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Intraparenchymal hemorrhage and cerebral venous thrombosis in an adult with congenital porencephalic cyst presenting for generalized tonic-clonic seizures

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ABSTRACT

Prothrombotic conditions are known risk factors for porencephalic cyst formation and cerebral vein thrombosis. Intracerebral hemorrhage is a potential complication of a cerebral vein thrombosis. Porencephaly is a risk factor for intracerebral hemorrhage and cerebral vein thrombosis formation. We present the case of an adult patient with a past medical history of epilepsy and congenital porencephalic cyst with de novo mutation of the COL4A1 gene who presented for episodes of generalized tonic-clonic seizure after a substantial symptom-free period. A brain CT scan showed an intracerebral hemorrhage with porencephalic cyst and superior sagittal sinus thrombosis despite negative thrombophilia work-up. A CT perfusion study, CT angiography, and brain MRI confirmed the diagnosis. The cause-and-effect relationship between porencephalic cysts, cerebral venous thrombosis, and intracerebral hemorrhage is still not clear in the literature.

Introduction

Porencephalic cyst is a rare condition of cerebrospinal fluid accumulation within the brain parenchyma, with incidence of 3.5 per 100,000 live births [1]. It may be classified as sporadic or familial related to de novo or inherited mutations of COL4A1 gene. Type IV a1 collagen chain, encoded by COL4A1 gene, is essential for the structural integrity of the vascular basement membrane [2]. In addition to their contribution to

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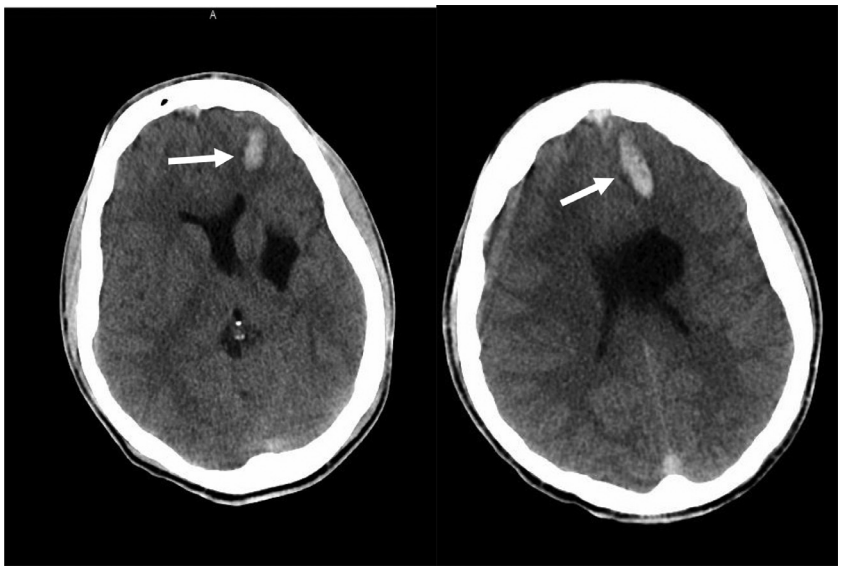

Fig. 1 – (A) and (B) (Right to left): Axial CT scans brain without contrast showing a left frontal lobe intraparenchymal hemorrhage (A) and another left frontal lobe intraparenchymal hemorrhage located more inferiorly (B).

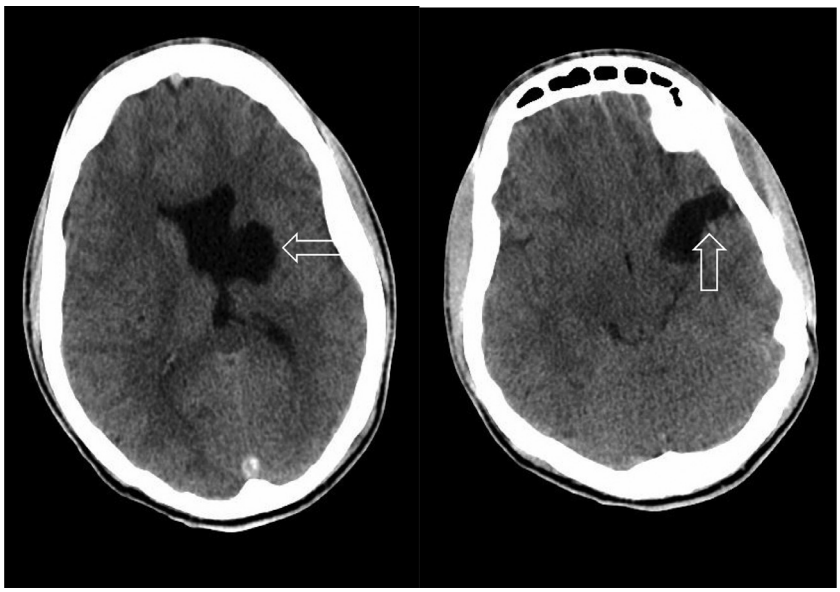

Fig. 2 – (A) and (B): Axial CT scan brain without contrast showing a porencephalic cyst in the left sylvian fissure (A) and inferior basal ganglia (B).

the formation of porencephalic cysts, COL4A1 mutations have been largely confined to the neonatal and adult-onset recurrent hemorrhagic and ischemic strokes [3].

We describe the case of a 20-year-old patient with a past medical history of congenital porencephalic cyst who presented for episodes of generalized tonic-clonic seizure. Brain CT scan, MRI, and CT angiography confirmed the presence of intracerebral hemorrhage along with sinus thrombosis despite a normal thrombophilia work up.

Case report

A 20-year-old man with a past medical history of epilepsy, congenital right hemiparesis, and congenital porencephalic cyst presented to the emergency department with witnessed generalized tonic-clonic seizures, three episodes at home and one in the emergency department. The patient was diagnosed with epilepsy and porencephalic cyst since birth associated

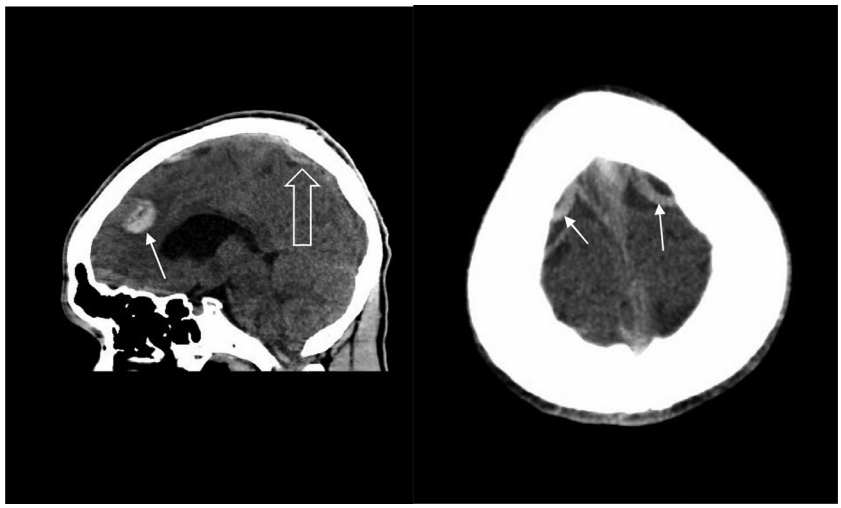

Fig. 3 – (A) and (B): Sagittal CT scan brain without contrast showing a left frontal lobe intraparenchymal hemorrhage (Arrowhead) as well as a hyperdensity within the superior sagittal sinus suggestive of a Superior sagittal vein thrombosis (Arrow). Axial CT scan brain without contrast showing multiple hyperdensities within the cortical veins suggestive of acute venous thrombosis (Arrows).

with congenital right hemiparesis. He tested positive for a heterozygous mutation of the COL4A1 gene although his parents tested negative. He was on anti-seizure medications up until when he turned 10 years old, and had a seizure free period for 10 years as per family.

On physical exam, the patient was drowsy, and had altered mental status. There was no evidence of muscle weakness. He was given intravenously (IV) 10 mg of Midazolam to go into postictal phase with slow return to baseline later on.

A brain CT scan without IV contrast revealed two parasagittal hyperdense lesions within the white matter of the left frontal lobe (Fig. 1A and B). These lesions were consistent with intracerebral hemorrhage. Within the same scan, a porencephalic cyst in the left sylvian fissure and inferior basal ganglia was present (Fig. 2A and B).

A hyperdensity within the superior sagittal sinus as well as within multiple cortical veins was present suggestive of acute venous thrombosis (Fig. 3A and B).

A brain MRI showed an acute/subacute gyriform infarct in the high left parietal lobe, consistent with an acute/subacute left MCA territory ischemia. The infarct was hyperintense on FLAIR, T2-weighted, diffusion weighted images, and hypointense on ADC and T1-weighted images (Fig. 4A-C).

A CT perfusion study showed a matched region of increased mean transit time, decreased blood flow, and decreased blood volume in the left occipital and parietal lobes, consistent with a completed infarct likely due to a superior sagittal sinus as well as a transverse sinus thrombosis.

A CT angiography of the head revealed intact intracerebral arteries, with no evidence of intra-arterial thrombosis. However, there was evidence of a left transverse sinus thrombosis (Fig. 5).

The intraparenchymal bleed was stable for 3 days. The thrombophilic work up including serum levels of factor V, prothrombin, protein S and protein C, and antithormbin III was negative Rivaroxaban was started three weeks after the patient was admitted. The patient had no recurrent seizures in a follow-up for 2 months.

Discussion

A porencephalic cyst is a congenital or less commonly acquired cavity within the cerebral hemisphere which contains cerebrospinal fluid [4]. Congenital cysts can result from intrauterine vascular injury or infections [5]. Acquired cysts are secondary to injury because of a trauma, surgery, ischemia, or infection [4].

Porencephaly might rarely present with seizures which may be partial or generalized [4]. Qureshi and colleagues reported a patient with a history of perinatal intracerebral hemorrhage who presented for a witnessed new-onset generalized tonic-clonic seizure. Brain imaging showed a large frontal lobe porencephalic cyst, with encephalomalacia in the right parietal lobe and temporal lobe [6]. Another patient reported by Oommen and colleagues presented with postictal confusion following a new onset of focal seizures with secondary generalization. Brain imaging exhibited a porencephalic cyst in the left posterior parietal lobe [4].

Intracerebral hemorrhage can be either a cause or a complication of a cyst [7]. Porencephaly by itself is a prothrombotic state. Nicolini and colleagues reported that fetuses born with porencephaly had a higher hematocrit level than other normal fetuses [8]. It has been noted that local causes including porencephaly can determine dural sinus and cerebral sinus occlusions [9]. However, the occurrence of both hemorrhage and thrombosis in the same patient with porencephaly has not been clearly reported in the literature so far. The de novo mutation of the COL4A1 mutation in our patient could have contributed to the co-occurrence of both states.

The diagnosis of porencephalic cyst associated with intraparenchymal hemorrhage and sinus thrombosis in a patient

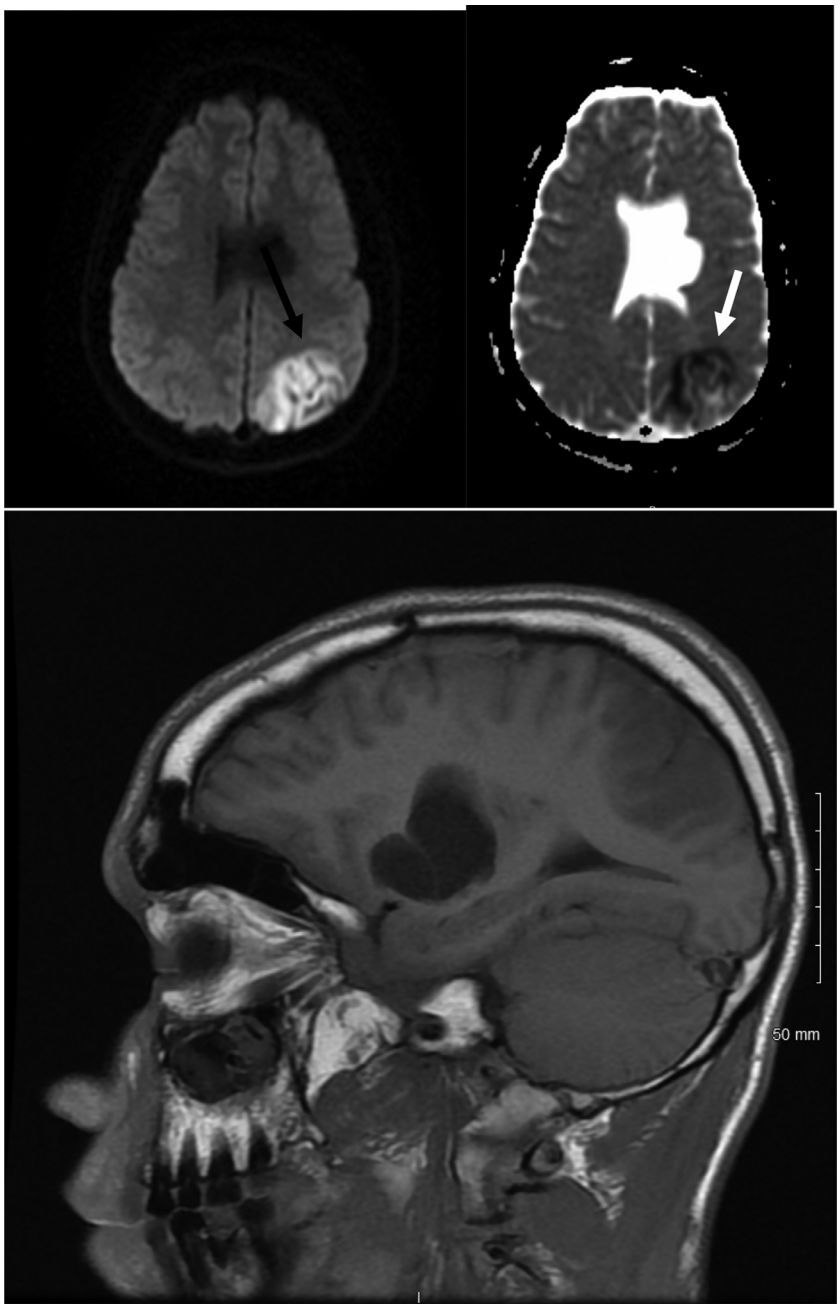

Fig. 4 – (A), (B), and (C): Brain MRI shows a hyperintense lesion (black arrow) on FLAIR, T2-weighted, diffusion weighted images at the level of the left parietal lobe that is hypointense on T1-weighted images (white arrow) consistent with an acute left parietal ischemia. The sagittal T1-weighted image shows hypointense lesion at the level of the right parietal lobe consistent with an ischemic area (C).

presenting for new onset seizures necessitates brain imaging. EEG might show a normal background, or slowing which may be focal or generalized [6]. CT without contrast is the most commonly used initial study to diagnose an intraparenchymal hemorrhage because of its wide availability and high sensitivity and specificity [10]. MRI without contrast has nearly similar sensitivity and specificity [10]. The diagnosis of cerebral sinus thrombosis is based on neuroimaging. A CT scan should be included in the diagnostic strategy for ruling out other acute cerebral disorders that sinus thrombosis can imitate including arterial stroke, abscess, tumors and subarachnoid hemorrhage [9]. CT venography is especially useful in the acute setting because it provides a rapid and reliable method for detection of sinus thrombosis, particularly in patients with contraindications to MRI. This modality can be performed immediately after brain CT scan [9]. The combination of MRI showing the thrombosed vessel and MR venography demonstrating no visualization of the same vessel is currently the

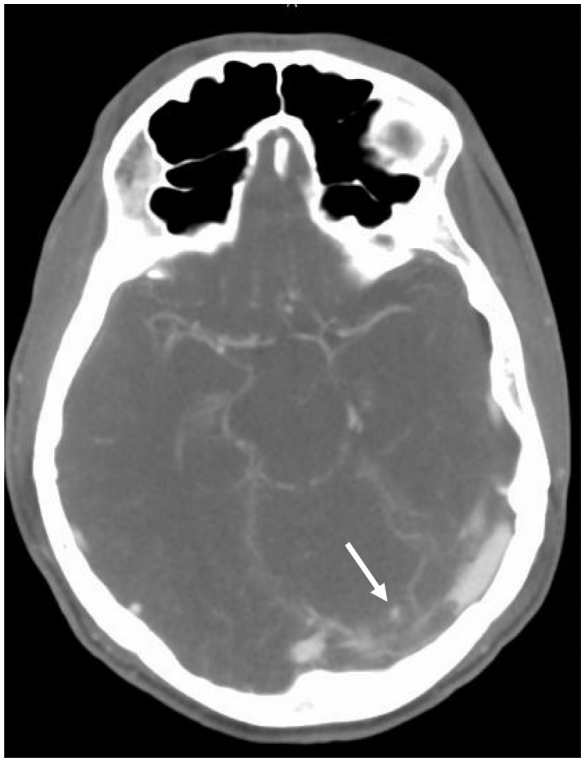

Fig. 5 – Axial CT angiography of the head with IV contrast reveals a patent circle of Willis with no evidence of intra-arterial thrombosis. However, a filling defect is noted at the level of the left transverse sinus (white arrow).

most sensitive method to confirm the diagnosis of sinus thrombosis in the acute, subacute, and chronic phases [11].

As performed in our case, an initial CT scan without IV contrast showed two areas of intraparenchymal hemorrhage and a porencephalic cyst with a suspicion for acute venous thrombosis which was confirmed by a CT angiography and brain MRI.

Treatment for porencephaly and its possible complications includes physical therapy for neurological deficits, anti-epileptic drugs for seizure control, and a shunt in case of hydrocephalus. Surgical choices are recommended for cases resistant to anti-epileptic drugs. Those choices include hemispherectomy and hemispherotomy [12]. Identifying the cause of intracerebral hemorrhage and venous thrombosis is vital. In our case, the patient had a negative thrombophilia work-up, and had no risk factors for developing both conditions simultaneously except for the presence of a congenital porencephalic cyst with positive mutation of the COL4A1 gene.

Conclusion

It is important for clinicians to consider a congenital porencephalic cyst as a potential cause for thrombosis, hemorrhage, and new-onset seizure. The cause-and-effect relationship between all of those conditions is still unknown. Brain imaging by CT scan, CT angiography, and brain MRI should be performed to visualize the cyst and assess for complications.

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