Vascular Malformations in Children: An Update

Jamshed Akhtar [1*]

Vascular malformations are congenital lesions and distinct from vascular tumors. They are composed of abnormally formed channels lined by endothelial cells.[1] Their growth pattern is different from that of vascular tumors and both are types of vascular anomalies. Historically, there was controversy about the nomenclature used for these anomalies. However, the classification system proposed by the International Society for the Study of Vascular Anomalies (ISSVA), which is the most widely accepted system, tried to address these issues. According to it the vascular anomalies are categorized as vascular tumors and vascular malformations.[2] The original classification of ISSVA of 1996 was been revised in 2014 and later in 2018 keeping abreast with the new knowledge gathered over the decades.[3,4]

There are four main groups of vascular malformations namely: simple, combined, anomalies of major vessels, and those associated with other vascular anomalies. The commonly used term infantile hemangioma is a type of vascular tumor and must not be confused with the vascular malformations. This is subdivided into rapidly involuting (RICH), non-involutong (NICH) and partially involuting hemangiomas types.[5] Vascular malformations on the other hand increase in size with the age of the patients. They are subdivided into capillary, venous, lymphatic, arterio-venous and combined malformations, depending upon which component predominates. The most common malformations are venous in origin and constitutes nearly 70% of the total followed by lymphatic malformations, arterio-venous malformations, and combined malformation syndromes. The capillary malformations are rare of all the lesions.[6]

Vascular malformations have varied clinical presentations. Based upon the predominant type of the channel involved and nature of the blood flow, they can be sub-classified as slow-flow and fast-flow malformations. The symptoms and signs therefore varies. They may be associated with different syndromes like Klippel-Trénaunay syndrome (slow-flow type), Sturge-Weber syndrome (slow-flow type), Bonnet-Dechaume-Blanc syndrome (fast-flow type) and many others. These malformations may present with number of complications some of which may be life threatening.[7] The diagnosis is usually clinical and supported by investigations like color Doppler ultrasound and MRI. Treatment is planned based upon the nature, anatomical site, age of the patients, and clinical presentation. This include sclerotherapy, surgery, endovascular intervention (embolization), use of lasers and others.[8]

New terminology is also used for lymphatic malformations in ISSVA classification. These are low flow lesions. They drain the third space fluid that leaks out of capillaries, into venous system. Based upon their clinical presentations these are divided into microcystic, macrocystic, or combined (microcystic/macrocystic) types.[9] They are benign in nature and are mostly noted at birth. They may manifest early in infancy or later in childhood. These malformations are composed of cysts of varying sizes. They are commonly located in neck, axilla and groin as these anatomical sites are rich in lymphatics. They are also found in retroperitoneum, thorax and extremities. Diagnosis for superficial lesions is usually clinical though ultrasound and magnetic resonance imaging help in delineating extent and the nature of the cysts. The new research on the genesis of these lesions and growth helped in finding new pharmacological interventions for the treatment. Traditionally these were subjected surgical excision or sclerotherapy. The new drugs used for these malformations include rapamycin (like sirolimus) that inhibit lymphatic vessels overgrowth. It was initially used on off-label basis for complex lesions and showed promising results.[10] However, the complex lymphatic malformations are still a challenge and multidisciplinary approaches are recommended to deal with them.[11] Other modalities of treatment, in selected cases like those with mixed lesions and syndromic presentations,

[1] Department of Paediatric Surgery, National Institute of Child Health Karachi

Correspondence:
Dr. Jamshed Akhtar [1*]
Visiting Faculty
Department of Pediatric Surgery
National Institute of Child Health Karachi
E mail: jamjim88@yahoo.com

include drug like sildenafil and ablation of the lesions with laser.[12]

The International Society for the Study of Vascular Anomalies classification helps in understanding and reporting various lesions. However, the currently available classification is available in PDF format which is not machine readable. We are living in an era of artificial intelligence. The use of artificial intelligence in this context may facilitate specialists in identifying and diagnosing theses vascular anomalies. In a study authors describe a process by which the ISSVA classification is transformed into an ontology. They involved ontology experts and clinician to work on it. They believe that the ISSVA ontology may contribute to data collection for vascular anomaly research. They used an acronym; Findable, Accessible, Interoperable, and Reusable (FAIR) for this purpose. The ontology is made available athttps://bioportal.bioontology.org/ontologies/ISSVA.[13]

In conclusion the vascular malformations pose multiple challenges for the treating physicians. These are also the subject of research to explore hidden embryological events for their development and growth. This can further enlighten us about their clinical behavior and development of new treatment options

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