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EDITORIAL

Vascular Anomalies

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Vascular anomalies are complex diseases with different clinical presentations. In ancient times, some purplish spots were already seen as a curse, and over the centuries, they began to be described as part of other diseases, gaining different eponyms. In recent decades, the term "hemangioma" has been missused to describe a large part of vascular anomalies, causing a significant problem in diagnosing and treating patients.

European and American groups began dedicating themselves to the study of anomalies, and Mulliken and Glowacki proposed in 1982 a classification based on histopathological studies and clinical manifestations. Thus, a first draft of the classification was developed[1], which is now adopted by the International Society for the Study of Vascular Anomalies (ISSVA), also created by the initial group.

The ISSVA classification divides the anomalies into two major groups, vascular tumors and malformations. Hemangiomas are the most typical representative of the tumor group, which also includes hemangioendotheliomas and angiosarcomas. The term "oma", which refers to a tumor, started to be used exclusively for this group. Alternatively, vascular malformations were subdivided according to the type of vessel affected and may be capillary, lymphatic, venous, or arterial, combined or associated with malformations of other structures[2]. Within this group are the Klippel Trenaunay, Sturge Weber and Parkes Weber syndromes.

Patients' stratification and correct diagnosis have allowed exponential treatment advancement in the last two decades.I nfantile hemangiomas, the most frequent benign tumors of childhood, have accelerated growth in the first 2 years of life. They were previously treated with watchful waiting, steroids, and interferon with limited effectiveness and many sequelae. The use of b-blockers was studied after observing the regression of a giant hemangioma in a hypertensive child[3] and is currently the first treatment option, associated with other modalities such as laser therapy or surgery.

Vascular malformations, caused mainly by random somatic mutations during the embryonic period, can lead to different phenotypes such as small lymphatic cysts, venous lakes, or even malformations of the main vessels such as agenesis of veins. Symptoms can be present from birth or appear after stimuli such as local trauma, puberty, pregnancy, infection, or surgery. Many patients underwent unsuccessful surgical treatment, such as ligating arteries that nourish the malformations, with many sequelae and worsening symptoms. The treatment evolved less invasively, with the indication of sclerosing agents or embolization of the anomalous structures to occlude the lesions without harming other structures. Additionally, adjunctive treatments such as laser therapy for the superficial components, thermoablation of malformed truncal veins, and surgeries for partial or complete resection of some

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lesions have also been used successfully.

In the last decade, a new frontier has opened for the comprehension of vascular anomalies with the study of the genetic mutations present in each disease. Some vascular malformations share the previously identified mutations in tumors and may confer adaptive advantages to cells that contain these mutations. Unlike germ cell mutations, somatic mutations are known as "mosaic" as only mutated cells will develop the phenotype.

The great importance of genetic studies in patients with malformations is the possibility of using targeted therapy with medications that regulate cell signaling pathways compromised by mutations[4,5]. Simply put, these drugs can control systemic and local effects of malformations such as localized intravascular coagulation, recurrent lymphangitis, bone lymphatic disease, and even reduce the volume of lesions.

Sirolimus, an inhibitor of the mTOR receptor (part of the PIK3CA-mTOR AKT cell signaling pathway), was the first drug used as targeted therapy, already studied in clinical trials, with good results[6,7]. Other drugs, some already used for some types of cancer, are being tested with variable results, such as trametinib (MEK inhibitor, ras-mapk-MEK signaling pathway) and alpelisib (PIK3alfa unit blocker)[8].

Despite being a rare disease affecting 0.5%-3%[9] of the population, vascular anomalies can cause significant harm to patients' quality of life, especially if they are not correctly diagnosed and treated. The first step is to get to know this large group of diseases and, whenever possible or necessary, refer patients for follow-up in specialized multidisciplinary groups.

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