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CASE REPORT

Congenital pial AVF along the falx cerebri with complete agenesis of the corpus callosum and bilateral parasagittal pachygyria-polymicrogyria secondary to chronic ischemia

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Abstract

Objective Pial arteriovenous fistula (AVF) is an extremely rare entity due to direct arterial connection with the venous plexus without an intervening capillary network. The objective of this article is to describe a unique case of congenital pial AVF along the interhemispheric falx with complete callosal agenesis and malformation of cortical development within the bilateral anterior cerebral artery territories. We also demonstrated the distinctive feature of temporal stability of the extensive intracranial abnormalities without active intervention. Less than 100 cases have been reported thus far, most of which involve the adult rather than pediatric age group. A comprehensive literature review of congenital pial AVF will also be included.

Case description A 5-year-old child presented with headache and complex partial seizures. Imaging of the brain revealed the presence of polymicrogyria-pachygyria in the parasagittal frontoparietal lobes with associated underlying white matter hypodensities. Complete agenesis of the corpus callosum was also seen. In addition, enlarged and tortuous vessels were noted along the interhemispheric falx with no appreciable nidus. Bilateral dilated and tortuous ACAs were seen supplying the network of abnormal vessels along the falx. The radiological findings were stable on a follow-up MRI 12 years later.

Conclusion Our reported case adds to current limited knowledge of this rare entity in the pediatric age group, which is traditionally treated aggressively and urgently. Our case demonstrated temporal stability of this lesion with no detrimental complications observed. This suggests that the outcome of pial AVFs with conservative treatment may not be as grim as previously thought.

Keywords Pial arteriovenous fistula · Vascular malformation · Malformation of cortical development · Agenesis of corpus callosum

Introduction

Pial arteriovenous fistula (AVF) is a high pressure gradient vascular malformation (VM) with direct connection between the arterial and venous systems without an intervening capillary network [1]. These are rare, comprising only 1.6% of intracranial VMs [2]. Herein, we report a unique case of congenital pial arteriovenous fistula (AVF) along the interhemispheric falx with complete callosal agenesis and malformation of cortical development (MCD) within the bilateral anterior cerebral artery (ACA) territories, presumably a result of chronic ischemia during intrauterine development. Our case is also distinctive as we were able to establish temporal stability of both the VM and brain parenchymal changes without active intervention over a period of 12 years.

Case report

A 5-year-old child who was well at birth presented to our institution with headache and complex partial seizures. On examination, there was no focal neurological deficit and developmental milestones were age-appropriate. Seizures were well controlled with single anti-epileptic medication.

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Computed tomography of the brain (Fig. 1), magnetic resonance imaging (MRI), and time-of-flight magnetic resonance angiography of the brain (Fig. 2) revealed the presence of polymicrogyria-pachygyria in the parasagittal frontoparietal lobes with associated underlying white matter signal abnormalities. Complete agenesis of the corpus callosum was also seen. In addition, enlarged and tortuous vessels were noted along the interhemispheric falx with no appreciable nidus. Bilateral dilated and tortuous ACAs were seen supplying the network of abnormal vessels along the falx. Catheter angiogram was suggested to delineate the extent and anatomy of the pial AVF, with view for surgical intervention. The family however opted for expectant management. A repeat MRI brain which was performed for the investigation of frontal headache 12 years after the initial presentation showed stable findings with persistently dilated ACA branches and cortical veins (Fig. 3).

Discussion

Pial AVF is a rare VM with direct connection between the arterial and venous systems without an intervening capillary network [1]. It is distinct from pial arteriovenous malformation because it does not have a nidus and distinct from dural AVF because it is fed by pial artery rather than dural artery [1]. The high pressure gradient due to direct arteriovenous communication renders this lesion vulnerable to rupture and life-threatening bleed [1]. According to the updated 2014 International Society for the Study of Vascular Anomalies (ISSVA) classification, pial AVF is a high-flow simple VM [3]. Early presentation before 2 years of age is usually from high output cardiac failure, symptoms of increased intracranial pressure, giant varices presenting as a palpable mass or macrocephaly. Patients after 2 years of age can present with intracranial hemorrhage, seizure, neurological deficit, or headache [4, 5].

The underlying pathophysiology of this entity is unclear, and various theories have been proposed. They are divided into the congenital and acquired subtypes. Acquired pial AVFs had been associated with trauma [1, 2] while congenital pial AVFs had been proposed to be related to increased angiogenesis since a significant proportion of the reported cases had underlying vascular syndromes such as Rendu-Osler-Weber disease, Klippel-Trenaunay-Weber syndrome [6–11], and RAS-1 mutation [12]. Cases without associated vascular syndromes such as our reported case are extremely rare. We performed a thorough literature review, the results of which are delineated in Table 1 [11, 13–23]. Thus far, there are 12 case reports and case series of congenital pial AVF, none of which was associated with complete agenesis of corpus callosum or MCD. Also, none of the reported patients demonstrated temporal stability of imaging findings without active intervention.

We postulate that the pial AVF was the first of the three abnormalities to develop in this patient and is the most likely antecedent cause of the MCD and corpus callosal agenesis. The presence of VM along the interhemispheric falx, deriving its blood supply mainly from the bilateral ACAs, resulted in chronic ischemia due to the "steal phenomenon." Ischemic insult is a known cause of MCD [24]. The polymicrogyria-pachygyria seen in our patient was confined within the bilateral ACA territories, further supporting our hypothesis of an ischemic insult. Gliosis/signal abnormalities seen within the subjacent white matter, again confined within the ACA territories, were also likely ischemic in etiology. Mechanical inhibition to the midline crossing of the commissural fibers is hypothesized to be the cause of the high incidence of corpus callosal dysgenesis in individuals with interhemispheric lipomas [25]. Hence, it is not surprising that the presence of abnormal dilated vessels in the interhemispheric region due to the presence of pial AVF in our reported case could interrupt with the normal intrauterine development of the corpus callosum. Intrauterine development of the corpus callosum commence as early as the eighth week of gestation and it assumes its final shape by 18 to 20 weeks of

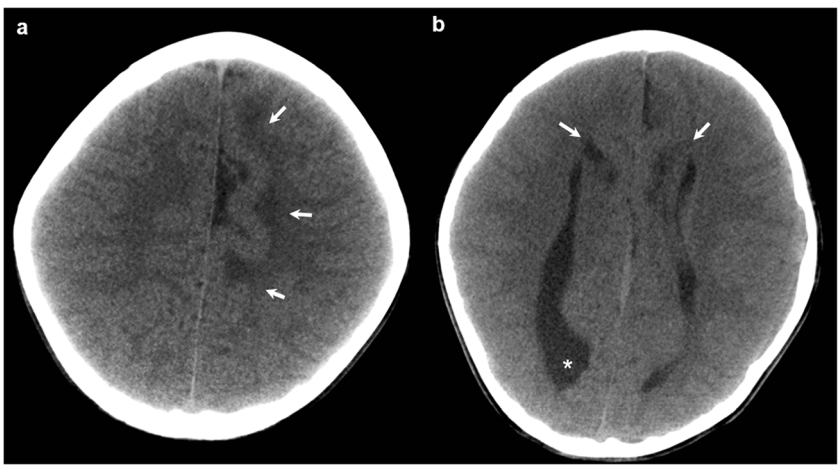

Fig. 1 **a**, **b** Axial CT images of the brain demonstrate the presence of abnormal gyral-sulcation pattern in the medial/parasagittal frontoparietal lobes with areas of low densities within the underlying white matter (arrows), as well as parallel appearance of the lateral ventricles and colpocephaly (*), in keeping with complete agenesis of the corpus callosum

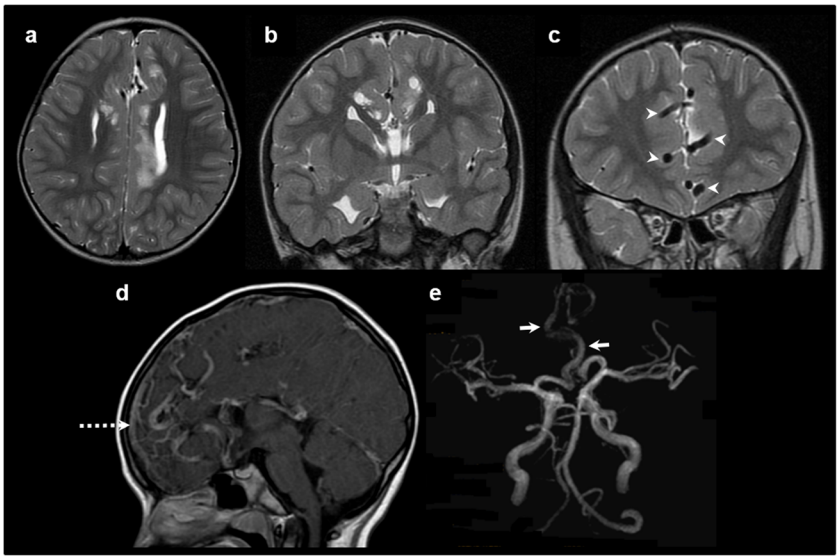

Fig. 2 **a-e**: **a** Axial and **b**, **c** coronal T2-weighted images of the brain confirmed the presence of polymicrogyria-pachygyria in the medial/parasagittal frontoparietal lobes, signal abnormalities within the underlying white matter, and complete agenesis of the corpus callosum. In addition, enlarged and tortuous vessels (arrowheads) are seen along the interhemispheric falx, with no appreciable nidus. **d** Midline sagittal post-contrast T1-weighted image showed enlarged and tortuous vessels along the interhemispheric falx. There is also mild prominence of the anterior superior sagittal sinus (dotted arrow). **e** Time of flight magnetic resonance angiogram of the circle of Willis revealed enlargement of the bilateral anterior cerebral arteries (solid arrows), supplying the network of abnormal vessels along the falx

gestation [26, 27]. The complete absence of the corpus callosum in our reported patient suggests that the pial AVF developed in the first trimester.

Owing to the rarity of pial AVF, its natural history is not known and a consensus regarding the best treatment strategy has not been established. Historically, pial AVF has been treated on urgent basis, either by endovascular or surgical approach [4, 28, 29]. Studies showed that patients who were treated surgically or endovascularly demonstrated good clinical outcomes [6, 7]. One study reported that five (63%) of eight patients managed conservatively expired due to acute or subsequent fatal bleeding [10]. Development of giant venous aneurysm had also been reported before, leading to cardiac failure [16]. Although extremely rare, spontaneous

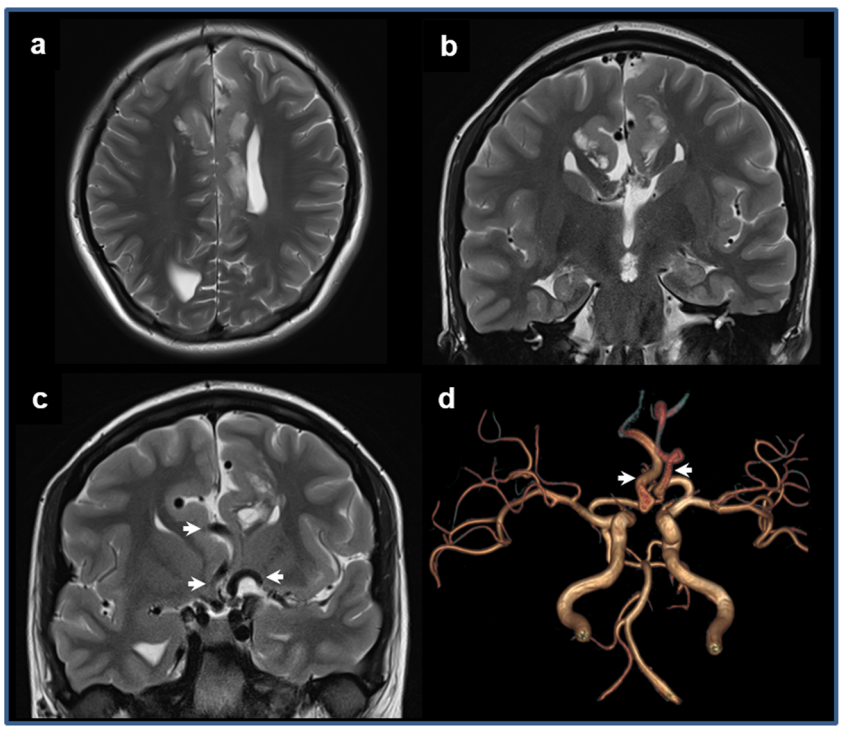

Fig. 3 **a-d** Follow-up magnetic resonance imaging 12 years after the initial presentation again showed polymicrogyria-pachygyria in the medial/parasagittal frontoparietal lobes, stable signal abnormalities within the underlying white matter, complete agenesis of the corpus callosum, and network of abnormal vessels along the falx, supplied by the bilateral anterior cerebral arteries which are dilated and run a tortuous course (arrows)

Table 1 Summary of literature review of case reports and case series of congenital pial arteriovenous fistula (AVF) [11, 13–23]

Case reports/case series	Age at presentation	Duration of follow-up	Presenting symptoms	Associated intracranial findings	Management	Outcomes
Zhang et al. [11]	4 months	6 months	Progressive eye redness	Nil	Endovascular embolization with microcoils	Stable until the date of publication
Aroyo et al. [14]	6 months	36 months	CHF	Nil	Craniotomy and clipping	Stable until the date of publication
Kraneburg et al. [15]	1 month	2.5 months	Intraventricular hemorrhage and hydrocephalus	Nil	Endovascular embolization	Stable until the date of publication
Batista et al. [13]	3 years	5 months	Abnormal facial features and retarded growth	Encephalocranio-cutaneous lipomatosis syndrome	Endovascular embolization with coils and glue	Stable until the date of publication
Ago et al. [16]	At birth	6 months	Cranial bruit	Nil	Surgical resection	Stable until the date of publication
Komiyama et al. [17]	1-month-old (2 cases)	10 years and 2.5 years (2 cases)	CHF	Nil	Endovascular embolization with coils	Stable until the date of publication
Boet et al. [19]	2 years	3 years	Progressive unsteadiness	Nil	Endovascular embolization	Stable until the date of publication
Martinez et al. [20]	Prenatal	N/A	N/A	Nil	N/A	Intra-uterine demise
Pedicelli et al. [21]	Prenatal	4 years	N/A	Nil	Surgical excision	Stable until the date of publication
Koruglu et al. [22]	Prenatal	3 years	N/A	Nil	Endovascular embolization via umbilical artery	Stable until the date of publication
Garel et al. [23]	Prenatal (3 cases)	Patient 1: 2 weeks Patient 2: 1.9 years Patient 3: 6 years	N/A	Nil	Endovascular embolization in all 3 patients	1 died and 2 were stable until the date of publication
Paramasivam et al. [18]	Prenatal	Varies from 1 day to 18 months (most cases were followed up for 6 to 12 months)	Ventriculomegaly	Nil	All 16 cases were embolized with N-butyl-cyanoacrylate (NBCA) whereas coils were also used in 10 of the 16 cases along with NBCA.	4 cases developed subsequent dural arteriovenous fistula. 2 cases had transverse sigmoid sinus occlusion. 1 neonate presenting with congestive heart failure, following embolization developed subarachnoid hemorrhage and subsequently died.
	3 months		Intracranial hemorrhage	Nil		
	14 months		Prominent facial veins	Nil		
	neonate		CHF, pulmonary hypertension	Nil		
	24 months		Seizures	Nil		
	7 months		Macrocephaly, seizures	Nil		
	7 months		Macrocephaly, developmental delay	Nil		
	Neonate		CHF, bluish discoloration	Nil		
	18 months		Staring spells, breathlessness	Nil		
	62 months		Headache	Nil		

Table 1 (continued)

Case reports/case series	Age at presentation	Duration of follow-up	Presenting symptoms	Associated intracranial findings	Management	Outcomes
	36 months		Headache, right hemiparesis	Nil		
	20 months		Seizures	Nil		
	Neonate		CHF	Nil		
	Neonate		CHF	Nil		
	48 months		Intracranial hemorrhage	Nil		
	60 months		Increased head circumference	Nil		

CHF congestive heart failure, *N/A* not applicable

closure has been documented [30]. Our case demonstrated temporal stability of both the VM and brain parenchymal changes over a period of 12 years with no interval development of giant venous aneurysm or intracranial hemorrhage. This suggests that the outcome of pial AVFs with conservative treatment may not be as grim as previously thought. Our case is exceptionally unique due to the associated findings not previously documented, its location along the interhemispheric falx as well as its unpredictably stable natural history despite the absence of active intervention.

Conclusion

Our understanding of pial AVFs is very limited especially in the pediatric age group, largely due to the rarity of the condition. Our case adds incremental value to this rare entity by showing the associated findings that were not previously documented, its location along the interhemispheric falx as well as its unpredictably stable natural history despite the absence of active intervention.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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