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CASE REPORT

Sixty-four year old male reports lesions in hands and oral cavity for nearly 40 years, accompanied by recurring epistaxis that became progressively more severe, culminating in hospital admission during the last episode. He denies gastrointestinal bleeding and refers that his father had a similar case. Dermatological examination: telangiectasia in fingertips, tongue, lips, perioral and nasal regions (Figures 1 and 2). Anatomopathological exam of fingertip skin showed histological skin sections with epidermal acanthosis and a thick layer of compact hyperorthokeratosis, numerous vessels with tortuous and ectatic lumen were noted in the dermis, sometimes touching and compressing the basal layer of the epidermis. These vessels are lined by endothelial cells without atypias and have variable calibers (Figure 3). Colonoscopy showed vascular ectasia in the sigmoid. Magnetic resonance angiography of brain, chest and abdomen, and upper gastrointestinal endoscopy showed no vascular alterations.

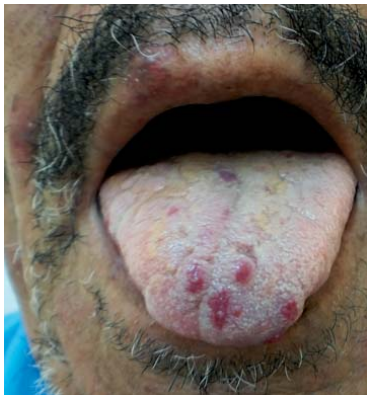

FIGURE 1: Erythematous-violaceous papules with teleangiectasias in tongue

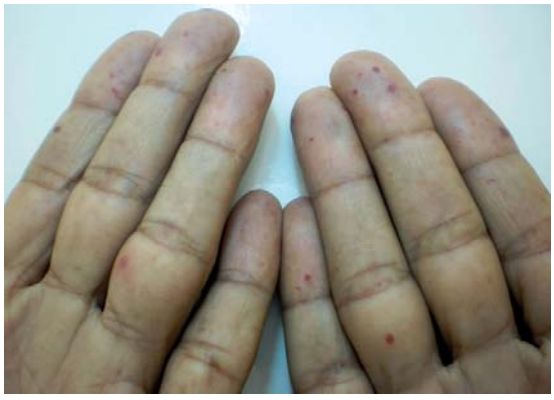

FIGURE 2: Teleangiectasias in fingertips and palms

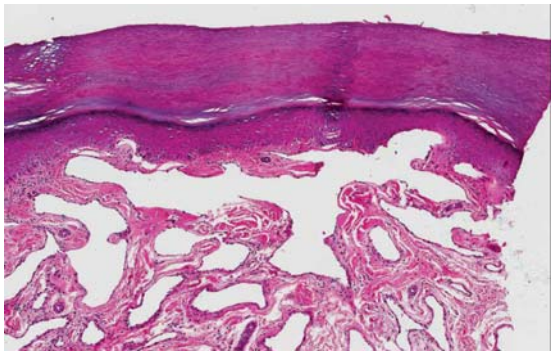

FIGURE 3: Anatomopathological exam of fingertip skin lesion: numerous vessels with tortuous and ectatic lumen were noted in the dermis, sometimes touching and compressing the basal layer of the epidermis. The vessels are lined by endothelial cells without atypias and have variable calibers

DISCUSSION

The patient reported was classified as having Rendu-Osler-Weber syndrome for presenting more than three of the four diagnostic criteria: spontaneous and recurrent epistaxis, multiple visible telangiec-

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tasias (lips, tongue, hands and nose); visceral lesion (gastrointestinal telangiectasias) and positive family history (first-degree relative as disease carrier).

Rendu-Osler-Weber syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT), is a rare systemic fibrovascular dysplasia that leaves vascular walls vulnerable to trauma and rupture, causing mucocutaneous bleeding.[1-3] It has a dominant autosomal inheritance, but in 20% of cases there is no positive familial history.[3] HHT can arise from mutations in at least five genes, but mutations in two genes (ENG and ACVRL1/ALK1) represent approximately 85% of cases.[4] The incidence is variable and affects both genders.[1-3]

Diagnosis is based on the presence of at least three of the following criteria: recurrent epistaxis, mucocutaneous telangiectasias; evidence of family history and visceral arteriovenous malformations.[1-4] Recurrent epistaxis is the first and main symptom and it is present in about 90% of cases. Patients with this syndrome can develop telangiectasias on skin, nasal and oral mucosae, gastrointestinal tract and also arteriovenous fistulae in lung, liver and brain.[3] Investigation of these alterations should be done through endoscopy, colonoscopy and MRI of head, thorax and abdomen.

Mortality rate increases in patients over 60 years old, however, the disease is characterized by the high morbidity among younger patients due to visceral involvement, particularly lungs and brain.[5] Treatment consists of supportive care and prevention of complications. Angiogenesis modulation therapy may be an effective alternative.[4] Bevacizumab (recombinant humanized monoclonal endothelial growth antibody - VEGF) has been used as a treatment option in these patients.[6,7] Several other treatment options such as propranolol, tranexamic acid, and other forms of laser ablation have been reported in the literature, with varying response rates.[5-8]

The Rendu-Osler-Weber syndrome is a rare disease that should be well understood by dermatologists, because the correct diagnosis of a patient at the start of symptoms and his early referral to a multidisciplinary team can avoid more severe disease complications. ❑

Abstract: Rendu-Osler-Weber Syndrome also known as Hereditary Hemorrhagic Telangiectasia is a rare systemic fibrovascular dysplasia, with dominant autosomal inheritance. It is characterized by recurrent epistaxis, mucocutaneous telangiectasia, visceral arteriovenous malformation and positive family history. There may be hematologic, neurologic, dermatologic and gastrointestinal complications. Therapy is supportive and aimed at preventing complications. In this article we report a case of Rendu-Osler-Weber in a 64 year-old man, with history of mucocutaneous telangiectasia since the third decade of life, recurrent epistaxis, positive family history and vascular ectasia in the gastrointestinal tract.

Keywords: Arteriovenous fistula; Epistaxis; Gastrointestinal hemorrhage; Skin abnormalities; Skin diseases, vascular

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