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Review Article

Vascular anomalies of the head and neck: current overview

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Vascular anomalies are disorders of the endothelium and surrounding cells that can affect the vasculature and involve any anatomical structure. The most common problem associated with vascular anomalies is psychological distress related to disfigurement as well as functional defects, as many lesions affect the head and neck. This article provides an overview of the current clinical features that distinguish the major types of vascular anomalies that affect the head and neck.

Keywords: Vascular anomalies / Head / Neck

INTRODUCTION

Vascular anomalies are disorders of the endothelium and surrounding cells that can affect the vasculature and involve any anatomical structure. The estimated prevalence is 4.5%, and the anomalies are usually diagnosed during infancy or childhood [1]. The most common problem associated with vascular anomalies is psychological distress related to the disfigurement as well as functional defects because many lesions affect the head and neck. Vascular anomalies lead to local complications (including bleeding, infection, obstruction, pain, thrombosis, ulceration, and destroyed anatomic structures) and can also cause general complications such as congestive heart failure, disseminated intravascular coagulation, pulmonary embolism, thrombocytopenia, and sepsis [2].

Vascular anomalies were once limited owing to their nomenclature by the 19th century, but a schema proposed in 1982 has helped to clear the terminological confusion [3]. Accurate histopathological description is important for diagnosis and treatment of vascular anomalies, in correlation with clinical and radiological evaluation. Nevertheless, traditional nomenclature continues to be responsible for mistaken diagnosis, treatment, and research. The International Society for the Study of Vascular Anomalies (ISSVA) recognized this persistent problem and developed a classification system for vascular anomalies, derived in part from the system proposed by Mulliken and Glowacki [3], in which vascular anomalies were divided into tumors and malformations according to the presence or absence of endothelial mitotic activity. Accumulated evidence has shown that endothelial mitotic activity alone is not sufficient to classify vascular anomalies because mitotic activity stimulated as a secondary effect of ischemia and turbulence. The ISSVA classification has expanded over the years and is now widely adopted (Table 1) [4]. This article provides an overview of the current clinical features that distinguish the major types of vascular anomalies that affect the head and neck.

VASCULAR TUMORS

Vascular tumors of childhood are usually benign. The common types of vascular tumor are infantile hemangioma (IH), congenital hemangioma, kaposiform hemangioendothelioma, pyogenic granuloma, and others [5].

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Table 1. Overview of the ISSVA classification of vascular anomalies (approved at the 20th ISSVA Workshop, Melbourne, April 2014, last revision May 2018)

Vascular anomaly
Vascular tumor
Benign
• Infantile hemangioma
• Congenital hemangioma
Rapidly involuting (RICH)
Non-involuting (NICH)
Partially involuting (PICH)
• Tufted angioma
• Spindle-cell hemangioma
• Epithelioid hemangioma
• Pyogenic granuloma
• Others
Locally aggressive or borderline
• Kaposiform hemangioendothelioma
• Retiform hemangioendothelioma
• Papillary intralymphatic angioendothelioma (PILA), Dabska tumor
• Composite hemangioendothelioma
• Pseudomyogenic hemangioendothelioma
• Polymorphous hemangioendothelioma
• Hemangioendothelioma not otherwise specified
• Kaposi sarcoma
• Others
Malignant
• Angiosarcoma
• Epithelioid hemangioendothelioma
• Others
Vascular malformation
Simple
• Capillary malformations (CM)
• Lymphatic malformations (LM)
• Venous malformations (VM)
• Arteriovenous malformations (AVM)[a]
• Arteriovenous fistula (AVF)[a]
Combined[b]
• CVM (CM+VM), CLM (CM+LM)
• LVM (LM+VM), CLVM (CM+LM+VM)
• CAVM (CM+AVM)[a]
• CLAVM (CM+LM+AVM)[a]
• Others
Of major named vessels
• See details on ISSVA website (http://www.issva.org/classification)
Associated with other anomalies
• See list and known genetic associations on ISSVA website (http://www.issva.org/classification)

The International Society for the Study of Vascular Anomalies (ISSAVA) classification for vascular anomalies. The updated classification was approved at the May, 2018 General Assembly in Amsterdam, the Netherlands.
[a]High-flow lesions; [b]Defined as two or more vascular malformations found in one lesion.
Adapted from International Society for the Study of Vascular Anomalies [4].

Infantile hemangioma

IH is benign and the most common tumor in infancy [3]. IH affects approximately 4%–5% of Caucasian infants and is rare in other populations [6]. It is more frequent in premature (23% of infants < 1,200 g) and female (3:1 to 5:1) [7]. It usually occurs singly (80%) and involves the head and neck (60%), followed by the trunk (25%) and extremities (15%) [8]. Of all IH cases, 30%–50% are found at birth as a telangiectatic stain or ecchymotic lesion; however, the mean age at appearance is 2 weeks [9]. During the first 9 months after birth, IH grows faster than the growth rate of the child (proliferating phase). IH may appear red and involves the superficial dermis, whereas the overlying skin may appear bluish when IH is located beneath the skin. Until 9 to 12 months after birth, the growth of IH plateaus. After 12 months of age, IH begins to shrink, its color fades, and the lesion flattens (involuting phase). By 5 to 7 years of age, involution stops in most patients (involuted phase). Thereafter, local complications are observed in one-half of patients, including residual telangiectasias, loss of elastic fibers, scarring, redundant skin, or destroyed anatomic structures [10,11]. The immunostaining for an erythrocyte-type glucose transporter (GLUT1), which is specifically expressed in IH, can differentiate IH from other tumors and malformations [12]. Propranolol, a nonselective adrenergic blocker, is the first-line treatment of complicated IH. Most IH are managed by observation because 90% are small, localized, and involute without sequelae. But some patients may require operative treatment because of residual fibrofatty tissue, redundant skin, or damaged structures after involution [9].

VASCULAR MALFORMATIONS

Vascular malformations, as congenital abnormalities, result from abnormal vessel development and morphogenesis. In general, they are present at birth (but may be hidden in a deep location) and grow in proportion to the child's growth, persisting throughout the lifetime.

Capillary malformations

Capillary malformations (CMs), also known as port-wine stains or nevus flammeus, are the most common type of congenital vascular malformations [13]. These lesions are initially flat and bright pink, red, or violaceous and typically affect the face (90%), followed by the neck, trunk, leg, arm, and hand [14-16]. They often seem to lighten significantly over the first few months of life. This is not indicative of spontaneous resolution but is probably due to a decrease in circulating blood hemoglobin concentration [17]. In contrast to other similar birthmarks, most CMs become darker, thicker, and more nodular over time. This is particularly true of facial lesions [18]. The incidence rate is reported as 0.3% in newborns, with an equal sex distribution, occurring spontaneously within the population [6]. In most affected individuals, CMs occur as a sporadic unifocal lesion and

are not associated with any underlying abnormalities. However, CMs are sometimes associated with other underlying syndromes such as Sturge-Weber syndrome, macrocephaly-capillary malformation syndrome, capillary malformation-arteriovenous malformation syndrome, and overgrowth syndromes such as Klippel-Trenaunay syndrome [19,20]. The pathogenic mechanism of CM is still unknown. Shirley et al. [21] identified a somatic mutation in *GNAQ* with isolated CMs, disrupting vascular development. Facial CMs initially appear as a faint pink macule; however, some patients may develop soft tissue hypertrophy, bony hypertrophy, and/or nodule formation during adulthood. Depending on the size and location, these changes can cause functional deficits in vision, speaking, or eating, and significant psychological distress related to the resulting stigmatization or disfigurement. The gold standard therapy for facial or aesthetically sensitive CM is still the pulsed dye laser treatment. In patients with associated soft tissue or bony hypertrophy, surgical management can be helpful in restoring the normal anatomy and in re-establishing a symmetric contour [15,16,22-24].

Lymphatic malformations

Lymphatic malformation (LM) results from errors in the development of the lymphatic system; lymphatic tissue may form in an abnormal location [25]. LM is divided into three types according to the size of the malformed channels, namely, microcystic, macrocystic, or combined (microcystic/macrocystic) [3,8]. LM is a soft and compressible lesion that usually appears at birth; however, a small or deep lesion may not become evident until the lesion has grown large enough to cause deformity or symptoms. LM is most commonly located on the head and neck, causing a deformity and psychosocial morbidity. The overlying skin appears in various shapes, which may be normal, have a bluish hue, or have pink vesicles similar to CM. LM is problematic because of its progression, its slow expansion over time, and its recurrence. The common complications are bleeding and infection. Intralesional bleeding occurs in up to 35% of LMs, causing pain or swelling [26]. LM is vulnerable to infection because the malformed lymphatics contribute less to antibody production and protein-rich fluid provides a good environment for bacterial growth. Somatic *PIK3CA* mutations were found in several malformative or overgrowth syndromes, including LMs as a component. Small or asymptomatic lesions may be observed. Symptomatic lesions causing pain, deformity, or threatening vital structures necessitate operative treatment [27].

Venous malformations

Venous malformation (VM) results from errors in vascular morphogenesis. Thin-walled veins with abnormal smooth muscle are dilated, and then the VM expands and the flow stagnates with clotting. VM is present at birth but may not become evident until it has grown large enough to cause a deformity or symptoms. VMs are blue, soft, and compressible; sometimes phleboliths may be palpable. VMs may appear from localized skin lesions to diffuse malformations involving multiple tissue and structures [28]. VMs are sporadic and solitary in 90% of patients; 50% of patients have a somatic mutation in the endothelial receptor TIE2, which is involved in angiogenesis [29,30]. VMs are usually larger than 5 cm (56%) and involve the skin, mucosa, or subcutaneous tissue; 50% of VMs also involve muscle, bone, and viscera; appear singly (99%); and are located on the head/neck (47%), followed by the extremities (40%) and trunk (13%) [31]. Complications related to VMs depend on the extent and location and cause psychosocial morbidity because of their appearance. Common complications are ulceration, bleeding, compression of adjacent structures, and chronic low-grade consumptive coagulopathy in large and extensive lesions. Pain and swelling are dependent on position or are secondary to thrombosis and phlebolith formation. In the head and neck, VMs may severely affect compression of adjacent structures. Although nonproblematic lesions can be observed, symptomatic lesions causing pain, deformity, or threatening vital structures necessitate sclerotherapy or operative treatment [28].

Arteriovenous malformations

Arteriovenous malformation (AVM) results from errors in vascular development during embryogenesis; absent capillary beds lead to shunting directly from the arterial to venous circulation through a fistula or nidus (abnormal channels between feeding arteries and draining veins) [32]. The most common site of extracranial AVM is the head and neck, followed by the limbs, trunk, and viscera [9]. AVM is present at birth but may not become evident until childhood. AVM has a pink-red cutaneous stain with a palpable thrill or bruit, and it is important to distinguish AVM from a CM or hemangioma. Arteriovenous shunting reduces capillary oxygen delivery, causing ischemia. Common complications are pain, ulceration, bleeding, and congestive heart failure. AVM can cause disfigurement, compression, or destruction of adjacent tissues. Although AVM is considered a quiescent lesion, angiogenesis (growth of new blood vessels from preexisting vasculature) and/or vasculogenesis (de novo formation of new vasculature) may be involved in AVM expansion [33]. AVM is not a static malformation, progresses over time, and recurs. Genetic abnormalities cause certain types of familial AVMs. Capillary malformation-arteriovenous malformation (CM-AVM) results from a mutation in *RASA1* [34].

The goal of treatment usually is to control AVM. For superficial AVMs, patients should prevent desiccation and subsequent ulceration, and compression garments for extremity lesions may reduce pain and swelling. Intervention including embolization, resection, or a combination is focused on reducing symptoms, preserving vital functions, and improving deformities [3,35].

CONCLUSION

The number of physicians interested in the treatment of vascular anomalies has grown over the last several decades. Hundreds of articles about vascular anomalies are published every year, and criteria for classification, diagnosis, and treatment are also changing rapidly. The common vascular anomalies of the head and neck region are summarized, and the authors hope that this article will be helpful in the research and treatment of patients.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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