What the Vascular Surgeon Should Know about Congenital Vascular Malformations

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Congenital vascular malformations (CVMs) present a formidable challenge to the physician. Patients suffering from gross disfiguring lesions go from doctor to doctor in search of someone with the interest and experience to tackle their challenging problems. Most physicians have little or no experience in the diagnosis and management of CVMs. For this reason, these patients are referred to multidisciplinary specialized centers with the resources, both human and material, to deal with these problems.

The incidence of CVM in the general population is 1.5%. They are defined as "an embryologically developed, inborn error of vascular morphogenesis leading to true structural anomalies. By definition, they are congenital (present at birth)."[1] The variable presentations of CVMs have led to errors in diagnosis and consequently misguided treatment. For this reason, it is essential to classify them. Because of its simplicity, we have followed the classification described by Malan, which, with somewhat different terminology, is similar to the classification adopted in Hamburg in 1988.[2] In Malan's classification, CVMs are divided into five main groups: arterial (aneurysms, atresia, ectasias), venous (localized or diffuse), arteriovenous (localized or diffuse), lymphatic (previously called cystic hygroma), and mixed. Mixed malformations involve several vascular systems; the most common is the combination of the venous and lymphatic systems.

Therefore, the vascular surgeon should be familiar with the classification of CVMs and be able to distinguish between the two main types of congenital vascular anomalies as described by Mulliken.[1] The latter made an important contribution in classifying the vascular birthmarks according to their embryologic characteristics. He classified them into hemangiomas and malformations. Clinically, the hemangioma may be either not present at birth or shown as a small reddish birthmark. In the first few months of life, some of these lesions proliferate very rapidly and then proceed in a few years to a slow regression. They are present most commonly in females. Some large hemangiomas may produce thrombocytopenia (Kasabach-Merritt syndrome) and present with bleeding. On the other hand, these malformations are usually present at birth even though sometimes they are not noticeable. They may be capillary, venous, arterial, lymphatic, or mixed. Generally, they have a parallel growth with the child and may flair up as a consequence of an accident or hormonal changes (puberty). Angiography may show a low flow malformation in cases of venolymphatic malformations such as the Klippel-Trénaunay syndrome (KTS), Maffucci's syndrome, and others. CVMs with localized or diffuse arteriovenous shunting show a high flow with enlarged arteries and tortuous dilated veins. The bone may be affected by either hypertrophy or hypoplasia.

Venolymphatic malformations, in particular, may be associated with large varicosities that, for some physicians, may appear as simple varicose veins easily managed by the usual ablation procedures for the treatment of varicose veins. However, the vascular surgeon must be aware that some venous malformations, particularly those that extend throughout the entire extremity, may be associated with anomalies of the deep venous system. In our experience with 392 patients with CVM, two-thirds of the CVMs were of venous predominance and in almost one-half there were anomalies of the deep venous system presenting as phlebectasias, aplasia/hypoplasia, venous aneurysms, and avalvulia. Of course, excising the large superficial venous outflow of the extremity in a patient with atresia of the deep venous system may lead to disastrous hemodynamic consequences.

Diagnostic Procedures

The vascular surgeon interested in CVM must become familiar with the most frequently used diagnostic procedures. Identification of the vascular system involved in a malformation is essential for its treatment. Given its complex nature, it is necessary to have the collaboration of a multidisciplinary group of experts that includes radiology, interventional radiology, cardiology, general and vascular surgery, and plastic, orthopedic, and neurosurgeons. The collaboration of this team of experts is essential for the benefit of the patient.

The vascular surgeon should know when to intervene in the course of a CVM. In general, it has been our policy that if a child has a congenital malformation that does not interfere with his/her lifestyle, this patient should not be submitted to diagnostic examinations that may be painful and predispose the child to avoid future encounters with a physician. Therefore, the timing to carry out diagnostic examinations and therapeutic interventions is entirely dependent of the severity of the patient's symptoms. If a malformation is disfiguring or localized to anatomic areas submitted to frequent trauma such as the knee, elbow, foot, then it should be treated as soon as possible. The same applies to those malformations localized in vital areas such as mouth, nose, pharynx, and eyes. If symptoms are life-threatening, the malformation should be aggressively investigated to proceed with any therapeutic procedure necessary to solve the problem. Malformations involving larynx and eyes are particularly troublesome since they may produce respiratory difficulties and/or permanent visual impairment.

The vascular surgeon should be aware of those diagnostic studies that are most useful for the diagnosis of CVM. Most commonly, a careful clinical examination will establish a working diagnosis. However, very often it is necessary to resort to diagnostic procedures such as Duplex scanning with color flow imaging, strain gauge plethysmography (mostly used in research protocols), magnetic resonance imaging (MRI), and positron emission tomography (most useful in patients with malformations involving the brain). Invasive studies such as arteriography and phlebography must be performed when surgery or invasive treatments such as embolization of diffuse or localized arterial venous fistulae are being considered.

During the past decade, an awareness of the presence of hidden lesions distant to the apparent CVM has led to the development of techniques such as transarterial radioisotope studies with labeled microsphere albumin (whole body blood pool scan), magnetic resonance imaging, and lymphoscintigraphy.[3] MRI has been particularly useful in the assessment of the magnitude of the involvement of the vessels, muscle, and soft tissue in malformations such as KTS and other diffuse malformations. Contrast MRI has been useful in diagnosing hypoplastic or absent venous segments. Invasive angiography is used in complex cases where the noninvasive diagnostic methods are not conclusive. Ascending or descending phlebography is essential in those patients in whom surgery for large superficial varicosities is considered and the surgeon must investigate the anatomy and integrity of the deep venous system. Some intractable venous ulcers that are not unusual in patients with diffuse venolymphatic malformations may be secondary to hemodynamic venous obstruction owing to aplasia or hypoplasia of the venous system.

Phlebography, both ascending and descending, and selective varicography are of importance in the diagnosis of venous aneurysms, phlebectasias, and avalvulia. In the latter, the contrast material injected in retrograde fashion may be observed refluxing from the groin to below the knee without the presence of any apparent valves such as occurs in an arteriogram. Venous aneurysms are particularly dangerous because they are usually asymptomatic and their first manifestation may be an episode of severe or fatal pulmonary embolism.[4]

Management

The vascular surgeons should be familiar with the therapeutic alternatives available in CVM. In the case of rapidly growing, threatening hemangiomas affecting vital organs such as the nose and larynx or those that manifest with bleeding and ulceration, systemic corticoids should be the first line of treatment.[5] Because of its angiogenic nature, hemangiomas respond well to antiangiogenic therapy. Intralesional corticoids are useful in localized lesions in the lips, nose, and eyelids.[6] In children with life endangering hemangiomas not responding to corticoids, the subcutaneous administration of daily injections of recombinant interferon-? (2a or2b) has been proven effective and often life saving. Laser therapy has become an important therapeutic adjunct in the management of superficial hemangiomas (capillary) localized to the face, neck, and other exposed areas.

Surgery is indicated for most forms of relatively localizedmalformations either venous or arteriovenous. Excision of the malformation after intravascular embolization with glues, microcoils, sclerosant agents, or even absolute alcohol has proven to be an effective therapeutic measure.[7] In patients with arteriovenous fistulae, a multidisciplinary approach combining embolization, sclerotherapy, and surgery offers a reasonable therapeutic alternative in these otherwise intractable patients. Finally, the vascular surgeon must know that in most diffuse malformations, cure is not possible but an acceptable degree of palliation is always possible.

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