Osler-Weber-Rendu Syndrome

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Osler-Weber-Rendu syndrome, also known as hereditary hemorrhagic telangiectasia (HHT), is an autosomal dominant disorder; however, in about 20% of the cases there is no family history. The syndrome is characterized by abnormal blood vessel formation in the skin, the mucous membranes and often within various organs such as the lungs, liver and brain. The incidence of the disorder in the general population reaches 1–2/100,000 and the race and gender distribution is homogeneous [1]. Otorhinolaryngologic manifestations are the most frequent, with recurrent epistaxis being the main complaint. Blood vessels in other organs may also be involved in the aforementioned various organs as well as the gastrointestinal tract [2,3].

A recent study hypothesized that arterial aneurysms may occur in HHT as the result of a weakened arterial wall due to impaired transforming growth factor-beta (TGFβ) signaling. In clinical practice patients should be screened for occurrence of arterial aneurysms, particularly patients with known hepatic vascular disease [5].

Telangiectasia may be identified by visual inspection during physical examination of the skin and oral cavity or by endoscopy. Diagnosis is made after clinical examination and genetic testing based on the Curacao criteria: telangiectasia in the face, hands or oral cavity; recurrent epistaxis; arteriovenous malformations with visceral involvement; and a positive family history. Diagnosis is confirmed upon the presence of at least three of these manifestations [1].

Recently, a validated questionnaire known as the HHT Epistaxis Severity Score (ESS) was developed. Although not much is known about the relationship between epistaxis and quality of life, a recent study established that the ESS is a major determinant of health-related quality of life and should be considered as a measurement of treatment efficacy in HHT-related epistaxis [4].

The patient in the photograph is a 52 year old woman, already diagnosed with HHT, who presented to our department due to abdominal pain and nasopharyngeal bleeding. The telangiectases on the tongue [Figure 1], lip [Figure 2] and fingers [Figure 3] are characteristic of Osler-Weber-Rendu syndrome.

Visceral involvement was also recorded in this patient as demonstrated by the computed tomography scan exhibiting numerous arteriovenous malformations within the liver parenchyma [Figure 4].

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