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Case Report

Congenital Brachio-Brachial Arteriovenous Fistula: Presentation of a Rare Case and Literature Review

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Abstract: Congenital arteriovenous fistula is an extremely rare vascular malformation. Its symptomatology is variable and depends on its size as well as its location. We report the case of a 12-year-old girl with no known medical or surgical history, who was referred for an ultrasound examination at the Abidjan Heart Institute. Doppler ultrasound of the upper limb revealed a high-flow brachio-brachial arteriovenous fistula. The patient was subsequently transferred to the vascular surgery department for management.

Keywords: Arteriovenous Fistula, Congenital Malformation, Ultrasonography.

INTRODUCTION

Congenital arteriovenous fistula (AVF) is an extremely rare vascular malformation characterized by an abnormal and direct connection between an artery and a vein, bypassing the normal capillary bed [1]. Unlike acquired arteriovenous fistulas, which typically result from surgical procedures performed prior to dialysis, congenital fistulas arise from developmental anomalies of the embryonic vascular system [1, 2]. This diversion of blood flow leads to sometimes significant hemodynamic alterations, whose clinical consequences largely depend on the size of the shunt and its anatomical location [2]. Although any anatomical region can be affected, involvement of the vessels in the arm remains the least common [3]. While some arteriovenous fistulas (AVFs) remain asymptomatic, others may present with suggestive clinical signs such as a pulsatile mass, vascular murmur, or peripheral circulatory disorders that can lead to amputation [3]. In progressive forms, the high shunt flow can impair cardiac function and result in heart failure [2–4]. The etiology of congenital AVFs is not yet fully elucidated; however, various specific genetic mutations implicated in the occurrence of these anomalies have been identified and are currently under extensive investigation [5, 6]. Diagnosis is based on a combined approach incorporating clinical examination, morphological and functional imaging techniques [3]. In this context, the Schöbinger clinical classification, which assesses symptomatology and disease stage, as well as the Yakes angiographic classification, which characterizes the vascular morphology of the lesions, constitute essential tools for guiding therapeutic management [7, 8].

This case report aims to present a rare case of congenital arteriovenous fistula (AVF) and to discuss its specific features in light of current literature data.

CASE PRESENTATION

A 12-year-old girl with no significant medical or surgical history, was referred to the Abidjan Heart Institute for ultrasound evaluation of a pulsatile vascular dilation in the right upper limb. Physical examination revealed, in addition to the pulsatile vascular dilation, a palpable thrill and a continuous murmur localized to the affected area. No signs of local circulatory disturbance were observed. The cardiovascular examination and the remainder of the physical exam were unremarkable. Vital signs were within normal limits: blood pressure 100/60 mmHg, heart rate 100 bpm, respiratory rate 20 breaths/min, temperature 36.6°C, and oxygen saturation of 98% in ambient air (left side versus right side). Anthropometric parameters indicated a weight of 45 kg, height of 1.50 m, and body mass index of 20 kg/m².

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Doppler ultrasound of the right upper limb revealed two arteriovenous communications between the brachial vein and the brachial artery, measuring 5.5 mm and 3.7 mm respectively (Figure 1). Color Doppler imaging demonstrated aliasing was observed at the fistula site (Figure 2). The fistula exhibited high flow, with an estimated mean flow of 3227 ml/min (Figure 3). Transthoracic echocardiography showed no abnormalities. The patient was referred to the vascular surgery department for specialized management.

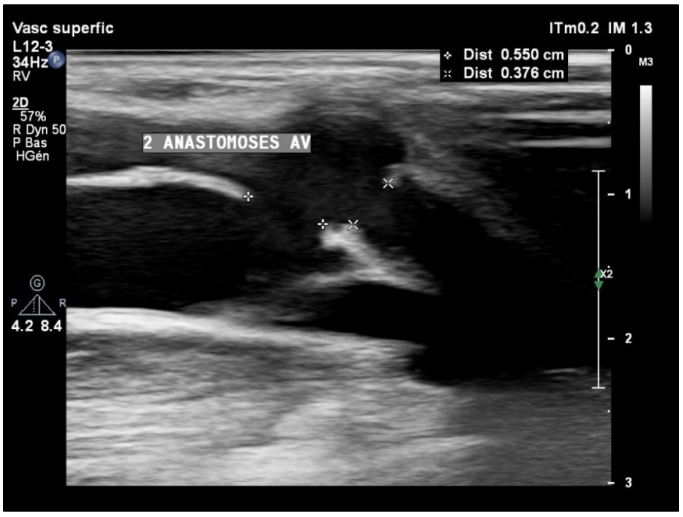

Figure 1: Doppler ultrasound of the right upper limb revealed the presence of two arteriovenous anastomoses between the brachial vein and the brachial artery

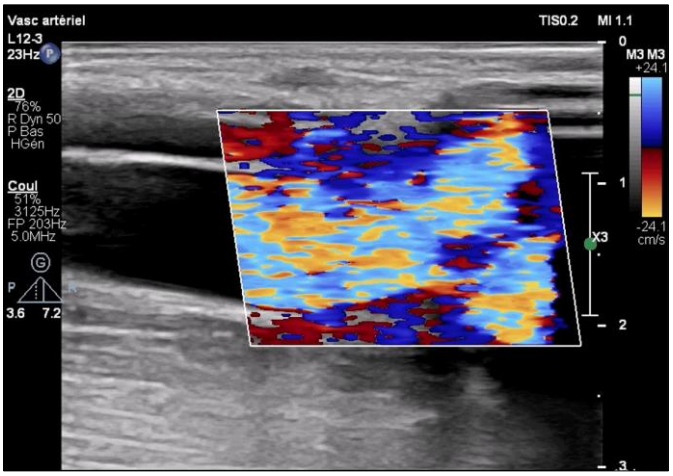

Figure 2: Color Doppler ultrasound of the right upper limb noted Aliasing at the fistula site

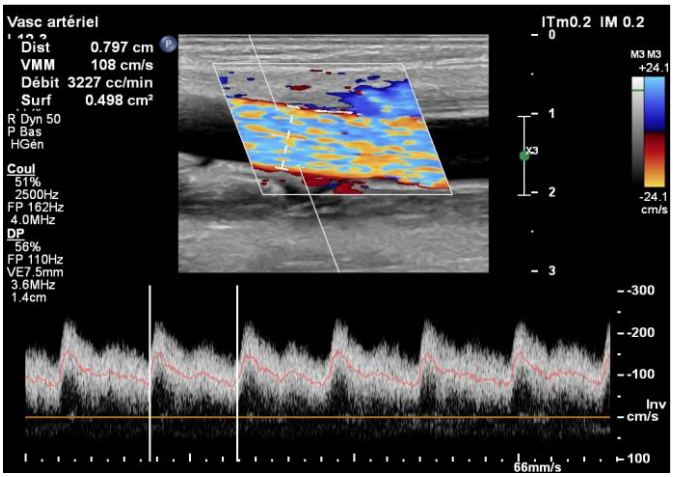

Figure 3: Color Doppler ultrasound of the right upper limb demonstrated High flow of the brachio-brachial arteriovenous fistula

DISCUSSION

Congenital arteriovenous fistula (AVF) is a rare vascular anomaly with a variable clinical spectrum [2, 3]. A 10-year study (2010 to 2020) conducted by Subramanian AP *et al*., [3], in a pediatric population estimated its incidence at 0.009%. Some genes involved in cellular growth and differentiation, including MAP2K1, which encodes the protein Mitogen-Activated Protein Kinase Kinase 1, and KRAS, which encodes the Kirsten Rat Sarcoma Viral Oncogene Homolog protein, have been investigated. Mutations in these genes lead to excessive endothelial cell proliferation and abnormal vascular remodeling, resulting in the direct arteriovenous shunt and thereby implicated in the development of arteriovenous malformations [6]. The symptomatology of AVFs is polymorphic, ranging from minimally or asymptomatic masses to extensive tissue lesions, which can progress to severe heart failure in cases of large, high-flow fistulas [1-9]. These malformations may manifest at any age and affect various anatomical locations, including the scalp, neck, limbs, or internal organs [1-11]. Schwarz K and Pitcher DW [12], reported an asymptomatic congenital subclavian AVF diagnosed in adulthood, illustrating the variability in circumstances of discovery. In contrast, Jabari S *et al*., [4], and Dogan R *et al*., [9], have documented severe congenital AVF cases complicated by heart failure, highlighting the potential progressive nature of these vascular anomalies. Diagnosis is based on clinical examination combined with imaging. Doppler ultrasound is the primary tool, supplemented if necessary by magnetic resonance imaging (MRI) and angiography, particularly for deep or cerebral fistulas [1-3]. The treatment strategy for arteriovenous fistulas (AVFs) depends on their size, location, and associated symptoms [7]. Asymptomatic forms may be subject to regular clinical and imaging surveillance, whereas progressive or symptomatic cases require more aggressive intervention, either via endovascular or surgical approaches. In the absence of formal guidelines, management remains individualized, taking into account the lesion's potential progression and the risk of disabling complications in the event of delayed treatment [13, 14].

There is variability in the approach among centers: some prefer intervention starting from Schöbinger stages III or IV, whereas others opt for early treatment to prevent adverse progression [13]. In our case, the patient was classified as stage II. Several authors recommend embolization as first-line therapy, reserving surgery for complex cases [7-14]. Targeted therapies based on inhibition of the RAS/MAPK signaling pathway, guided by genetic analyses, are currently under extensive investigation. These aim to reduce recurrence risk and offer alternatives to invasive surgical interventions, particularly in cases with multiple lesions [6]. The prognosis is generally favorable for systemic congenital fistulas when detected and treated early, with Doppler ultrasound being vital for both diagnosis and monitoring [1-3]. A study involving 7 pediatric cases demonstrated successful closure in 6 out of 7, with no recurrence over a median follow-up of 4 years [1]. A study by Pinelo A *et al*., [7], on 9 cases reported clinical improvement with reduction of Schöbinger stage in 77.8% of cases and noted complications in some patients when therapeutic decisions were made at advanced stages [7]. Rarely, some small fistulas may close spontaneously. Regardless of whether radical treatment or observational management is chosen, regular follow-up is essential to detect potential recurrence or progression [3]. The recent identification of genetic mutations opens new avenues for genetically guided medical therapies to improve long-term outcomes and reduce associated morbidity [6].

CONCLUSION

Congenital arteriovenous fistulas remain a diagnostic and therapeutic challenge due to their polymorphic nature and often unpredictable progression. Early recognition and multidisciplinary management are essential to limit complications and improve prognosis. Advances in imaging techniques, minimally invasive approaches, and targeted therapies based on genetics now offer promising prospects for safer management of these vascular anomalies.

REFERENCES

1. Subramanian AP, Jayranganath M, Bharath AP, Barthur A, Sastry UMK, Moorman AFM. Congenital systemic arteriovenous fistulas: Interventional strategies and embryological perspectives. *Ann Pediatr Cardiol.* 2022 Mar-Apr;15(2):138-146.
2. Jayroe H, Foley K. 2022. Arteriovenous fistula. StatPearls. *Treasure Island (FL):* Stat Pearls Publishing; 2022.
3. Shoab SS, Scurr JH. Arteriovenous malformations of the upper limb. In: Chang JB, editor. *Textbook of Angiology.* New York: Springer; 2000.
4. Jabari S, Hartmann A, Cesnjevar R. A rare case of congenital thoracic arteriovenous fistula between the brachiocephalic truncus and the superior vena cava resulting in heart failure. *J Pediatr Surg Case Rep.* 2016;15:50–2.
5. Revencu N, Boon LM, Mendola A, Cordisco MR, Dubois J, Clapuyt P, et al. RASA1 mutations and associated phenotypes in 68 families with capillary malformation-arteriovenous malformation. *Hum Mutat.* 2013;34:1632–41.
6. Gilbert P, Dubois J, Giroux MF, Soulez G. New treatment approaches to arteriovenous malformations. *Semin Intervent Radiol.* 2017;34:258–71.
7. Pinelo A, Loureiro L, Almeida P, Mendes D, Veterano C, Castro J, Rocha H, Almeida R. Endovascular approach as first-line therapy for congenital arteriovenous malformations – a case series. *Angiol Cir Vasc*. 2023 Jan. 7.

8. Yakes W, Baumgartner I. Interventional treatment of arterio-venous malformations. *Gefasschirurgie.* 2014;19:325–30.
9. Dogan R, Yilmaz M, Ozkutlu S, Elsharshari H. Congenital subclavian artery to subclavian vein fistula presenting with congestive heart failure in an infant. *Pediatr Cardiol.* 2000;21:269–70.
10. Valdeci WP, Gilmone RL, Rosa RM, Rafael C. Giant scalp arteriovenous malformation. *Rev Assoc Med Bras.* 2016;9:828–30.
11. Lim JJ, Jung JI, Lee BY, Lee HG. Prevalence and types of coronary artery fistulas detected with coronary CT angiography. *AJR Am J Roentgenol.* 2014;203:W237–43.
12. Schwarz K, Pitcher DW. Congenital subclavian arteriovenous fistula. *BMJ Case Rep.* 2009;2009:bcr09.2009.2312.
13. Lee BB, Do YS, Yakes W, Kim DI, Mattassi R, Hyon WS. Management of arteriovenous malformations: a multidisciplinary approach. *J Vasc Surg.* 2004;39:590-600.
14. Samir El Youbi, Salma Boulman, Hamza Naouli, Hamid Jiber, Abdellatif Bouarhroum, Surgical management of a congenital arteriovenous fistula of the forearm, *Journal of Surgical Case Reports*, Volume 2022, Issue 6, June 2022, rjac259.

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