OPHTHALMOLOGIC DIAGNOSIS OF HEREDITARY HEMORRHAGIC TELANGIECTASIA OR RENDU-OSLER-WEBER DISEASE

DIAGNÓSTICO OFTALMOLÓGICO DE UN CASO DE TELANGIECTASIA HEMORRÁGICA HEREDITARIA O ENFERMEDAD DE RENDU-OSLER-WEBER

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ABSTRACT

Case report: Our patient was a 45-year-old woman who had recurrent episodes of hematic epiphora, repeated epistaxes for which no cause was found and a family history of gastric hemorrhage. One of her daughters also suffered from spontaneous hemorrhages.

Discussion: Hereditary hemorrhagic telangiectasia is rarely diagnosed by an ophthalmologist; however the occurrence of bloody tears occurring spontaneously in a patient with epistaxis or gastric hemorrhage should lead to suspicion of hereditary hemorrhagic telangiectasia or Rendu-Osler-Weber disease. It should never be forgotten that clinical examination and appropriate investigations are basic components of disease diagnosis (Arch Soc Esp Oftalmol 2008; 83: 381-384).

Key words: Hereditary hemorrhagic telangiectasia, Rendu-Osler-Weber disease, hemorrhage, hematic epiphora.

RESUMEN

Caso clínico: Mujer de 45 años con episodios recurrentes de epífora hemática espontánea. La paciente presentaba epistaxis de repetición no filiadas y, además, una de sus hijas sufrió de epistaxis.

Discusión: El diagnóstico de telangiectasia hemorrágica hereditaria por oftalmólogos es un hecho infrecuente. La presencia de lágrimas con sangre o epífora hemática espontánea, en un paciente con historia de hemorragias de repetición en forma de epistaxis o hemorragia digestiva, nos tiene que hacer sospechar y buscar la presencia de esta enfermedad. La historia y exploración clínica son fundamentales en el diagnóstico de dicha enfermedad.

Palabras clave: Telangiectasia hemorrágica hereditaria, enfermedad de Rendu-Osler-Weber, hemorragia, epífora hemática.
INTRODUCTION

Hereditary hemorrhagic telangiectasia (HHT) or Rendu-Osler-Weber disease is a hereditary autosomic dominant disease characterized by abnormalities in one or two genes (9q33-34 and 12q13 chromosomes) involved in repairing the vascular wall. Telangiectasias are vascular dilations that spontaneously bleed from the smallest trauma.

There are different types of telangiectasias according to morphology: linear and spider (Table 1). The location of the telangiectasias can guide us toward finding the underlying pathology. Thus, periungual telangiectasias are pathognomonic of systemic lupus erythematosus, scleroderma and dermatomyositis.

Telangiectasias may appear anywhere on the skin and mucous membranes and are frequently seen on the lips, tongue, face and the palm of the hand. They first appear during childhood and grow larger during adolescence and adulthood. There are sometimes also small vascular dilations in the digestive tract, genitals and urinary apparatus and central nervous system, which may erode and start bleeding.

HHT is a particular form of telangiectasia due to its distribution and genetic pattern. These vascular lesions typically appear in the nasal, oral and conjunctival mucosa, on the face and tips of the extremities, such as the fingernail beds. The clinical signs and symptoms are determined by haemorrhagic episodes such as epistaxes and gastric bleeding. Morphologically speaking telangiectasias are radial and make up authentic arteriovenous malformations in the mucous membranes.

CASE REPORT

A 45-year-old patient comes in for recurring episodes of «eye bleeding», and she mentions that she «cries blood». The episodes spontaneously remit without any sort of treatment.

The patient’s medical history reveals the presence of repeated epistaxes with no ear, nose and throat tests. There is no other pertinent personal systemic or ophthalmologic history. When delving deeper into the patient’s history, she mentioned that her father suffered from repeated upper digestive bleeding. He died from this due to a massive haemorrhage and hypovolaemic shock. The patient also has two daughters that have repeated epistaxes.

The biomicroscopic exam of the subtarsal conjunctiva, both superior and inferior, shows the existence of isolated telangiectatic lesions (Figures 1-3). One of the telangiectasias showed signs of recent coagulation in connection with an associated haemorrhagic event (fig. 4). The fundoscopy did not show alterations in the vascular tree, with no alterations in the arterial or venous vessel calibre.

Table I. Classification of telangiectasias

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<tr>
<th>Primary or congenital</th>
<th>Secondary or acquired</th>
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<tr>
<td>General essential telangiectasias</td>
<td>Post traumatic</td>
</tr>
<tr>
<td>Hemorrhagic congenital Rendu-Osler-Weber telangiectasias</td>
<td>Due to physical agents: sunlight, radiations and cold (Raynaud syndrome)</td>
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<tr>
<td>Louis-Bar athaxia telangiectasia</td>
<td>Drugs: topical fluorinated corticoids and oral contraceptives</td>
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<tr>
<td>Congenital telangiectasic erithema or Bloom syndrome</td>
<td>Systemic diseases: dermatomiositis, systemic erythematous</td>
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<tr>
<td>Vascular spiders</td>
<td>lupus, scleroderma, CREST syndrome, carcinoid syndrome, systemic mastocitosis (eruptive macular telangiectasia)</td>
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<td></td>
<td>Pregnancy</td>
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<td>Liver cirrhosis</td>
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<td>Varicose veins and post-flebitic syndrome</td>
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haemorrhaging phenomena, exudates or associated neovascularizations.

The general examination of the patient showed telangiectasias in oral and nasal mucosa, aside from the telangiectatic lesions on the tongue and inside the upper and lower lips (figs. 5-7).

Referral to the Otorhinolaryngology and Internal Medicine Departments confirmed the presence of telangiectasias in nasal and pharyngeal mucosa, with none in the digestive mucosa. With all this information, a diagnosis of Rendu-Osler-Weber disease was made.

DISCUSSION

HHT was described at the end of the 19th century in groups of relatives characterized by spontaneous episodes of epistaxes, gastric haemorrhage, hae-
moptysis or haematuria. Such phenomena were reported independently by three physicians: Rendu, Osler and Weber. It was Hanes (1) who, in 1909, established the diagnostic triad of hereditary haemorrhagic telangiectasia, i.e. family history, haemorrhages and telangiectasias.

Ocular involvement is not frequently seen (2) and rarely constitutes the foundation for this diagnosis (3), as in the case reported here. The ocular signs and symptoms can be placed into two groups. On one hand we have those found in the anterior segment, such as tears with blood, telangiectasias and post haemorrhagic granulomatous lesions on the conjunctiva, and on the other hand macular and choroidal retinal telangiectasias (4).

A suspected diagnosis can be reached by looking at the patient’s clinical situation. A full clinical work-up requires endoscopic study of the digestive mucosa and the ear, nose and throat. In our case, since the patient came to our department first, an examination of the oral and nasal cavities was performed and she was then referred to the Otorhinolaryngology and Internal Medicine Departments in order to complete the diagnosis and continue with the corresponding follow-ups.

This case shows us that, as with any disease with vital implications from massive spontaneous haemorrhagic phenomena, it can go unnoticed by the patient and the untrained physician.

The conjunctiva affected rarely requires treatment due to spontaneous self-limitation. In some cases cauterization therapy has been used for telangiectasias (3).

In conclusion, the existence of spontaneous ocular haemorrhagic phenomena in the absence of culprit aetiological factors (5) (haemophilia, hysteria, conjunctivitis, vascular tumours, trauma, granulomas, silver nitrate treatments and tear-duct haemorrhagic regurgitation) has to alert us to the possible existence of vascular abnormalities and lead us to take a precise systemic medical history in search of haemorrhagic phenomena with the same characteristics in other bodily structures.

REFERENCES