CASE REPORT

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Surgical Excision of a Childhood Congenital Vascular Malformation in the Leg: A Rare Inborn Lesion

Hamit Serdar BAŞBUĞ[a], Volkan KIZILGÖZ[b]

[a]Department of Cardiovascular Surgery, Kafkas University Faculty of Medicine, Kars, Türkiye
[b]Department of Radiology, Erzincan Binali Yıldırım University Faculty of Medicine, Erzincan, Türkiye

ABSTRACT Congenital vascular malformations are rare anomalies resulting from an inborn error during the vascular network development. They are rare inborn lesions characterized by deficient smooth muscle layer leading to vascular dilatation with impaired functions. Congenital vascular malformations may occur in a spectrum of clinical presentation ranging from ectasia or varicosity to a more complicated lesion that can affect any organ system. Although they are present at birth, they grow proportionally during childhood and adolescence, becoming clinically evident later in life. The symptoms differ depending on location, extension, and size. They may bleed and result in disfigurement, functional impairment, and pain. In this case report, successful surgical treatment of a child with a congenital vascular malformation in his right leg was presented. This case and its detailed intraoperative and radiological images are original as there is no similar example in the available literature.

Keywords: Congenital abnormality; lower extremity; vascular malformations

Congenital vascular malformations (CVMs) are rare entities caused by the defective development of the vascular system during embryogenesis.[1] CVMs are present at birth as a congenital disability arising from different embryogenic stages and may involve arterial, venous, lymphatic, and capillary systems.[2] Histologically, these malformations have an irregular vascular coil with thickened walls and extensive anastomoses that dissect the surrounding tissues.[3] CVMs are a mixture of various vascular defects with different characteristics. A CVM may appear at various localizations, conditions, morphology, dimensions, and severities. They may also occur as an independent or a complex lesion.[1] According to the modified Hamburg classification, CVMs are classified based on the malformation's predominant vascular structure. This primary classification divides the CVMs into 6 subgroups: arterial, venous, arteriovenous, capillary, lymphatic, and combined-vascular malformations.[4] CVMs may be profound without skin involvement. Therefore, they often remain undiagnosed until a swelling, pain, or functional impairment happens.[5] The CVMs are also misdiagnosed as benign subcutaneous tumors like lipomas.[6] We present a 13-year-old boy with a CVM on his right leg that was successfully excised with a surgical operation. This paper about a childhood CVM is original with its detailed intraoperative and radiological images.

CASE REPORT

A 13-year-old boy applied to the cardiovascular surgery outpatient clinic with a localized mass on his right leg. Anamnesis revealed that the mass was present and palpable when he was 3-year-old with a hazelnut size and grew proportionally with his growth. The inspection and physical examination

Correspondence: Hamit Serdar BAŞBUĞ
Department of Cardiovascular Surgery, Kafkas University Faculty of Medicine, Kars, Türkiye
E-mail: s_basbug@hotmail.com

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demonstrated a painless solid 5x4 cm mass on the anterolateral mid-tibial portion of his right leg. There were no signs of circulatory defect, and pedal pulses were palpable. The contrasted magnetic resonance imaging (MRI) was then planned. The MRI was performed using a 1.5 T MR machine (Magnetom Essenza; Siemens, Erlangen, Germany) and an 8-channel extremity coil. The axial (Figure 1A) and coronal (Figure 1B) planes of fat-saturated T2 weighted, fat-saturated T1 weighted, and contrast-enhanced T1 weighted images were used as imaging sequences in the standard extremity MRI. The child was diagnosed with a CVM located beneath the dermis, superficial to the crural muscles and fascia.

The mass was surgically removed with a longitudinal skin incision under local anesthesia (Figure 2). A single small artery which was the feeder of this vascular malformation was separated and cauterized. The excised material was sent to the pathology lab (Figure 3). The histopathology report demonstrated a benign mesenchymal-origin vascular structure surrounded by a thin fibrous capsule compatible with a CVM. There were no mitosis, necrosis, and atypic structures within the lesion, proving the lesion was not malignant. The boy was discharged on postoperative second day with a total cure. The twenty-eighth-day follow-up revealed a clean and healed skin incision with no hematoma inside. Written informed consent was obtained from the patient's parents to publish this case report and accompanying images.

DISCUSSION

CVMs are rare pathologies affecting children with different morphology and multiple diagnostic features. They may be present as bony, intramuscular, subfascial, epifascial, subcutaneous, and cutaneous locations.[7] The cutaneous and subcutaneous CVMs may be manifested with blue skin color.[8] In this case, there was no skin discoloration as the CVM was located subcutaneously and epifascially. However, a localized soft swelling was easily observed as an epifascial CVM was present. CVMs may rarely cause skeletal changes with osseous involvements.[7] No bony deformity of the tibia or fibula was seen in this case.

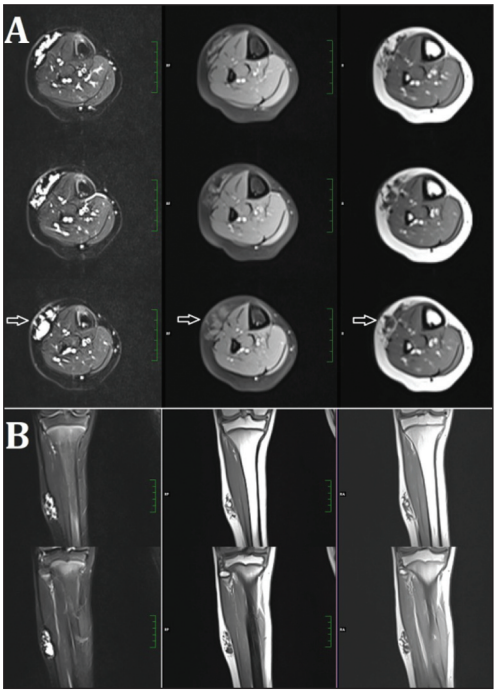

FIGURE 1: Fat saturated T2 weighted (left column), fat-saturated T1 weighted (middle column), and contrast-enhanced T1 weighted (right column) magnetic resonance images of the same axial planes are shown. The vascular lesion was observed in the middle crural level, located in the subcutaneous fat planes **(A)**. Coronal plane magnetic resonance images of the same patient. Fat saturated T2 weighted (left column), fat-saturated T1 weighted (middle column), and contrast-enhanced T1 weighted (right column) magnetic resonance images of the same coronal levels are shown. The vascular lesion was observed in the middle crural level, located in the subcutaneous fat planes **(B)**.

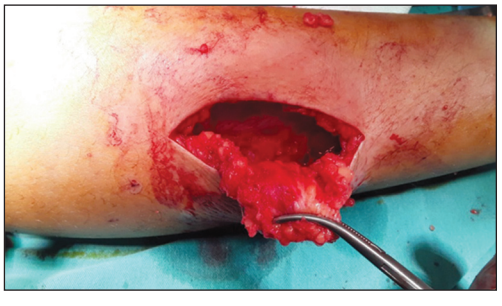

FIGURE 2: Intraoperative image of enucleation and removal of the mass.

The spectrum of CVMs is evolving at the interaction between various medical specialties. Its nomenclature and its descriptive terminology have camouflaged this subject for a long time. Similar terms have been used for entirely different lesions leading to a misunderstanding of the pathology. For example, the term "hemangioma" is the most com-

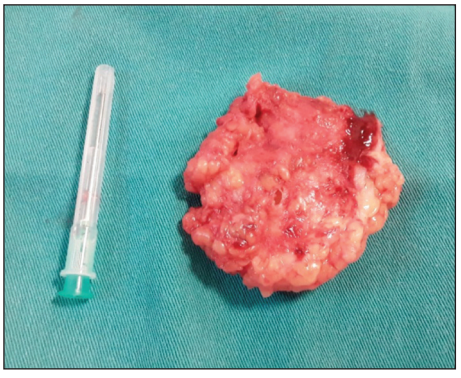

FIGURE 3: Morphology and size of the excised mass.

mon example applied to all vascular lesions of different etiologies and clinical presentations. This bias in the terminology then causes misdiagnosis, improper treatment, and misdirected academic facilities. Interdisciplinary dialogue is then interrupted as each specialty has its terminology.[9] According to Mulliken and Glowacki's classification, vascular anomalies were divided into vascular tumors and vascular malformations.[10] According to the clinical behavior and cellular kinetics, vascular tumors demonstrate endothelial hyperplasia, while the vascular malformations exhibit average endothelial turnover and arise by dysmorphogenesis.[9] Vascular tumors may tend to regress, whereas vascular malformations increase in size with age and never regress.[11] CVMs are rheologically subdivided into slow-flow (capillary, lymphatic and venous malformations) and fast-flow (arterial malformations -e.g., aneurysms, ectasia, arteriovenous fistulas, stenosis- and arteriovenous malformations) lesions.[12] According to their incidence, venous malformations are the most common (70%) among all other CVMs. Lymphatic malformations (12%), arterio-venous malformations (8%), combined malformation syndromes (6%), and capillary malformations (4%) are the other types of CVMs.[11]

Discussing the etiology of vascular anomalies, knowledge about the genetic causes has evolved in recent years. Postzygotic somatic mutations play a role in vascular anomaly formation with many specific genes that cause inherited malformations.[13] The endothelial receptor tyrosine kinase gene TEK has a vital role in angiogenesis and vascular development. Somatic changes causing loss of function in this angiopoietin receptor gene play a role in the etiology of solitary and multiple sporadic CVMs.[14]

Treatment of CVMs is a major clinical challenge. Surgery is often unsuccessful and hard to perform. Endovascular intervention with embolization has an ill-defined role, and conventional foam sclerotherapy has little contribution.[12] Surgical approaches mainly target to reduce the hemodynamic activity of the lesion, eliminate the malformation, and reconstruction.[15] In this case, we performed a successful surgical intervention resulting in the total excision of the well-delineated mass. The success was because the CVM was encapsulated and located between the fascia and skin. The removal was easy as the CVM has no surrounding infiltration.

In conclusion, CVMs are a challenge that requires a dedicated multidisciplinary diagnostic approach and therapeutic management, especially in the treatment of children. Establishing an integrated multidisciplinary medical evaluation will lead to diagnose and treat these rare complex vascular entities better. Patient care reaches an optimum standard only in this manner.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Hamit Serdar Başbuğ;* ***Design:*** *Hamit Serdar Başbuğ, Volkan Kızılgöz;* ***Control/Supervision:*** *Hamit Serdar Başbuğ, Volkan Kızılgöz;* ***Data Collection and/or Processing:*** *Voklan Kızılgöz;* ***Analysis and/or Interpretation:*** *Hamit Serdar Başbuğ;* ***Literature Review:*** *Hamit Serdar Başbuğ;* ***Writing the Article:*** *Hamit Serdar Başbuğ, Volkan Kızılgöz;* ***Critical Review:*** *Volkan Kızılgöz;* ***References and Fundings:*** *Volkan Kızılgöz;* ***Materials:*** *Hamit Serdar Başbuğ.*

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