ARTERIOVENOUS MALFORMATIONS

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Summary

Cutaneous arteriovenous malformations (AVM) are post-flow malformations. They consist of a connection between arteries and veins. The can be congenital or acquired.

The lesions may be small to large and aggressive. There are many particular clinical forms

- *Stewart-Fluefarb*
- *Bonnet-Blanc Deschaumes (or Wyrburn – Mason)*
- *Capillary malformations –AVM syndrome with RASA-1 mutation*
- *Parkes-Weber syndrome*
- *Congenital lipomatous overgrowth vascular malformations and epidermal nevi (CLOVE) syndrome*
- *Cobb syndrome*

AVM can be complicated by pain, ulceration, haemorrhage and necrosis. Evolution has 4 stages (Schobringer classification).

Diagnosis is based particulary on two exams, doppler ultrasonography and MRI.

Treatment for voluminous forms used embolization, surgery, and new medicals treatments with some anti VEGF drugs.

Key words: *Angioma, malformations, arteriovenous malformations.*

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Rezumat

Malformațiile arteriovenoase cutanate (MAV) sunt malformații ale circulației de întoarcere. Ele constau în conexiuni între artere și vene. Acestea pot fi congenitale sau dobândite. Leziunile pot fi de mici dimensiuni sau extinse și agresive.Ele prezintă multe forme clinice particulare

- *Stewart-Fluefarb*
- *Bonnet-Blanc Deschaumes (sau Wyrburn – Mason)*
- *Malformații capilare –sindrom MAV cu mutația RASA-1*
- *Sindrom Parkes-Weber*
- *Sindrom CLOVE – malfromații vasculare congenitale lipomatoase și nevi epidermali*
- *Sindrom Cobb, MAV pot fi complicate de durere, ulcerații, hemoragii și necroză. Evoluția are 4 stadii. (Clasificare Schrobringer). Diagnosticul se bazează în special pe două examinări, ultrasonografie Doppler și IRM.*

Tratamentul pentru formele cu volum mare utilizează embolizarea, chirurgia și noile tratamente medicale cu medicamente anti VEGF.

Cuvinte cheie: *angiom, malformații, malformații arteriovenoase.*

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Cutaneous arteriovenous malformations (AVMs) are fast-flow malformations as opposed to low-flow venous, capillary and lymphatic malformations. They consist of a connection between arteries and veins without any interposal capillary bed, thus creating a „nidus" or shunt. They can be congenital or acquired. If acquired, they often occur after a trauma. If congenital, they are usually latent for many years and may be exacerbated at puberty or after a trauma or therapeutic procedure, even if minimal.

From a clinical viewpoint, the lesions may be small, such as located on a finger (1) or the nose. They involve a superficial telangiectatic vascular network and sometimes minimal swelling. The

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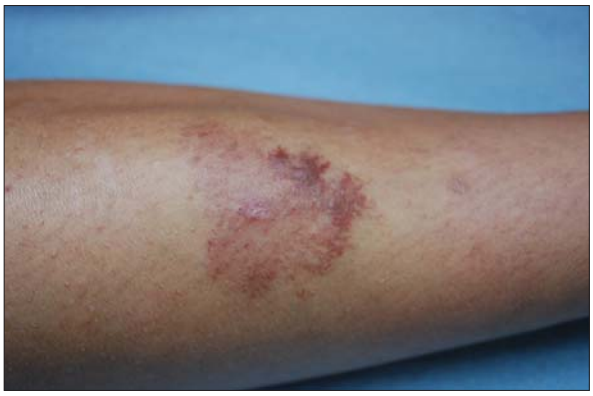

Fig. 1. Superfical arteriovenous malformation of leg

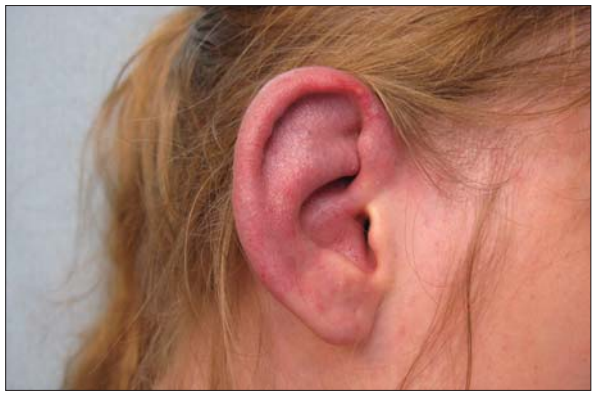

Fig. 2. Cutaneous arteriovenous malformation of ear

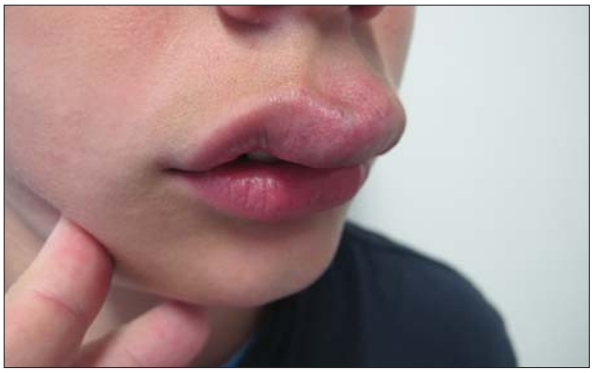

Fig. 3. Arteriovenous malformations of the upper lip

lesion may be located on a limb (fig 1) or on the face (fig 2-3) with various spreading. Sometimes the only features are superficial capillary malformations. The local temperature may be increased. Palpation can reveal a pulse. In contrast, large defects can affect any part of the face or a limb, with large swelling, superficial venous network of large diameter, and increased local heat. This lesion may be stable or may be the starting point of a large aggressive malformation. The evolution from this growth phase can be particularly aggressive, with haemorrhage, ulceration, and necrosis.

Clinical forms of AVMs

- Stewart-Bluefarbsyndrome is a purplish swelling tumor mimicking Kaposi's sarcoma. The lesions may become necrotic and infected.

- Bonnet-Blanc-Dechaume or Wyburn-Mason syndrome stain consists of a forehead capillary malformation associated with an underlying AVM of the brain.

- Capillary malformation-AVM syndrome (or arteriovenous fistula) (2) combines multiple capillary malformations, rounded or oval, often surrounded by a lighter halo of vasoconstriction (3) (4). In other family members, it can be observed as hypertrophy of a limb (5) with extended capillary malformation and multiple arteriovenous fistulas indicating Parkes-Weber syndrome (6) (7). It can also be revealed as an AVM of the spinal cord (8). These cases exhibit mutation of the gene RASA-1 (9) (3) (6).

- Congenital Lipomatous Overgrowth Vascular malformations and Epidermal nevi (CLOVE) syndrome. The most stricking features are vascular malformations, lipomas, scoliosis, and enlarged bony structures. Spinal malformation of the arterio-venous type may be responsiblefor neurological injury (10) (11) (12).

- Cobb syndrome (13) involves a segmental vascular malformationof the skin. This malformation may be arterio-venous or capillary. The same part of the trunkmay show a spinal cord AVM. The spinal cord lesions can cause neurological disorders and paraplegia. The lesions are located in the same territory: arteries and veins, muscles and bones. This syndrome is also called Signal arteriovenous Venous Syndrome Metameric 1-31).

Evolution

AVM lesions can increase in volume. They can be complicated by pain, ulceration, haemorrhage, and necrosis. Heart failure is possible. AVM may rapidly progress during puberty or pregnancy. Trauma and infections are also factors of progression. Evolution shas 4 stages (Schobinger

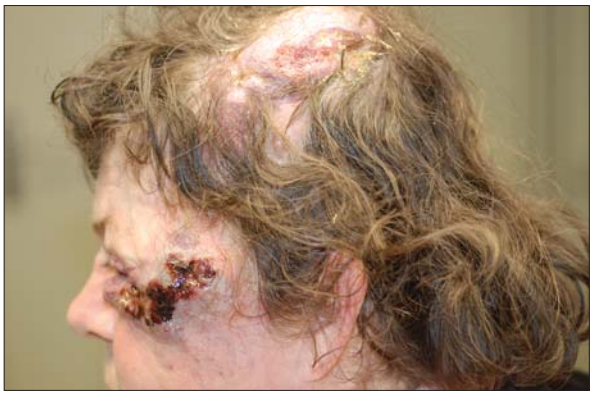

Fig. 4. Giant arteriovenous malformation of face and scalp

classification): stage 1, quies-cence with macular lesions; stage 2, expansion, enlarging lesions, palpable pulse; stage 3, infiltration;stage 4, destruction, necrosis, heart failure (fig 4).

Pathophysiology

AVMs are due to failure of complete involution of the fetal capillary bed. This situation leads to connections between arteries and veins. Most AVMs do not have a hereditary family character of transmission. The discovery of the mutation in RASA-1 has introduced an element of complexity with the realization of familial forms, but clinical manifestations are varied in the same family (9) (6) (5) (4).

In contrast, somatic mutations are sometimes identified on biopsy, in particular mutation in the PI3K gene (14) in the same pathway as the Proteus syndrome gene (AKT) (14). Somatic mutations play an important role in many AVMs. DNA libraries of tissues and circulating blood samples must be collected for future investigations of these malformations.

Diagnosis

The diagnosis is made on clinical examination: clinical features, location, and stage. Some areas such as the ear are more at risk. Local heat, which increasesthe hypertrophy of the auricle (15), palpation of pulse or thrill require exploration. Doppler ultrasonography allows for visualizingincreased arteriovenousflow. MRI can visualize the enlarged superficial tissues, important for visualizing depth. Gadolinium injection allows for better visualization of vessels. MRI angiography can reveal the nidus squads that are vascular arteries and veins corresponding to extension zone angiomas.

Treatment

For quiescent, minor forms of AVMs, monitoring only is the rule; any action may lead to exacerbation, so the treatment should be conservative. Embolization can be performed in case of progression or complications.

For voluminous forms, the definitive treatment, when possible, is complete surgical wide excision of the nidus. This surgery may be difficult or impossible because of the location or volume of the AVM. It sometimes requires rapid intervention for heavy bleeding or rapid extension (16). Amputation may be necessary. Embolization is useful immediately preoper-atively to improve intraoperative hemostasis. Embolization or ligation of vessels without subsequent surgery are of little use for large AVMs, and often a reversal in condition is fast and exacerbation may even succeed embolization.

Medical treatment

The aim is to decrease the action of growth factors (vascular endothelial growth factor [VEGF]). The immunosuppressant sirolimus can be tried as can some anti-VEGF drugs with or without embolization or surgery. Clinical trials are underway.

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Conflict of interest
NONE DECLARED

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