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A case of lingual manifestation of Rendu-Osler Syndrome

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***Abstract*: The authors present a single case of lingual manifestation of Rendu-Osler Syndrome with haemorrhagic manifestations originating from multiple lingual angiomatous lesions in a patient with a family history of hereditary haemorrhagic telangiectasia.**

***Keywords*: Hereditary hemorrhagic telangiectasia, Rendu-Osler, tongue, hemorrhage.**

1. INTRODUCTION

Rendu-Osler syndrome, also known as hereditary hemorragic teleangiectasia (HHT) is an autosomal dominant disorder characterized by appearance of multiple mucocutaneous teleangiectasias, that is small artero-venous malformations whit frequent bleeding from nose, stomach and other places. It is possible the manifestation in many organs such as lung, brain and liver. There are two many types of HHT.HHT1 involves a mutation in endoglin and is at risk of pulmonary and cerebral artero-venous malformations. HHT2 involves a mutation ofactivin –A receptor-like type 1 and is at risk o liver artero-venous malformation.These mutations determine an aberrant development of blood vessel and artero-venous malformations. Normallt epistaxis is the most common manifestation of HHT. In the course of life, normally after puberty teleangiectasias appear in face, lips, tongue, fingers and palms.

2. PATIENTS AND METHODS

A 40-year-old male patient with a family history of epistaxis, postoperative haemorrhage after surgery for nasal polyposis (father), labial angiomatous lesions (sister) came to our attention. The patient arrived for the first time in an emergency due to a lingual haemorrhage coming from an arteriovenous lesion present on the lingual back; a suture was needed to stop the bleeding that had lasted relentlessly for over an hour.

The patient's tongue showed many angiomatous-looking lesions of which three (on the lingual back, on the right lingual margin and on the left osteral area) were larger than 6 millimeters and bluish in color, while many others were no larger than 2 millimeters and bright red (Fig. 1)

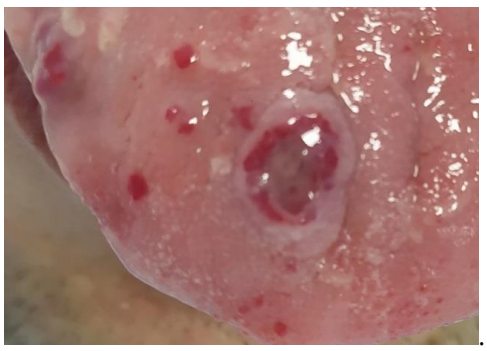

Fig 1: Lingual artero-venous malformations in the patient

The patient presented small capillary varicosities at the level of the nasal septum and some small arteriovenous malformations at the labial level, but the most important manifestation was undoubtedly the lingual lesions.

First a series of radiological investigations were prescribed to look for the presence of cerebral, pulmonary or hepatic localizations of the disease.

Once these were excluded, the surgical removal of the largest arteriovenous lesion was planned which, causing frequent bleeding, made it difficult to feed the patient who for some time had been forced to a liquid and cold diet. The removal took place without particular complications and the patient was discharged in the afternoon of the same day of the operation.

3. RESULTS

The patient is currently being followed up to monitor the growth of small lingual lesions. A coagulation of these with DIODE LASER could be a valid alternative to surgical removal if there is evidence of the growth of the lesions that currently do not bother the patient as they do not cause hemorrhagic manifestations.

4. DISCUSSION

Small lingual manifestations in HHT are quite frequent, but generally they are not so marked and lingual bleeding is almost never the first reason for consulting the specialist. We considered it appropriate to report this clinical case precisely because of the preponderance of lingial bleeding symptoms in the absence of other clinical manifestations.

The clinical diagnosis of HHT is based on some criteria which are recurrent epistaxis, mucocutaneous telangiectasias, visceral telangiectasias and the presence of HHT in first degree relatives. The presence of three or more criteria makes it possible to diagnose a certain HHT, while with 1 or 2 criteria we speak of suspected HHT.

Screening of cerebral arteriovenous lesions by brain MRI, hepatic by ultrasound and MRI, pulmonary by CT, gastroesophageal by gastroscopy is very important as these lesions, if present, can cause life-threatening complications .

In doubtful or not clinically defined cases, especially when there is a family history of HHT, a genetic investigation can be used to dispel doubts.

Periodic evaluation of the blood count to detect bleeding that is sometimes not clinically evident is advisable. If followed carefully to prevent possible complications, patients with HHT can have an almost normal life expectancy. For this reason it is important to clinically and genetically identify the carriers of this genetic disorder and subject them to the necessary controls and treatments.

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