

Reversible posterior encephalopathy and cerebral vasoconstriction in acute intermittent porphyria

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Section: Neuroradiology

Area of Interest: Neuroradiology brain

Procedure: Comparative studies

Imaging Technique: CT

Imaging Technique: MR

Imaging Technique: MR-Angiography

Special Focus: Acute Oedema Metabolic disorders

Pathology Case Type: Clinical Cases

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Patient: 31 years, female

Clinical History:

The patient presented with severe abdominal pain and hypertension. Laboratory findings revealed mild hyponatraemia and hyperchromia. Abdominal US, CT, colonoscopy, gastroscopy and gynaecological examination were unremarkable. On day four she had two subsequent epileptic seizures with respiratory arrest and successful resuscitation and was transferred to the intensive care unit.

Imaging Findings:

Head CT performed after resuscitation showed a single nonspecific hypodensity in the subcortical white matter of the left frontal lobe. (Fig. 1) The next day brain MRI showed bilateral symmetric FLAIR/T2 hyperintensities located cortically and subcortically in the posterior parietal lobes. Additionally, few and asymmetric white matter lesions of subcortical and deep white matter in the frontal and parietal lobes were noted in the frontoparietal region bilaterally. (Fig. 2) DWI showed increased diffusion and there was no enhancement after gadolinium contrast media administration. TOF MRA showed segmental narrowing of intracranial arteries, which was more pronounced in the posterior circulation. (Fig. 3) DWI showed diffusion and there was no enhancement after gadolinium contrast media administration.

Subsequent brain MRI after 2 weeks showed disappearance of cortical and subcortical lesions in posterior parietal lobes. (Fig. 4) TOF MRA showed marked reversal of segmental narrowing in cerebral arteries. (Fig. 5)

Discussion:

Subsequent tests revealed elevated levels of porphobilinogen and d-ALA in urinary samples. EMG revealed severe motor axonal neuropathy in proximal upper limbs. Based on clinical data, laboratory and imaging findings, the patient was diagnosed with acute intermittent porphyria and treated with human hemin.

Acute porphyrias are rare inherited disorders due to deficiencies of haem synthesis enzymes. [1] Inheritance is usually autosomal dominant, although penetrance is low and most gene carriers remain asymptomatic. Clinical presentation is typically with acute neurovisceral attacks characterised by severe abdominal pain, vomiting, tachycardia and hypertension. Severe attacks may be complicated by hyponatraemia, peripheral neuropathy

sometimes causing paralysis, seizures and psychiatric features. Attacks are triggered by drugs, alcohol, hormonal changes, fasting or stress. The diagnosis is based on increased porphobilinogen excretion in a urine sample. [2] Prognosis is good if the condition is recognised early and treated aggressively. [3]

Authors have reported acute porphyria in association with reversible cerebral vasoconstriction [6], posterior reversible encephalopathy syndrome [7], multiple reversible cerebral lesions [8], reversible diffuse gyriform cortical enhancement [9] and bilateral occipital lobe lesions [10], where either vascular or hypertensive aetiology were suggested as the causative factor.

Reversible cerebral vasoconstriction syndrome (RCVS) and posterior reversible encephalopathy syndrome (PRES) overlap significantly in their clinical and radiographic features, and the two entities are frequently encountered as complications of various medical conditions, including intravenous immunoglobulin therapy, Guillain-Barre syndrome, immunosuppression, stem cell transplantation, blood transfusions, and septic shock. [4]

A large series of patients with clinical and radiographic features of PRES identified the presence of RCVS in over 24% of patients. [5] Moreover, a multifocal cerebral vasoconstriction has been noted in more than 85% of patients with PRES whenever investigations included angiography. Inversely, reversible brain oedema occurs in 8–38% of all cases of RCVS. [11]

Given the significant overlap between the two entities, RCVS and PRES may represent a spectrum of potential clinical manifestations of a common underlying pathophysiology involving various degrees of altered cerebral vascular tone and endothelial dysfunction. [4]

Presented imaging findings included elements of vasoconstriction as well as PRES-like lesions in cortico-subcortical distribution. Both were reversible as showed on control studies. Based on imaging findings, we concluded that the patient most likely suffered from a spectrum of reversible cerebral vasoconstriction and posterior reversible encephalopathy, similar to RCVS and PRES, respectively.

Differential Diagnosis List: Reversible posterior encephalopathy and cerebral vasoconstriction in acute intermittent porphyria, Vasculitis, Ischaemic lesions

Final Diagnosis: Reversible posterior encephalopathy and cerebral vasoconstriction in acute intermittent porphyria

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600 (PMID: [7677301](#))

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Figure 1

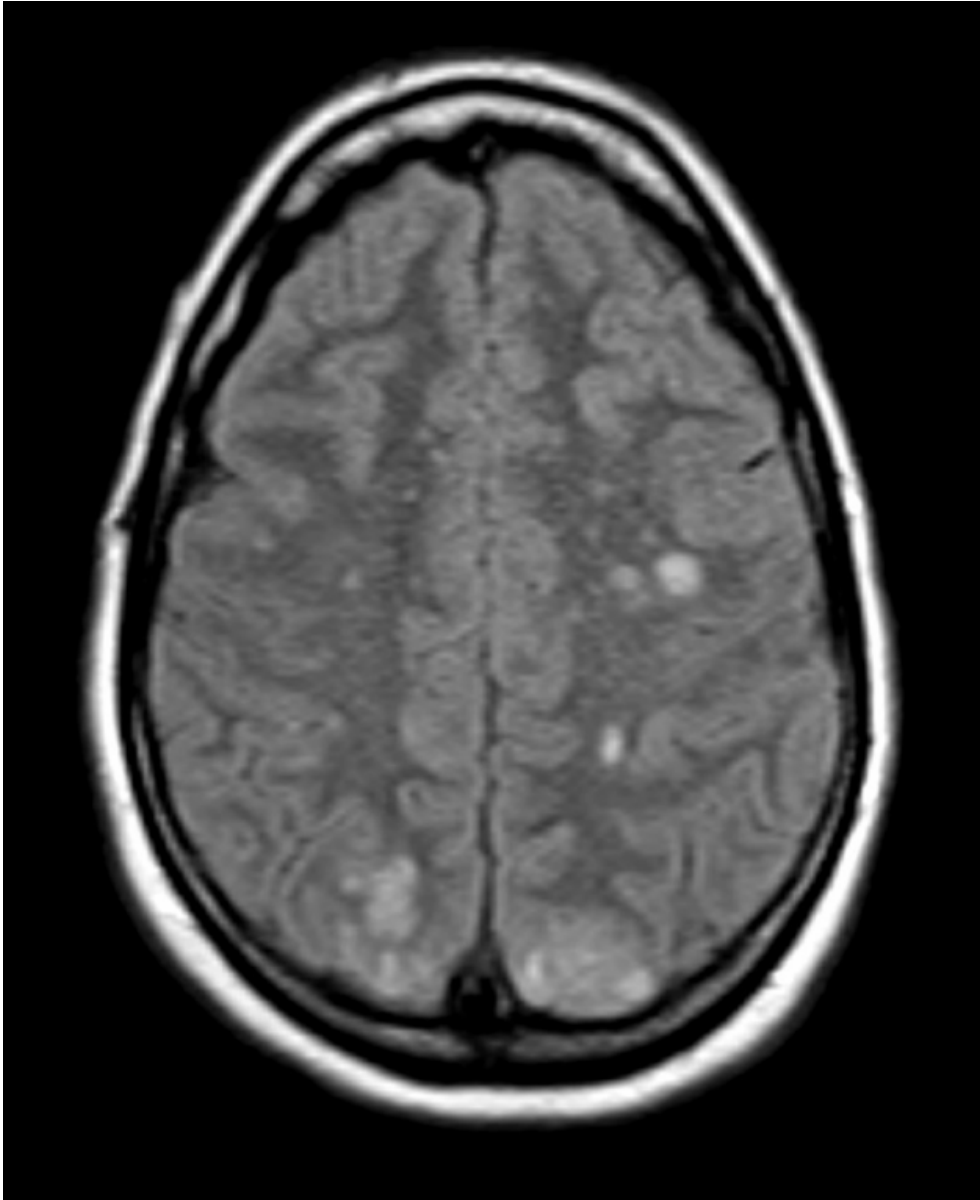
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Description: Head CT showed an small subcortical hypodensity in the left frontal lobe. **Origin:** Department of Radiology, University Medical Centre Ljubljana, Slovenia

Figure 2

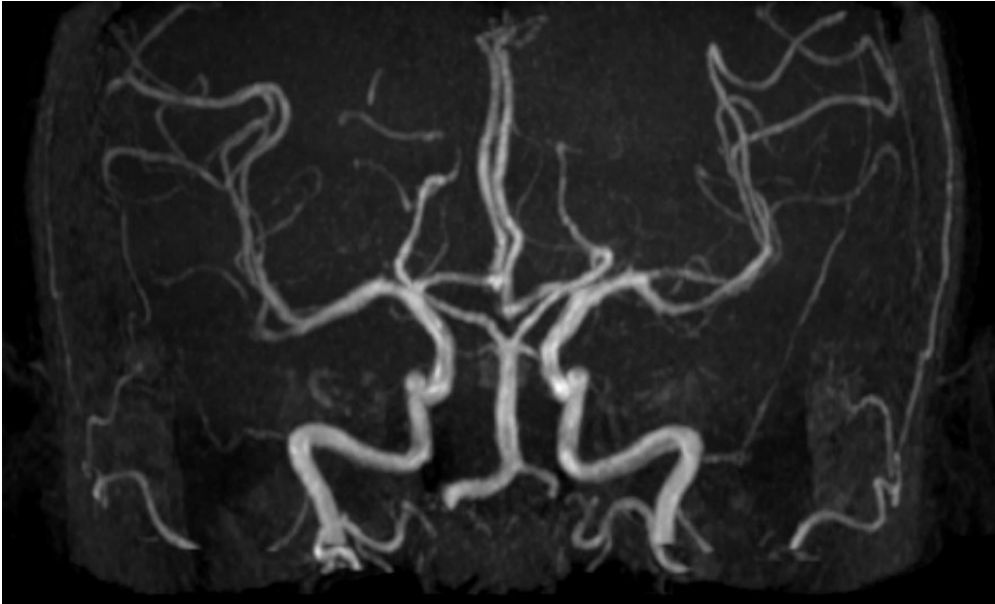
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Description: Relatively symmetric signal changes, located cortically and subcortically in both posterior parietal lobes with mild effacement of adjacent sulci were seen on FLAIR images. **Origin:** Department of Radiology, University Medical Centre Ljubljana, Slovenia

Figure 3

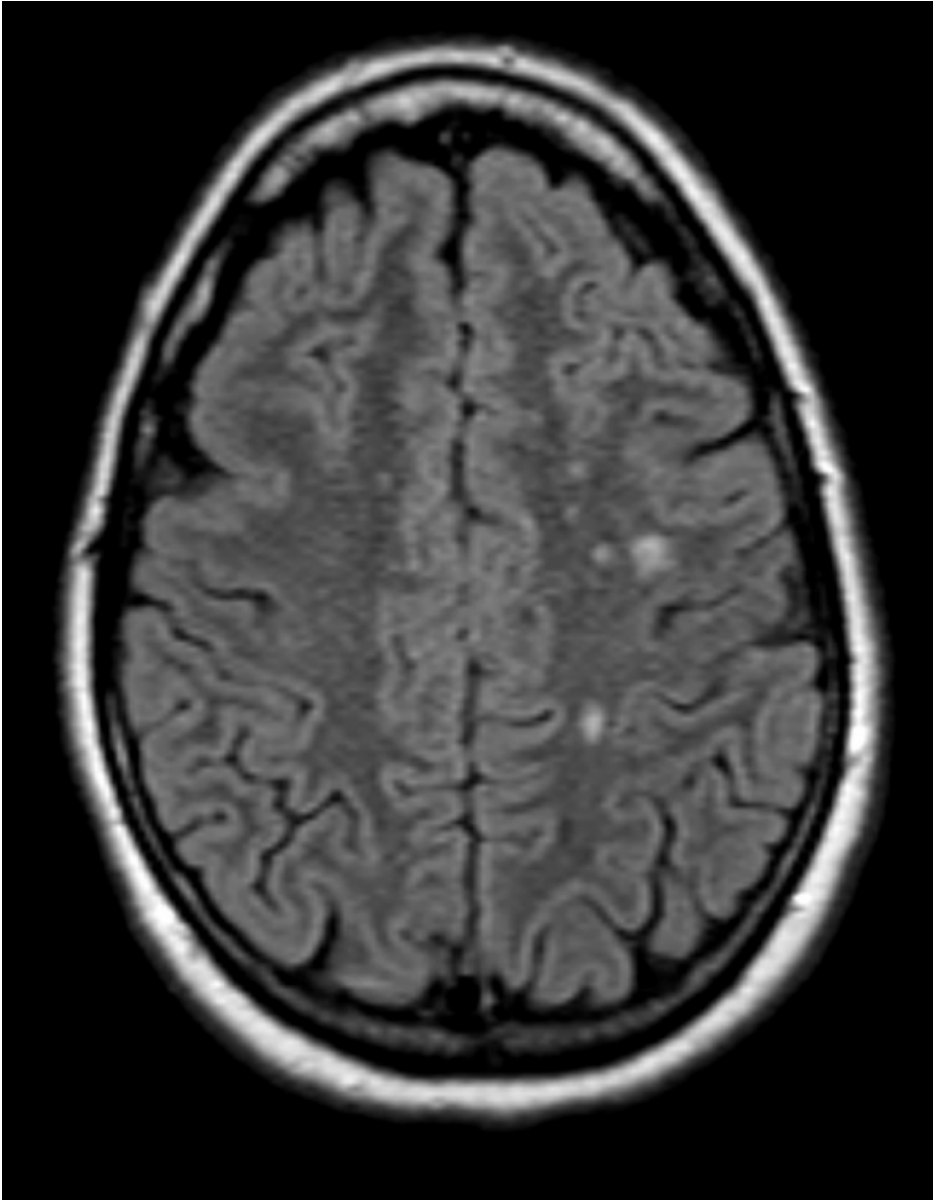
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Description: TOF MRA images showed segmental narrowing of intracranial arteries, which was most pronounced in the posterior circulation. **Origin:** Department of Radiology, University Medical Centre Ljubljana, Slovenia

Figure 4

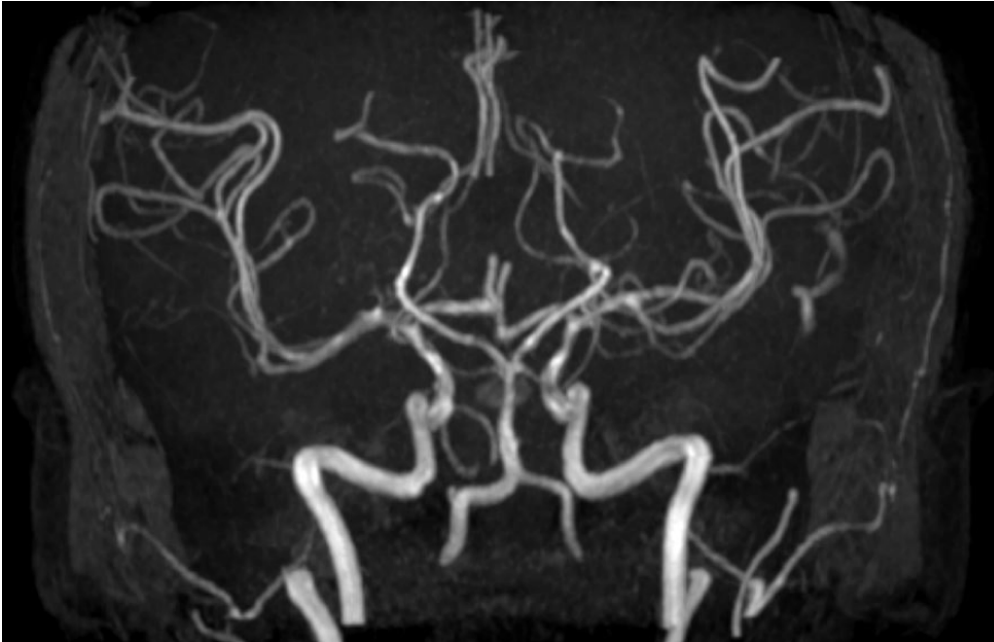
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Description: MRI after 2 weeks showed disappearance of signal changes in posterior parietal lobes on FLAIR images. Sulci appeared normal. **Origin:** Department of Radiology, University Medical Centre Ljubljana, Slovenia

Figure 5

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Description: TOF MRA images after 2 weeks showed marked improvement of segmental narrowing of cerebral arteries. **Origin:** Department of Radiology, University Medical Centre Ljubljana, Slovenia