Tips for Contemporary Management of Congenital Arteriovenous Malformations

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Congenital vascular malformations (CVMs) are a relatively rare and complex group of lesions, which may present significant pitfalls in diagnosis and treatment. These vascular lesions develop by embryologic dysmorphogenesis without increased endothelial proliferation that leads to structural and functional anomalies of the vascular system characterized by a wide range of presenting symptoms and frequently unpredictable clinical course.1-3 The underlying etiology responsible for the development and progression of CVMs remains to be elucidated, although recent genetic data suggest that reduced ability to regulate the signaling processes at different stages of vascular cells’ cycle has an important role in the pathogenesis of CVMs.4,5

The diagnosis and management of vascular malformations remains challenging because of a variety of factors. Historically, the classification and nomenclature utilized to describe CVMs have not been primarily based on lesion physiology and/or hemodynamics, leading to confusion and inconsistency about the true nature of these vascular lesions. Most commonly, the term hemangioma was erroneously applied to all vascular anomalies, including CVMs.6,7 In addition, the use of eponymous designations of certain lesions, which especially characterized a significant part of the early literature, didn’t provide information about lesion type and hemodynamics, leading to further confusion. A recent advancement in the diagnostic and treatment modalities coupled with a concept of the multidisciplinary team approach (characterized by full integration of expertise of different medical specialists) have resulted in a better understanding of the pathophysiology and natural history of CVMs as well as improved management of these lesions.8,9

Because the management of CVMs falls within the purview of many medical specialties, it is critical to establish a multidisciplinary approach for their treatment by combining the expertise of each specialty, in particular vascular, pediatric, reconstructive, and orthopedic surgery; adult and pediatric hematology; adult and pediatric dermatology; adult and pediatric ophthalmology; and diagnostic and interventional radiology. This approach affords the opportunity to streamline the evaluation process for patients with vascular anomalies, coordinate the care of these patients, and treat them comprehensively, reducing the need for multiple visits to different clinics as well as patient frustration.

The morphology, clinical presentation, and course of CVMs are variable in both their extent and severity, depending upon location, size or organ involved, and the type of vessel affected. They are rarely asymptomatic. Usually they cause discomfort, pain, hemorrhage, and thrombosis, and they negatively affect patients’ appearance and emotional well-being. In addition, these patients often have a significant reduction in daily functional capacity and quality of life. Arteriovenous vascular malformations are characterized by direct arterial flow, or direct connection between the arterial and venous system caused by a congenital disorder that results in fistulous arteriovenous tracts.10 Patients usually present with skin discoloration, elevated cutaneous temperature, dilated veins, and frequently a thrill or a bruit that can be appreciated over the affected area. Patients also can present with pain, cutaneous ischemia, hemorrhage or infection, and even ulceration. If extensive, these lesions can lead to high-output cardiac failure due to large-volume arteriovenous shunting.

Arteriovenous malformations confer the worst prognosis of all CVMs and are associated with progressive growth and recruitment over the patient’s lifetime. The most important initial step in the management is to differentiate CVMs from hemangiomas, as the clinical course and long-term consequences are distinctly different. Arteriovenous malformations are present (but not always apparent) at birth, they grow proportionately with the child and do not regress over time. In contrast, hemangiomas as true neoplastic disorders pathohistologically demonstrate increased endothelial cellular turnover rate and manifest during the first several weeks of life, proliferate rapidly with disproportionate growth relative to the child, then frequently slowly involute over a period of years.11 The second step in the diagnostic algorithm, which follows initial differentiation between CVMs and hemangiomas (as well as other vascular neoplasms), is hemodynamic assessment of the lesion, which leads to differentiation between low-flow vascular malformations (LFVMs) and high-flow vascular malformations (HFVMs). Following this, differentiation between extratruncular and truncular subtypes of lesions has been
clinically validated as a very important next step in the diagnostic algorithm due to the vastly different embryologic, morphologic, and clinical characteristics between these two lesions.12 Morphologically, truncular lesions are those that affect larger, named blood vessels and can be definitively and permanently corrected, usually with surgery. Although there is insufficient evidence in the current literature to make a conclusive determination regarding the exact underlying pathphysiologic mechanisms responsible for differentiation between these two forms of HFVMs, data from early morphologic studies demonstrated that preservation of the embryologic proliferative potentials determined the more aggressive clinical course, unpredictable progression, and higher recurrence rates associated with extratruncular malformations.12 In contrast, truncular malformations originate from cells that have already undergone the mesenchymal maturation stage and consequently do not retain proliferative embryologic potential.12 These pathphysiologic characteristics are responsible for the significantly better clinical course and treatment outcomes associated with truncular malformations. In addition to these above mentioned diagnostic steps, it is critically important that evaluation of the patency and anatomic variations of the entire vascular system (deep and superficial, proximal and distal) as well as involvement of relevant anatomic structures by modern imaging modalities be performed to establish a definitive treatment plan.

Traditionally, multiple diagnostic modalities were used (alone or combined) to evaluate vascular malformations and to confirm the initial clinical diagnosis including ultrasound (US) imaging, computed tomography (CT) scan, catheter-based angiography, and magnetic resonance imaging (MRI). Although still frequently used by some practitioners, US and CT provide variable degrees of diagnostic accuracy or frequently insufficient information for preprocedural planning.13 Consequently, a significant number of patients was left undiagnosed or required evaluation with catheter-based angiography. Given these limitations, MRI has become the imaging modality of choice in the confirmation, characterization, and differentiation between vascular malformations and their subtypes. It gives a bright signal on T2-weighted spin-echo sequences for the parenchymal portions of vascular lesions, which is not only useful to delineate the extent of the malformation throughout the involved tissues, but also allows for treatment planning and can be used for objective assessment of the treatment efficacy.14 The full extent of tissue involvement is readily depicted when T1 and T2 or short tau inversion recovery (STIR) images are acquired.14 However, with conventional MRI it is frequently not possible to differentiate between different types of CVs in more complex cases as it does not provide hemodynamic data. Recently, dynamic contrast-enhanced magnetic resonance imaging (dceMRI) was clinically validated as the imaging modality of choice as it can definitively distinguish HFVM from LFVM with accuracy of approximately 84%, mitigating the need for invasive angiography, which is now required only in inconclusive cases.15,16

The specific indications to intervene on CVs remain controversial, particularly for HFVMs. The decision for intervention must consider presenting symptoms, the size of the CVM, location, proximity to vital structures, the natural history of the lesion, the risk of complications, and the relative risk of surgical or endovascular intervention.17 Asymptomatic patients and patients with mild symptoms that do not interfere with daily life are treated conservatively. Patients with diffuse and complex HFVMs are informed that, in most cases, multiple treatment sessions and/or multimodal treatment strategy, which includes preoperative embolization followed by complete surgical resection and/or sclerotherapy of the remaining venous component, are required.9,18 We evaluated the efficacy and safety of above-described treatment algorithm implemented by our multidisciplinary team in a large series (N=136) of CVM patients.20 There were 105 (77.2%) LFVMs and 31 (22.8%) HFVMs. Patients with HFVMs required a multimodal treatment approach, which resulted in symptomatic improvement in 89.5% of patients.19 Our experience provides strong evidence that experienced providers working in the context of a coordinated and structured multidisciplinary team offer efficient treatment of patients with CVs. It must be emphasized that treatment of extensive arteriovenous malformations is palliative and goal oriented, rather than curative.

A multidisciplinary approach and diagnostic algorithm utilized to distinguish CVs from hemangiomata and HFVM from LFVM have been validated as clinically applicable for making an accurate anatomical and hemodynamic diagnosis of CVs. They also serve as a basis for proper treatment selection, and significantly facilitate communication among different medical specialists. Implementation of the proposed diagnostic protocols and therapeutic algorithms in a multidisciplinary setting results in favorable outcomes with an acceptable complication rates in this challenging patient population.

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REFERENCES


