Familial case of oral white sponge nevus - a rare hereditary condition *

Caso familiar de nevo branco esponjoso oral - uma rara condição hereditária

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Abstract: White sponge nevus (WSN) is an autosomal dominant skin disorder characterized by white, corrugated and diffuse plaques mainly affecting the oral mucosa. The condition has a high penetrance and variable expressivity, but familial reports are uncommon. This report presents a familial case of WSN in which two sisters are affected by the disorder.
Keywords: Leukokeratosis, Hereditary mucosal; Oral mucosa; Nevus

Resumo: O nevo branco esponjoso é uma desordem autossômica dominante, caracterizada por placas brancas difusas, rugosas, que afetam principalmente a mucosa bucal. A condição tem um alto grau de penetrância e expressividade variada, embora os relatos familiais sejam incomuns. Este artigo relata um caso familiar de nevo branco esponjoso em que duas irmãs são afetadas por esta condição.
Palavras-chave: Leucoceratose da mucosa hereditária; Mucosa bucal; Nevo

INTRODUCTION

White sponge nevus (WSN) is an autosomal dominant skin disorder first described by Hyde in 1909 and formally identified by Cannon in 1935. [1 2] Clinically, the disease is characterized by the presence of white, corrugated and diffuse plaques in the oral mucosa. [3] In some patients extra-oral sites such as the mucous membrane of the nasal cavity, esophagus, rectum and vagina are affected. The clinical manifestations tend to appear at an early age. Women are more affected than men by a ratio of 3:1. [4] WSN has a high degree of penetrance and varied expressivity, although familial reports are uncommon. [5] This paper presents a familial case of WSN in which two sisters are affected by the disease.

CASE REPORT

Two sisters, 13 and 15 years-old, presented complaining about the presence of asymptomatic rough, diffuse, white patches of unknown etiology and progress, located bilaterally in the oral mucosa, mucosa of the upper lip, gum and ventral surface of the tongue (Figures 1A and 2A). A review of family history revealed that the adolescents mother had similar lesions in her mouth, confirmed by clinical examination, suggesting the presence of an autosomal dominant disorder. No lesions in other body sites were reported in any of the cases. The medical history of the patients revealed no systemic type changes. Cytology was performed on the two adoles-

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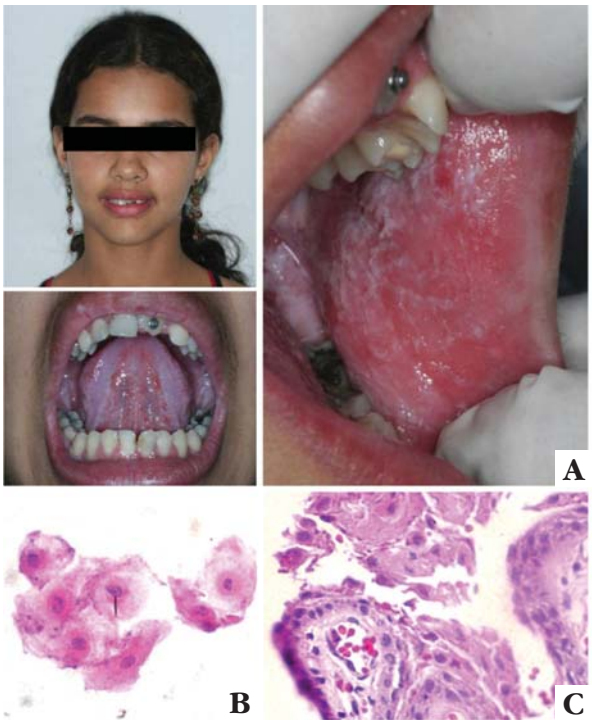

Figure 1: A. WSN in the vestibular mucosa and ventral surface of the tongue; **B.** Cytology showing epithelial cells with perinuclear eosinophilic condensation (Papanicolaou stain); **C.** Presence of hyperparakeratosis and acanthosis with vacuolization of keratinocytes in the suprabasal layers (hematoxylin and eosin, 200x)

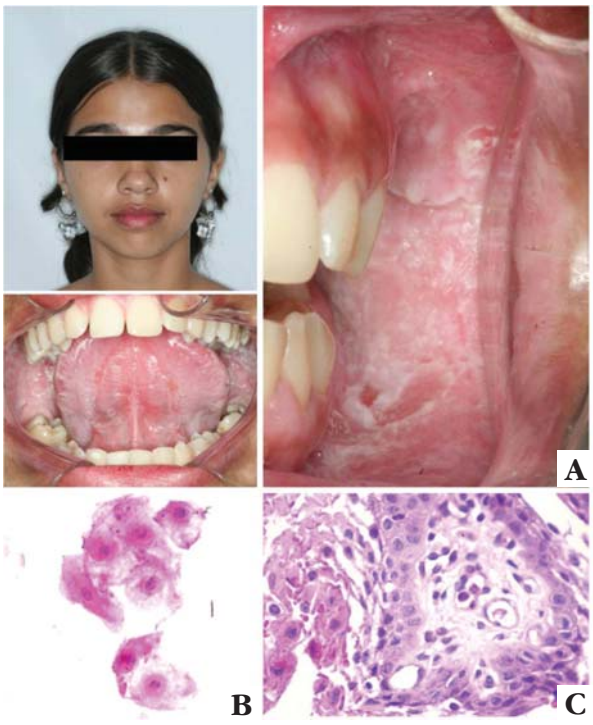

Figure 2: A. WSN in the vestibular mucosa; **B.** Exfoliative cytology showing epithelial cells with perinuclear eosinophilic condensation (Papanicolaou stain); **C.** Presence of hyperparakeratosis and acanthosis with vacuolization of keratinocytes in the suprabasal layers (hematoxylin and eosin, 200x)

cents, which showed the presence of perinuclear eosinophilic condensation of epithelial cells. An incisional biopsy was performed in the cheek mucosa and the specimens were sent for histopathological analysis. In both cases, hyperparakeratosis and acanthosis with *vacuolization* in *keratinocytes* in the suprabasal layers was observed (Figures 1B, C and 2B,C). After WSN was diagnosed no treatment was instituted and the patients are being monitored.

DISCUSSION

WSN is considered a rare disorder, affecting one in 200,000 people. [6] Some authors claim that the condition is related to mutations in *K4* and *K13* genes, characterized by defects in the maturation and desquamation of epithelial cells. [5,7] According to Terrinoni *et al*. [7], expression of WSN is highly variable, as well as the size of the plaques and their distribution in the oral mucosa. In our case, both sisters had similar lesions distributed in the oral mucosa, suggesting that the affected members of the same family might have the same expression pattern.

Lesions usually appear up to adolescence, with significant predilection for the cheek mucosa, followed by the ventral surface of the tongue, labial mucosa, the alveolar ridge and floor of the mouth. [7,8] In the case reported above the condition was found in two adolescent girls and in both cases the cheek mucosa and tongue dorsum were affected. The absence of pain is also an important clinical feature in patients with WSN. [4]

The differential diagnosis is made with other conditions presenting as white lesions on the oral mucosa. These include genodermatoses and acquired conditions such as leukoedema, linea alba, bitten mucosa, follicular keratosis, dyskeratosis congenita (DKC), congenital pachyonychia, focal epithelial hyperplasia, s*ystemic lupus erythematosus* (*SLE*), vegetative pioestomatitis, proliferative verrucous leukoplakia (PVL), oral florid papillomatosis, mucosal syphilids (*mucous plaques*), candidiasis, leukoplakia, frictional keratosis, and even squamous cell carcinoma. [9] However, the best differential diagnosis of white sponge nevus is done with the mucosal lichen planus (especially the reticular and plaque variants), since both lesions have the cheek mucosa as a preferred site and are usually present bilaterally. [10] In our case study, heredity was a primary factor for determining the

diagnosis as well as the patients´ age, given that lichen planus lesions are usually observed between the 4th and 6th decades of life. [11]

The histological findings are characteristic but not pathognomonic. [9] Family history leads to definitive diagnosis. [3] As for diagnostic tests, Messad *et al.* [9] have argued that the cytology of the epithelial cells shows characteristic perinuclear condensation better than the results of histological cuts. The histological features include acanthosis of the WSN, hyperparakeratosis and vacuolization of keratinocytes in the suprabasal layers. [12] In the event, the WSN diagnosis in our study was supported by family history as well as by cytological and histological findings.

Although no treatment is necessary for this condition, partial remissions have been documented with the use of systemic antibiotics. [13] However, in our case our chosen approach was to continue monitoring the two patients.

The above report describes a family case of WSN in which the disease is transmitted as an autosomal dominant inheritance with complete penetrance. Our main conclusion is that family history, together with cytology and histology, are vital clues for correctly diagnosing this condition and avoiding unnecessary treatment. ❑

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