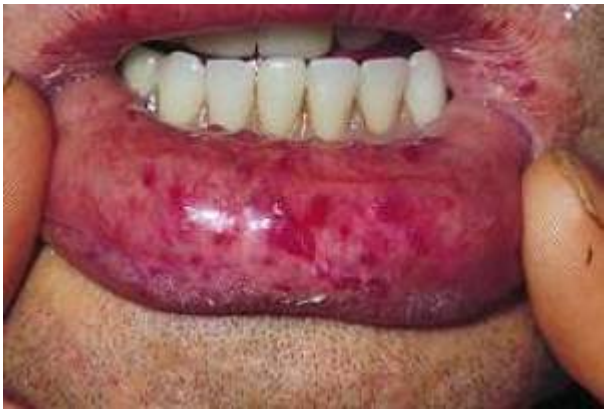
Osler-Weber-Rendu syndrome



Capillary telangiectasias involve small, dilated capillaries that may not be connected to larger vessels. They are usually considered benign and may not cause symptoms. However, they can sometimes be associated with other vascular malformations.

The disease carries the names of Sir William Osler, Henri Jules Louis Marie Rendu, and Frederick Parkes Weber, who described it in the late 19th and early 20th centuries.

General information

AKA hereditary hemorrhagic telangiectasia (HHT), AKA capillary telangiectasia: slightly enlarged capillaries with low flow. It cannot be imaged in any radiographic study. Usually incidentally found at necropsy without clinical significance (risk of hemorrhage is very low, except possibly in brain stem). Has intervening neural tissue [1] (unlike cavernous malformations). Usually solitary, but maybe multiple when seen as a part of a syndrome: Osler-Weber-Rendu, Louis -Barr (ataxia telangiectasia), Wyburn-Mason Syndrome, Sturge-Weber syndrome. Associated cerebrovascular malformations (CVM) include telangiectasias, AVMs (the most common CVM, seen in 5–13% of HHT patients [2]), venous angiomas and aneurysms. Patients are also prone to pulmonary arteriovenous fistulas with an associated risk of a cerebral paradoxical embolism which predisposes to embolic stroke and cerebral abscess formation

Epidemiology

Rare autosomal dominant genetic disorder of blood vessels affecting ≈ 1 in 5,000 people. 95% have recurrent epistaxis.

Clinical features

It is an autosomal dominant genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes, and often in organs such as the lungs, liver, and brain.

The occurrence of concomitant multiple cerebral arteriovenous malformations (mAVMs) is often

associated with hereditary hemorrhagic telangiectasia (HHT) or craniofacial arteriovenous metameric syndrome (CAMS) and frequently occurs in the pediatric population.

It may lead to nosebleeds, acute and chronic digestive tract bleeding, and various problems due to the involvement of other organs.

Diagnosis

Imaging CT

May show a well-demarcated homogeneous or mottled high density [3] (high density due to hematoma, calcification, thrombosis, hemosiderin deposition, alterations in BBB, and/or increased blood volume [4] with some form of contrast enhancement (around or within lesion) in 17 of 24 patients [5]. Surrounding edema or mass effect is rare (except in cases that have recently hemorrhaged).

MRI

May demonstrate previous hemorrhage(s), [6] (may be important when the presence of multiple occurrences affects therapeutic choices). T2 weighted image finding: a reticulated core of increased and decreased intensity, a prominent surrounding rim of reduced-intensity may be present (due to hemosiderin-laden macrophages from previous hemorrhages). GRASS image demonstrates flow-related enhancement in ≈ 60%of cases, which allows signal dropout from flowing blood on other sequences to be differentiated from that due to calcium (and thus, bone) or air (limitations: hemosiderin causes signal dropout, and slow in-plane flow does not enhance) [7].

Complications

Patients diagnosed with hereditary hemorrhagic telangiectasia (HHT) are at risk of developing intracranial arteriovenous malformations (AVM).

HHT-related AVMs are smaller in size with lower Spetzler-Martin AVM grading system and less temporal lobe involvement than sporadic AVMs. Patients with HHT frequently present with multiple intracranial AVMs. Conservative management is generally recommended due to lesion multiplicity and relatively low hemorrhagic risk [8]

Vascular malformations in the lungs may cause a number of problems. The lungs normally “filter out” bacteria and blood clots from the bloodstream; AVMs bypass the capillary network of the lungs and allow these to migrate to the brain, where bacteria may cause a brain abscess in up to 5 % and blood clots may lead to stroke.

Treatment

Osler-Weber-Rendu syndrome treatment.

References

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