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.

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 telangiectasias in tongue, erythematous-violeaceous papules with telangiectasias in tongue, and vascular ectasia in the sigmoid.
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 telangiectasias; evidence of family history and visceral arteriovenous malformations. 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. 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. 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.

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. Treatment consists of supportive care and prevention of complications. Angiogenesis modulation therapy may be an effective alternative. Bevacizumab (recombinant humanized monoclonal endothelial growth antibody - VEGF) has been used as a treatment option in these patients. 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.

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.

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