General Overview

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Congenital vascular malformations (CVMs) remain a significant clinical challenge due to the nature and complexity of the vascular lesions encountered in clinical practice [1, 2]. CVMs are often confused with hemangiomas [3]. The term "hemangioma" is commonly used to name different types of vascular tumors and vascular malformations, despite the fact that these vascular anomalies are distinct vascular lesions. CVMs and hemangiomas not only have different etiologies, anatomy, and pathophysiology, but they also exhibit unique hemodynamic and embryologic characteristics [2–6]. Both of these vascular anomalies have entirely different clinical courses and long-term prognoses. Furthermore, the management strategies of both conditions are fundamentally different [2–6].

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Hemangioma

A hemangioma is vascular tumor, the most common type being the infantile hemangioma [3, 6]. The hemangioma is a "self-limited" vascular tumor, while the CVM is a "self-perpetuating" embryologic tissue remnant. The hemangioma originates from endothelial cells and appears in the early neonatal period. Unlike CVMs, hemangiomas undergo self-limited growth followed by subsequent involution that usually occurs before the age of 5–10 years [3, 6].

Most hemangiomas are small and produce only minor clinical problems before they involute and become clinically silent. However, about 20 % become clinically significant and require treatment [3]. Indications for treatment include aggressive growth, tumor proximity to vital anatomic structures, ulceration, and bleeding. In addition, the disfiguring nature of certain lesions may prompt parents to seek early intervention [3, 6].

Congenital Vascular Malformation

A CVM is a vascular anomaly that presents at birth and continues to grow at a rate proportional to the growth rate of the body regardless of its type. Data reported by Tasnadi et al. show an overall incidence of CVMs to be 1.2 % [7]. These birth defects involving the vascular system are present at birth in over 90 % of cases, with a male

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to female ratio of 1:1 [7]. The majority of CVMs are either venous malformations (VMs) or lymphatic malformations (LMs) [7–9]. VMs represent approximately 2/3 of all CVMs [7–9].

CVMs are classified by vessel type according to the Hamburg classification (refer to Table 9.2) [9, 10]. In each class of vascular malformation, "extratruncular malformations" composed of small vessels intimately embedded in the host tissue and "truncular malformations," affecting individual large vessels, are recognized [1, 2, 4, 5].

Extratruncular vascular malformations arise when developmental arrest has occurred during the reticular stage of embryonic development [1, 2, 4, 5]. The mesenchymal cell properties persist, and the lesions retain the potential to proliferate when stimulated (e.g., menarche, pregnancy, hormone, surgery, and trauma).

Truncular vascular malformations arise when developmental arrest has occurred during vascular trunk formation at the later stage of embryologic development [1, 2, 4, 5]. These lesions no longer possess the potential to proliferate. Truncular lesions present as poorly developed, immature vessels (aplasia or hypoplasia) or as hyperplastic vessels (hyperplasia) [1, 2, 4, 5].

Arteriovenous Malformations

The incidence and prevalence of arteriovenous malformations (AVMs) is reported to be approximately one per 250,000 individuals in the USA based on cerebrospinal AVMs [11]. They present in the age range of 20–40 years with a male to female ratio of 1:1 or 1:2 [11]. Peripheral AVMs represent approximately 10–20 % of all clinically significant CVM lesions [12].

The AVM is the most challenging of all the CVMs due to the wide range of clinical presentations, unpredictable clinical course, complex anatomy and hemodynamics, pathophysiology, and high morbidity [13, 14]. The AVM anatomic defect results in shunting of arterial blood into the venous system [13, 14]. The majority of AVMs present as single component lesions but may have other CVM lesion components, complicating the diagnosis and treatment [13, 14]. These mixed CVMs often become a clinician's nightmare (e.g., Parkes-Weber syndrome) where management is difficult, and treatment results are often disappointing (e.g., micro-shunting AVM) [13, 14].

Extratruncular AVM lesions maintain the original reticular network resulting in AV shunting with no capillary check valve system [13, 14]. The "nidus" of the lesion retains its "non-fistulous" condition (in contrast to the truncular lesion with its "fistulous" condition). The extratruncular lesion produces significant hemodynamic alterations to both the arterial and venous systems. Extratruncular AVM lesions proliferate in response to stimulation (e.g., trauma, surgery, hormone, menarche, pregnancy), resulting in an increase in its size, extent, and severity. Suboptimal treatment of extratruncular AVMs often results in lesion recurrence [13, 14].

In contrast, the truncular AVM lesions affect individual large vessels and carry no risk of recurrence. However, these lesions often have more serious hemodynamic effects on the vascular system compared with the extratruncular form. Truncular AVMs persist as "fistulous" lesions with a direct connection between an artery and vein with no defined "nidus." This fistulous lesion produces significantly more serious hemodynamic sequelae such as cardiac failure, arterial insufficiency, and chronic venous insufficiency [13, 14].

Venous Malformations

Venous malformations clinically present as a single component lesion or combined with other CVMs: lymphatic malformation (LM), arteriovenous malformation (AVM), and/or capillary malformation (CM) [15, 16].

Extratruncular VMs often present as infiltrating lesions or as clusters of primitive venous tissue without direct involvement of the main venous trunk. Truncular VMs present as poorly developed, immature venous structures (aplasia or hypoplasia) or as hyperplastic venous vessels (hyperplasia) [15, 16]. Hypoplastic or hyperplastic truncular VM lesions produce venous obstruction or dilatation, respectively. Examples of truncular VMs include popliteal vein aneurysms, azygous vein stenosis, and intraluminal defects within the

vein (e.g., vein webs or membrane) resulting in stenosis or obstruction [15, 16]. Truncular VM lesions may also present as a persistent fetal remnant vein that has failed to involute or regress normally (e.g., marginal and sciatic embryonic veins) [15, 16]. Truncular VM lesions have more significant hemodynamic consequences than do extratruncular VMs, due to the direct involvement of the lesion with the truncal venous system often producing venous stenosis or obstruction [15, 16].

Lymphatic Malformations and Capillary Malformations

Lymphatic malformations are made up of variously dilated lymphatic channels or cysts, lined with endothelial cells with a lymphatic phenotype [9, 17, 18]. The extratruncular LMs are classified as microcystic, macrocystic, and mixed subtypes. There is no uniform consensus regarding the definition of microcystic or macrocystic LMs [9]. A useful distinction is whether the cysts can be successfully aspirated/sclerosed, resulting in a decrease in LM size, with the smaller cysts being more challenging [19]. Radiographic features also can help to define the difference because macrocystic LMs often have discernible fluid-filled areas. The primary lymphedemas are considered truncular forms of LMs and are characterized by a poorly developed lymphatic system or agenesis of the lymphatic network. LMs are often associated with other CVMs [9, 17, 18].

Capillary malformations (CM), including fading capillary stains and port-wine stains, are among the most common vascular malformations affecting the skin and mucosa [9]. CMs occur in approximately three of 1000 infants, are present at birth, and have a male to female ratio of 1:1 [9]. They are of minimal clinical significance (mainly a cosmetic issue) and are most often associated with other CVMs [1, 2, 9].

Evaluation

CVMs commonly occur as mixed lesions presenting with AVM, VM, LM, and/or CM components [1, 2, 4, 5]. Therefore, the evaluation of any suspected CVM should proceed in a logical, stepwise manner, bearing in mind that any suspected CVM lesion may actually prove to be a mixed lesion. Diagnosis of a suspected VM or AVM requires specific evaluation and confirmation as a single component lesion or mixed lesion.

As a general rule, the extent and severity of any CVM affecting the vascular system (anatomically and hemodynamically) usually determine the type of clinical manifestations observed [1, 2, 4, 5]. The history and physical examination should be followed by diagnostic imaging in order to determine the type of CVM suspected. When an AVM is suspected, workup should proceed to confirm the diagnosis and to distinguish the AVM from among the various CVMs (e.g., duplex ultrasonography and MRI). Most AVMs occur as single component lesions [1, 13, 14].

A mixed CVM with VM and LM components is classified as a hemolymphatic malformation (HLM) [1, 10]. Klippel-Trenaunay syndrome is an example of a HLM. When an AVM is present in a patient with Klippel-Trenaunay syndrome, it is also known as Parkes-Weber syndrome [20]. In this situation, the initial priority of investigation should be to confirm the presence of the AVM component.

Initial diagnostic imaging should be performed with a combination of baseline noninvasive imaging [2, 4, 5, 15, 16]. More specific diagnostic imaging should then follow for further assessment of the embryological subtype (extratruncular vs. truncular) of the AVM or VM. The recommended initial studies include duplex ultrasonography, magnetic resonance imaging (MRI) with T1- and T2-weighted imaging, and computed tomography (CT) angiography with three-dimensional reconstruction. The final diagnosis should be confirmed with angiography/phlebography to further define the lesion and plan appropriate treatment [2, 4, 5, 13, 15, 16].

Duplex ultrasonography allows hemodynamic assessment of the arterial and venous components involved with an AVM and VM. LMs often have discernible fluid-filled areas. Duplex ultrasound is extremely valuable for clinical follow-up and remains the initial study of choice for CVM evaluation [2, 4, 5].

MRI remains the major diagnostic study for the entire group of CVMs. MRI allows assessment of lesion extent, severity, and anatomic relationship with the surrounding tissues and organs [2, 4, 5]. MRI of an AVM, VM, or LM lesion is usually followed up with CT angiography as a confirmatory study.

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