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CASE REPORT

Loss of consciousness, encephalopathy and hereditary haemorrhagic telangiectasia

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Introduction

Hereditary haemorrhagic telangiectasia is a rare genetic condition that leads to vascular malformations in virtually every organ. This article presents the case of a 78-year-old woman admitted to hospital on two occasions with loss of consciousness, requiring intubation and ventilation. Ultrasound and computed tomography imaging identified hepatic vascular malformations within the liver, causing portosystemic shunting of blood and leading to encephalopathy and subsequent coma. The use of osmotic laxatives and the antibiotic rifaximin helped prevent further encephalopathy. The case highlights the importance of recognising rarer causes of a confused and drowsy older patient.

Case report

A 78-year-old woman presented to hospital after being found unconscious at home. She had been confused over the preceding week and had been treated with antibiotics for a urinary tract infection by her GP. Background history included hereditary haemorrhagic telangiectasia, atrial fibrillation and gastrointestinal lymphoma.

Her Glasgow Coma Scale score on arrival was 5 (E2V2M1). Neurological examination identified nystagmus on left lateral gaze and increased tone in both legs. Computed tomography brain and magnetic resonance angiography excluded an acute intracerebral event and lumbar puncture did not identify an infective cause. The patient was intubated and taken to intensive care. An electroencephalogram suggested severe encephalopathy but no seizure activity.

Of note, the patient had presented similarly 2 months previously to another hospital with reduced consciousness requiring intubation. At the time, this was attributed to opiate toxicity from a buprenorphine patch.

Her serum ammonia level was elevated at 90 umol/litre (12–32 umol/litre). The patient was started on regular lactulose and her Glasgow Coma Scale score began to improve and she was extubated. The patient was subsequently transferred to a geriatric medicine ward. The '4AT tool' was used to help identify delirium. A score of 6 suggested possible delirium with or without cognitive impairment (Tieges et al, 2021).

Serum ammonia levels were consistently elevated and ultrasound of the abdomen identified an enlarged liver with numerous prominent intrahepatic vessels. The patient remained confused with fluctuating cognition and occasional agitation. Initially she was managed as having severe hypoactive delirium, but given the background hereditary haemorrhagic telangiectasia, elevated serum ammonia levels and numerous intrahepatic vessels on ultrasound imaging, it was hypothesised that vascular malformations within the liver may be causing a portosystemic shunt, elevated ammonia levels, encephalopathy and coma.

Previous liver computed tomography imaging from 1 year earlier performed at a different hospital was reviewed in view of the presentation. This showed abnormally large calibre common hepatic artery, multiple vascular collaterals in the porta hepatis, along with tortuous intrahepatic vessels. The patient continued on regular lactulose and rifaximin was started to treat or prevent further episodes of encephalopathy. A multiphase computed tomography scan of the liver was performed, which confirmed hepatic vascular malformations resulting in arteriovenous and portosystemic shunts (Figure 1). Six months after admission the patient had avoided further admissions with lactulose and rifaximin preventing encephalopathy.

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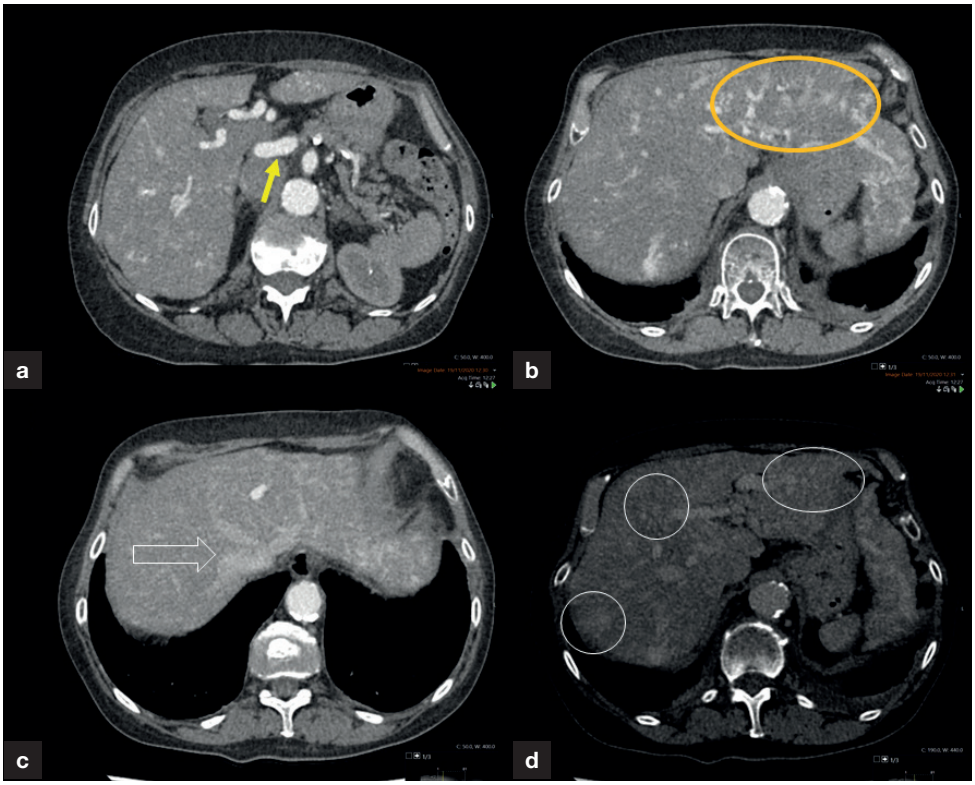

Figure 1. Axial computed tomography images. a. Arterial phase image showed a large common hepatic artery and its main branches (yellow arrow), also note the absence of contrast in the portal vein which lies between the artery branches. b. Arterial phase image showing numerous tortuous arterial branch vessels in the left lobe of liver (orange circle). c. Arterial phase image showing contrast in hepatic veins (white arrowhead) despite no contrast in portal vein, confirming arterio-venous shunting. d. Portal venous phase study shows multiple foci of persistent enhancement in the liver parenchyma (white circles), suggestive of venous malformations in the clinical context.

Discussion

Hereditary haemorrhagic telangiectasia is a rare genetic disorder characterised by widespread cutaneous, mucosal and visceral vascular malformations.

Hepatic vascular malformations are common, affecting 41–74% of patients with hereditary haemorrhagic telangiectasia. These malformations can cause the shunting of blood between the portal, systemic and arterial circulation (Buscarini et al, 2011). Symptoms are rare and generally do not occur before the age of 50 years. Hepatic lesions are more common in older patients (Lesca et al, 2007). High output cardiac failure, portal hypertension and biliary ischaemia are the more common complications of liver disease in hereditary haemorrhagic telangiectasia; portosystemic encephalopathy is rare (Guadalupe, 2007).

Diagnosis of liver involvement in patients with hereditary haemorrhagic telangiectasia is based on the identification of diffuse liver telangiectasias, an enlarged common hepatic artery and intrahepatic hypervascularisation on imaging. Angiography is the gold standard, but doppler ultrasound is commonly used (Faughnan et al, 2011). Portosystemic encephalopathy has been treated successfully with the use of osmotic laxatives and rifaximin. Hepatic artery branch embolisation or ligation has been performed in an attempt at shunt reduction. Reduction in symptoms is often transient and there is significant risk of hepatic or biliary necrosis. Liver transplantation is the only curative treatment for patients with hereditary haemorrhagic telangiectasia and liver involvement (Lesca et al, 2007).

This case highlights a patient who presented to hospital on two occasions with encephalopathy needing intubation. The cause of her altered level of consciousness was initially unknown and considered a severe hypoactive delirium. Recognising hereditary haemorrhagic telangiectasia and the associated liver complications allowed her condition to be managed.

Learning points

- Hereditary haemorrhagic telangiectasia is a rare genetic condition which causes vascular malformations in the skin, mucosal surfaces and visceral organs.
- Hepatic malformations can cause the shunting of blood between the portal, systemic and arterial circulation.
- Portosystemic encephalopathy is a rare complication of liver involvement in hereditary haemorrhagic telangiectasia.
- Portosystemic encephalopathy has been successfully treated with the use of osmotic laxatives and rifaximin.
- In older patients with persistent fluctuating cognition rare causes should be considered and a patient's medical history acknowledged.

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