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MR imaging of acute intermittent porphyria mimicking reversible posterior leukoencephalopathy syndrome

Abstract Reversible posterior leukoencephalopathy syndrome (PLS) is characterized by headache, altered mental function, visual disturbances and seizures. Neuroimaging studies suggest a white-matter oedema, predominantly in the posterior parietal-temporal-occipital regions of the brain. We present the case of a 30-year-old woman who had suffered her first attack of acute intermittent porphyria (AIP). Following 1 week of abdominal pain she developed several generalized seizures, and hallucinations, and exhibited a progressive deterioration of the consciousness. T2-weighted images, especially fluid-attenuated inversion recovery (FLAIR) sequences showed bilateral lesions in the posterior frontal, parietal and occipital cortex and subcortical white matter. Following treatment with haematin and a high carbohydrate diet the patient’s condition improved. Follow-up magnetic resonance imaging (MRI) revealed complete resolution of the lesions. To our knowledge, this is the first report concerning a completely reversible PLS in AIP.

Keywords Acute intermittent porphyria · Stroke · Reversible posterior leukoencephalopathy syndrome · Nitric oxide · MRI

Introduction

Acute intermittent porphyria (AIP) is an autosomal dominant disorder of haem biosynthesis that can affect the autonomic, peripheral, and central nervous system (CNS). Onset is usually not before puberty, from which time the clinical course is variable. The most frequent neurological clinical manifestations are peripheral neuropathy, seizures and mental disturbance [1].

Magnetic resonance imaging (MRI) examination during an acute attack with CNS involvement reveals predominantly cortical and subcortical lesions which can show mild contrast enhancement [2, 3]. We wish to present a case of AIP in which MRI revealed a lesion pattern as found in posterior leukoencephalopathy syndrome (PLS).

Case report

A 30-year-old woman with no family history of porphyria was admitted following development of diffuse abdominal pain. Appendectomy was performed 3 days later. Histological examination showed changes in keeping with chronic recurrent appendicitis. On the 10th day after admission she suffered three generalized seizures and was transferred to the neurological department.

The vital signs at time of transfer consisted of a blood pressure of 180/100 mmHg, a heart rate of 120, a respiratory rate of 16 and a temperature of 38.5 °C. The general physical examination was normal. The patient was orientated in person but not in time and place and was capable of following easy commands. There was no focal neurological deficit and no evidence of papilloedema or meningism. The cerebrospinal fluid was normal. Except for hyponatraemia of 130 mEq/l, the patient’s blood tests were unremarkable. Transcranial Doppler sonography revealed increased flow rates in all vessels, but no vasospasm. MRI of the brain, performed at the day of transfer, showed bilateral cortical and subcortical lesions in T2-weighted sequences. These signal alterations were predominantly located in the occipital cortex and subcortical white matter.
and in Rolando’s area and were detected best by fluid-attenuated inversion recovery (FLAIR)-sequences (Fig. 1). There was no breakdown of blood-brain-barrier in T1-weighted sequences after application of gadolinium-DTPA. Moreover it demonstrated a moderate hydrocephalus of both lateral and third ventricle without signs of acute transpendymal diapedesis of cerebrospinal fluid.

On the 11th day the patient suffered from two further seizures as well as a deterioration in mental state. At this time, an increased urinary excretion of porphyrins was discovered, and the diagnosis of AIP was made, which was subsequently confirmed by further blood, urine, and faecal analysis. Treatment with haematin, high carbohydrate diet and supportive therapy was given. On the 12th day the patient was somnolent, and again suffered several seizures and required intubation and ventilation.

A follow-up MRI performed on the 18th day showed a mild regression of the lesions. The final MRI on the 26th day proved complete regression of the lesions (Fig. 2). At this time transcranial Doppler sonography indicated normal flow rates. There were no abnormal neurological findings on the day of discharge, following 34 days of treatment in the intensive care unit.