At the time of admission, she was afebrile; blood pressure was 175/110 mm Hg, heart rate was 124 beats/min with regular heart sounds, and no murmurs were appreciated. She was confused and disoriented to time and place. Her visual acuity was decreased to light perception only, but the pupillary light reflex was preserved. The remaining cranial nerves functioned normally. Deep tendon reflexes and muscle power of the extremities were normal.

Laboratory studies including hemogram, liver function, kidney function, serum electrolytes, and osmolarity were normal. Tests for connective tissue disorders, paraneoplastic antibodies, viral infections, thyroid function, antithyroid antibodies, serum electrophoresis, and antiphospholipid antibodies were all negative. Plasma lead level was normal. Her cerebrospinal fluid was normal. Electroencephalography revealed generalized slowing in both hemispheres but no epileptiform discharges.

Two days later, she experienced nausea, vomiting, and constipation. Meanwhile, the abdominal pain reappeared. At this point, dark tea-colored urine became evident.

Although the girl had no family history of porphyria, AIP was considered because of the combination of CNS abnormalities (seizures and mental status changes), dysautonomia (hypertension and tachycardia), abdominal abnormalities, and the dark-colored urine, and a urine sample protected from light was sent for porphyrin metabolites analysis. The diagnosis of AIP was further confirmed by the following biochemical tests: PBG in the urine was 74 mg/24 hr (normal range $<$ 2 mg/24 hr), ALA in the urine was 107 mg/24 hr (normal range, 1-7 mg/24 hr), total porphyrins in the urine was 2123 μg/24 hr (normal range $<$ 200 μg/24 hr), and a decreased activity of erythrocyte PBG deaminase (50% of normal).

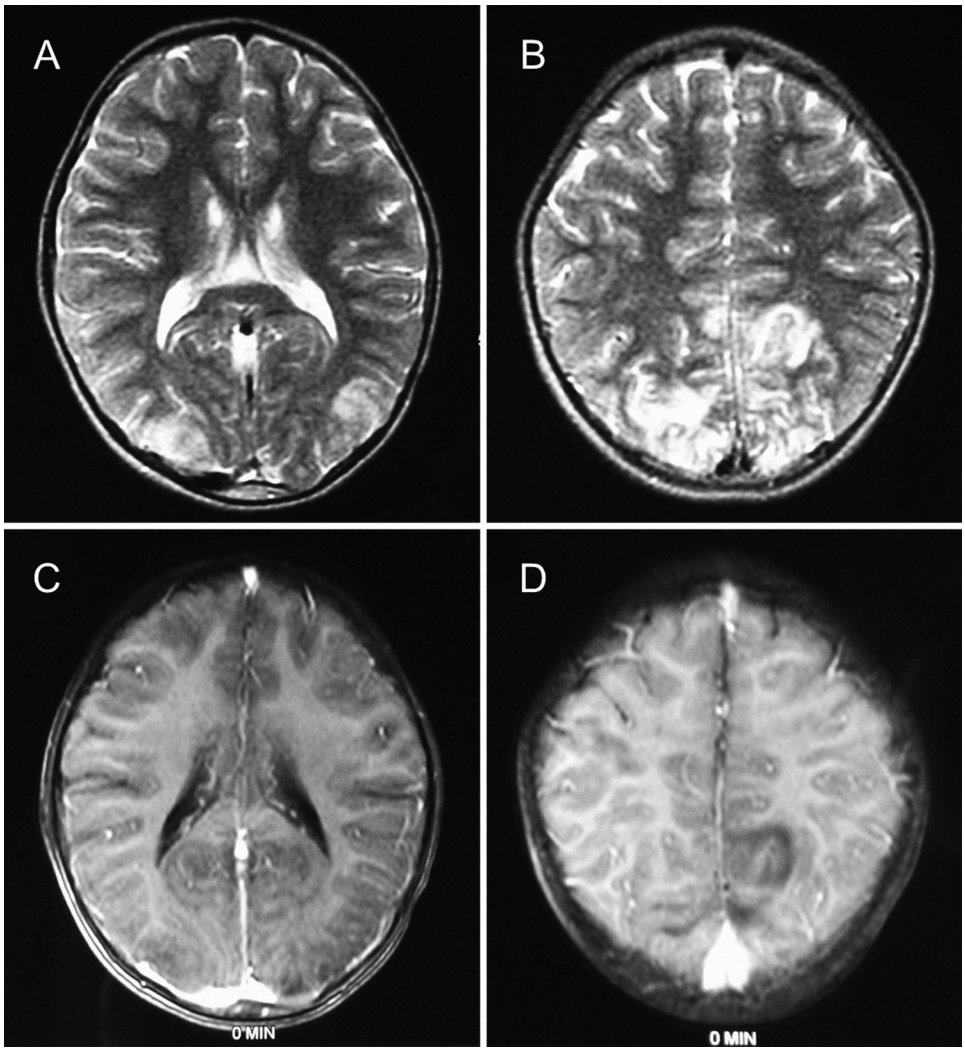

FIGURE 1.
Brain magnetic resonance imaging performed on the third postoperative day. Axial T_2-weighted images (A and B) revealed hyperintense gyriform lesions in both parieto-occipital lobes. Axial contrast-enhanced T_1-weighted images (C and D) revealed hypointense to isointense lesions with mild enhancement in both parieto-occipital lobes.