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EPISODIC ENCEPHALOPATHY AS A MASQUERADE: A RARE ASSOCIATION WITH PORTAL VEIN THROMBOSIS AND HEREDITARY THROMBOPHILIA

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ABSTRACT Encephalopathy is a syndrome of brain dysfunction caused by reversible or irreversible factors.Reversible causes of encephalopathy include hepatic encephalopathy,hashimoto's encephalopathy, metabolic encephalopathy, infections,braintumours,toxicexposure, non convulsive status epilepticus. We report a 51-year-old woman with recurrent episodes of hepatic encephalopathy secondary to portal vein thrombosis (PVT) caused by hereditary thrombophilia. This case highlights a rare cause of recurrent encephalopathy and emphasizes the importance of evaluating thrombotic disorders in such patients.

KEYWORDS :

INTRODUCTION

Encephalopathy refers to brain dysfunction caused by multiple etiologies, which may be reversible or irreversible. Reversible causes include hypoxic-ischemic encephalopathy, hepatic encephalopathy, metabolic encephalopathy, toxic encephalopathy, uremic encephalopathy, infection-related encephalopathy, and brain tumors.

Recurrent encephalopathy may result from metabolic disorders, substance abuse, demyelinating diseases, or hepatic encephalopathy. HE accounts for the majority of encephalopathy cases in patients with cirrhosis, affecting approximately 30–45% of such individuals[1].

HE is broadly defined as brain dysfunction resulting from liver insufficiency and/or portosystemic shunting. It manifests as a spectrum of neurological and psychiatric abnormalities, ranging from subtle cognitive impairment to coma[1]. Portal vein thrombosis (PVT) is a rare cause of HE [2,3]. The incidence of HE in patients with PVT is relatively high, particularly in those with advanced cirrhosis, with previous studies reporting 10–30% of patients affected[4-7].

PVT refers to the formation of a thrombus in the trunk of the portal vein or its intrahepatic branches. It can occur in patients with or without cirrhosis, and various conditions predispose individuals to PVT, including hereditary thrombophilia[8-11].

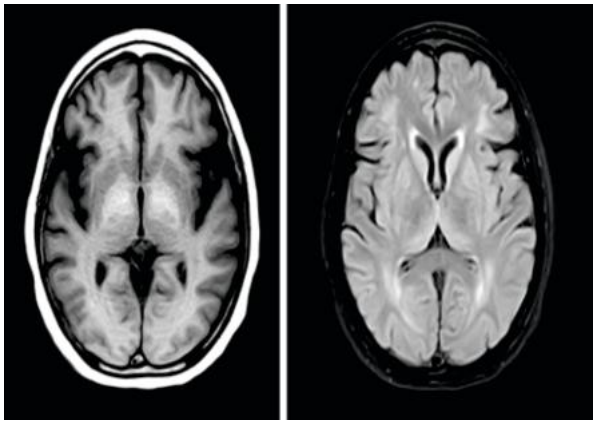
Protein C deficiency occurs in approximately 1 in 200–500 individuals, protein S deficiency in 1 in 500, and antithrombin III deficiency in 1 in 2000–5000 individuals[8,10].

Case Report

A 51-year-old woman presented with seizures followed by altered mental status, with a history of recurrent similar episodes over the past 2 years (2–3 episodes per year, each lasting about one week). She had no fever, headache, or history of drug use and was not known to have diabetes, hypertension, cerebrovascular disease, coronary artery disease, chronic liver disease, or chronic kidney disease.

On presentation, her Glasgow Coma Scale (GCS) was 3/15. Pupils were bilaterally normal and reactive; plantar responses were mute; no neck stiffness was noted. Laboratory investigations revealed persistent hypokalemia and mild elevations in liver enzymes (SGOT and SGPT). Thyroid profile and blood glucose were normal. Autoimmune encephalitis panel and cerebrospinal fluid analysis were unremarkable.

Serum ammonia was elevated at 90 mcg/dL (normal: 7–40 mcg/dL). Ultrasonography of the abdomen showed coarse liver echotexture, portal vein attenuation, and collaterals suggestive of chronic thrombosis. Contrast-enhanced CT confirmed severe attenuation of the portal vein with periportal and peripancreatic collaterals, consistent with chronic thrombus.



MRI Brain Findings:

MRI brain revealed symmetrical T1-weighted hyperintensities involving the bilateral basal ganglia and corona radiata. These findings are suggestive of hepatic encephalopathy changes secondary to hyperammonemia.

Thrombotic workup revealed antithrombin III 65% (normal 83–128%), protein C 32% (normal 70–150%), and lupus anticoagulant 55.9 (normal 28–30), with normal anticardiolipin and APA antibodies, confirming hereditary thrombophilia as the underlying cause of PVT.

DISCUSSION

The term encephalopathy originates from the Greek words enképhalos (brain) and páthos (suffering) and refers to any disorder of the brain leading to dysfunction. It is a syndrome of global brain dysfunction, with altered cognition, attention, orientation, sleep–wake cycle, or consciousness.

Encephalopathy may be reversible or irreversible. Reversible causes

include:

- **Hepatic Encephalopathy:** due to accumulation of toxins from liver dysfunction.
- **Hashimoto's Encephalopathy:** associated with autoimmune thyroid disease.
- **Metabolic Encephalopathy:** secondary to diabetes, kidney failure, or heart failure.
- **Infections**: including encephalitis, meningitis, or sepsis.
- **Brain Tumors**
- **Toxic Exposure:** long-term exposure to drugs, solvents, or metals
- Nonconvulsive status epilepticus

HE is a serious but potentially reversible condition in patients with advanced liver disease. It may result from underlying liver dysfunction or portosystemic shunts, such as TIPS. Common triggers include renal failure, gastrointestinal bleeding, constipation, infections, medication noncompliance, excessive protein intake, dehydration, electrolyte imbalances, alcohol use, and sedative medications.

PVT involves one or more components of Virchow's triad: reduced portal blood flow, hypercoagulability, or vascular endothelial injury[7]. Inherited thrombophilia (e.g., Factor V Leiden, prothrombin G20210A mutation, protein C/S deficiency, antithrombin deficiency, elevated factor VIII) significantly increases the risk of PVT.

Protein C deficiency is rare, occurring in 1 in 200–500 individuals, whereas clinically significant deficiency occurs in 1 in 20,000 people. Antithrombin III deficiency and protein S deficiency are even less common.

This case emphasizes the importance of considering hereditary thrombophilia in recurrent HE without overt liver failure, as early diagnosis allows targeted therapy and prevention of future thrombotic events

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