



# Angiokeratoma of the nasal vestibule: a case report

## Nazal vestibülde anjiokeratom: Olgu sunumu

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Angiokeratomas are benign mucocutaneous lesions which are composed of enlarged veins in the dermis of the skin. Although angiokeratomas may be part of a systemic disease, it is important to distinguish them from other vascular lesions. In this report, we present a rare case of angiokeratoma of the nasal vestibule. To our knowledge, this is the first nasal angiokeratoma case in the literature. The diagnostic and treatment features of this case and the association of angiokeratoma with systemic diseases like Fabry disease were discussed.

**Keywords:** Angiokeratoma; epistaxis; Fabry disease; nasal obstruction; nose.

Anjiokeratomlar derinin dermisinde genişlemiş venlerin oluşturduğu benign mukokütanöz lezyonlardır. Anjiokeratomlar sistemik bir hastalığın parçası olabileceği gibi, bunların diğer vasküler lezyonlardan ayırt edilmeleri önemlidir. Bu yazıda nadir bir nazal vestibül anjiokeratomu olgusu sunuldu. Bildiğimiz kadarıyla bu olgu literatürdeki ilk nazal anjiokeratom olgusudur. Bu olgunun tanısı ve tedavi özellikleri ve anjiokeratomun Fabry hastalığı gibi sistemik hastalıklarla birlikteliği tartışıldı.

**Anahtar Sözcükler:** Anjiokeratom; burun kanaması; Fabry hastalığı; burun tıkanıklığı; burun.

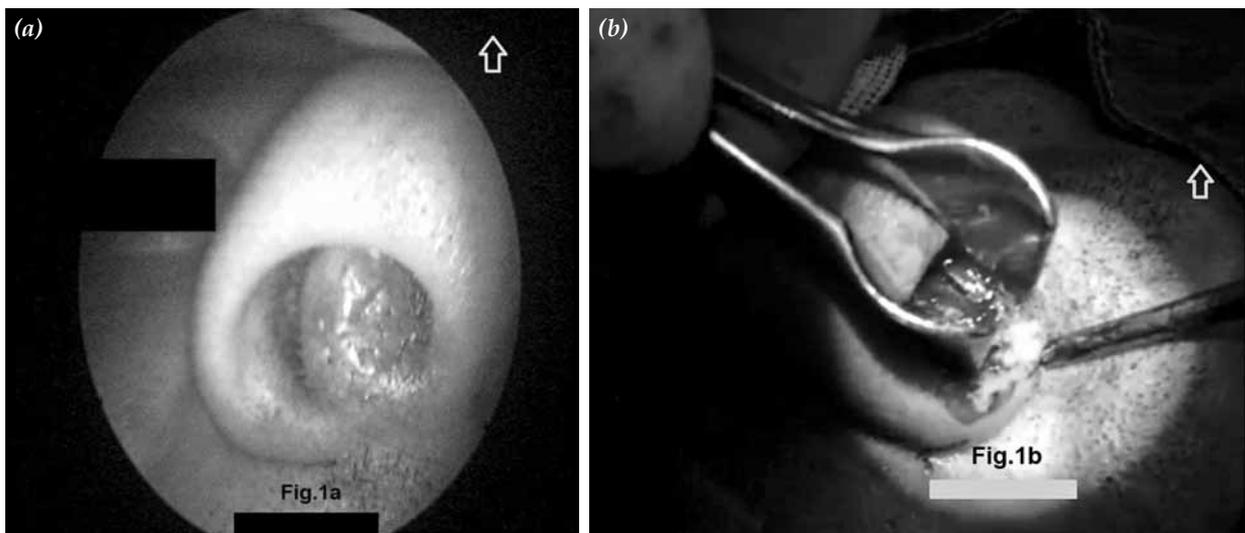
Angiokeratomas are benign mucocutaneous lesions composed of numerous dilated vessels in the superficial dermis, and hyperkeratosis overlying these dilated vessels of the skin.<sup>[1,2]</sup> They are usually asymptomatic but rarely may present with hemorrhage, cosmetic deformity, and other complaints depend on the location of the lesions. It is important to distinguish them from other vascular tumors. But more importantly, we have to determine the systemic conditions that may cause these lesions. They are usually encountered in various parts of body but to the best of our knowledge, have not been previously reported in

the nasal vestibule. We present what we believe to be the first case of angiokeratoma of the nasal vestibule and discuss an algorithm for its diagnosis and treatment.

### CASE REPORT

A 24-year-old man was referred to our otorhinolaryngology department complaining of a congenital nasal mass on the right nasal vestibule that caused nasal obstruction and intermittent epistaxis. There was no history of nose trauma. Physical examination showed an approximately 1 cm purple colored, painless, compressible, and





**Figure 1.** (a) One centimeter purple colored, painless, compressible, easily hemorrhagic nasal mass in the right nasal vestibule. (b) Preoperative view of the mass.

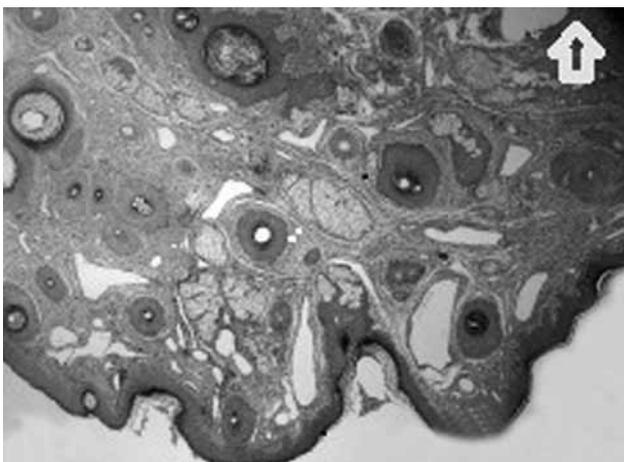
easily hemorrhagic nasal mass on the right nasal vestibule. It could be seen easily without using a nasal speculum (Figure 1a).

An incisional biopsy was performed under local anesthesia. The procedure was very bloody but controlled by bipolar cauterization. Histopathological analysis reported an angiokeratoma. We excised the mass totally under general anesthesia with 1 mm adjacent skin, and the defect was closed primarily (Figure 1b). Hyperkeratosis and hyperplastic epidermis, involving severely dilated blood vessels are seen on histopathologic examination

(Figure 2). We performed a general physical examination and a wider anamnesis to rule out a systemic condition such as Fabry disease and referred the patient to cardiology, ophthalmology and dermatology clinics for further examination. We also obtained a blood sample for nephrologic and genetic analysis for Fabry disease. No additional disease was determined by these consultations, and the blood tests and genetic analysis were negative for Fabry disease. Due to the solitary nature of this lesion, other metabolic disorders were not investigated. There was no recurrence on the postoperative first year control examination.

## DISCUSSION

There are several clinical types of this rare disease described according to the location and multiplicity of lesions.<sup>[3]</sup> They may be classified under two headings-- generalized and localized.<sup>[2-4]</sup> The systemic form is known as angiokeratoma corporis diffusum and is usually linked to some metabolic disorders, mainly Fabry's disease and fucosidosis.<sup>[3]</sup> The localized angiokeratomas include angiokeratoma of Mibelli (angiokeratomas on the dorsa of the fingers and toes), angiokeratoma of Fordyce (scrotal and vulvar form), angiokeratoma circumscriptum and solitary popular angiokeratoma.<sup>[4,5]</sup> Angiokeratoma circumscriptum presents at birth or early in life over the legs as a large, unilateral, hyperkeratotic plaque composed of confluent keratotic papules.<sup>[5]</sup> Solitary popular



**Figure 2.** Hyperkeratosis and hyperplastic epidermis, involving severely dilated blood vessels are seen on histopathologic examination (H-E x 4).

angiokeratoma is the last localized variant of angiokeratomas that is differentiated from others by its onset later in life.<sup>[4]</sup>

The histopathology of these lesions shows a vascular proliferation within the papillary dermis overlaid by acanthotic and hyperkeratotic epidermis. In addition to these, the presence of swollen and vacuolated (lipid-containing) endothelial cells are characteristically seen in Fabry's disease or fucosidosis-associated angiokeratomas.<sup>[5]</sup> In angiokeratomas, the vessels are covered by normal-appearing endothelium and occasionally filled with erythrocytes and organized thrombi.<sup>[1,5]</sup> Because of this thrombosis, angiokeratomas may mimic malignant melanoma but in contrast to hemangioma, do not include endothelial proliferation.<sup>[5]</sup>

A very important point in the management of this disease is performing a systemic physical examination to determine further lesions of the skin and mucous membranes which may be associated with systemic diseases such as Fabry disease, fucosidosis, sialidosis (Mucopolidosis Type 1), GM1 Gangliosidosis, galactosialidosis etc.<sup>[1]</sup> Fabry disease is the one most associated with the angiokeratomas.<sup>[3]</sup>

Fabry disease is an X-linked lysosomal storage disorder caused by the deficient activity of the lysosomal glycohydrolase  $\alpha$ -galactose A. Because of this deficiency, the sphingolipid globotriaosylceramide (Gb3) accumulates in the lysosomes, and causes multisystem pathologies.<sup>[6,7]</sup> The kidneys, heart, and brain may be affected. Since 2001, intravenous enzyme replacement therapy (ERT) has been used to increase the clearance of Gb3.<sup>[1,6,7]</sup> Therefore, it is very important to diagnose this disease and to begin treatment before the appearance of irreversible organ failures.

When an angiokeratoma is encountered the patient should be examined for other angiokeratomas on the body. If it is an isolated or localized angiokeratoma, the next diagnostic step is obtaining a wider anamnesis and family history from the patient for predisposing conditions. The family history must especially include sudden and/or premature deaths from cardiac or renal disease in relatives. We found premature cardiac disease in our patient's family history, and decided to make a wider investigation and genetic analysis. But a negative family history

for both sexes is not important to exclude this diagnosis because de novo mutations have been reported.<sup>[1]</sup> We can also suggest referring the patients to ophthalmology, cardiology and nephrology departments. In the presence of widespread angiokeratomas, measurement of alpha galactosidase is also recommended<sup>[1]</sup> but in females who can have normal levels of the enzyme in about 30% of cases, a mutational analysis may be required to determine the disease.<sup>[1,7]</sup> If the diagnosis of Fabry disease is ruled out, more rare metabolic disorders like fucosidosis, sialidosis, GM1 Gangliosidosis may be looked for.<sup>[1]</sup>

The main therapy for angiokeratomas is surgical excision. There are a few cases treated with laser excision in the literature,<sup>[3]</sup> but we did not choose laser treatment because of the localization of the lesion, and our desire to excise the entire lesion for histopathological examination.

In conclusion, we presented an angiokeratoma of the nasal vestibule that is not expected in this region. These lesions may be added to the list of nasal masses for rhinologists. It is important to make a differential diagnosis from vascular tumors of the nasal vestibule, and as a possible part of a systemic disease.

#### Declaration of conflicting interests

The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

#### Funding

The authors received no financial support for the research and/or authorship of this article.

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