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Maffucci's Syndrome: report of a case with oral manifestation

Síndrome de Maffucci: relato de caso com manifestação oral

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Abstract

Introduction: Maffucci's Syndrome is a rare nonhereditary mesodermal dysplasia consisting of multiple haemangioma of the soft tissue and enchondromas, mostly affecting phalanges and long bones. The syndrome can also be associated with a variety of other benign and malignant tumors. **Case report**: Here we report a case of Maffucci's Syndrome and haemangioma of lip and palate which is rare in this syndrome. This case report describes an 18 year old boy with multiple nodular soft tissue swellings involving the anterior hard palate and lower labial mucosa and bony abnormalities (enchondromas) involving the lower limb.

Keywords: Maffucci. Haemangioma. Chondroma.

Resumo

Introdução*: A Síndrome de Maffucci é uma displasia mesodermal não hereditária rara. Consiste de múltiplos hemangiomas de tecido mole e encondromas, a maioria afetando as falanges e os ossos longos. A síndrome pode também estar associada a uma variedade de tumores benignos e malignos.* ***Relato de caso****: O presente*

trabalho apresenta um caso de Síndrome de Maffucci e hemangioma de lábio e palato, o que é raro nessa síndrome, descrevendo o caso de um garoto de 18 anos de idade com inchaços nodulares múltiplos de tecido mole envolvendo o palato duro anterior e a mucosa labial inferior, além de anomalias ósseas (encondromas) acometendo um membro inferior.

***Palavras-chave**: Maffucci. Hemangioma. Condroma.*

Introduction

Multiple enchondromas with soft tissue haemangioma involving oral mucosa is termed as Maffucci's Syndrome, a unique condition seen less often in clinical practice. It consists of multiple soft tissue oral lesions and multiple bony abnormalities involving long bones. Till date, only about 150 cases have been reported in the literature (1). Maffucci's Syndrome does not exhibit any gender predilection and manifests with equal frequency in both the sexes. Tubular long bones are the most often affected in the skeleton, especially those of the upper extremity. Involvement of oral mucosa is a rare feature and the presence of haemangiomas is a very rare finding of this syndrome (1). A systematic review has shown that only about 5% to 10% of the reported cases have manifestations in head and neck region (1). Here we report a case of Maffucci's Syndrome with oral manifestation consisting of multiple haemangiomas, the exact incidence of which is not known and believed to be an occasional oral finding (1).

Case report

An 18 year old boy presented with a complaint of nodular masses on palate and lower lip region since six months. Patient reported that it interfered with normal chewing and was also of concern aesthetically. The patient's medical history revealed that he had undergone multiple resections of enchondromas performed earlier on his right lower limb two years ago with no other relevant history reported by him. A thorough physical examination revealed deep surgical scars of these resections on lower limb (Figure1). Intraoral examination revealed multiple bluish-red nodular growths involving oral mucosa of which three were seen in the lower labial mucosa, just lateral to the lower labial frenum and another was present in anterior hard palate. The labial and palatal nodules measured approximately 5mm in diameter (Figures 2, 3).On palpation, these swellings were soft in consistency and did not elicit tenderness. There was no evidence of any overlying ulceration or bleeding from the swellings. The swellings, however, showed blanching when diascopy was performed using a microscopic glass slide with restoration of the bluish red coloration on withdrawal of the applied pressure.

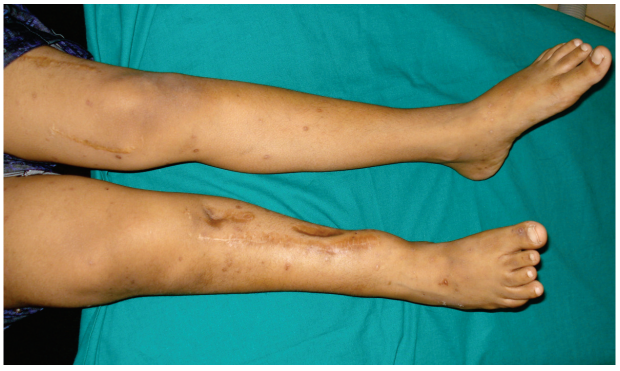

Figure 1 - The surgical scars remaining after the osteotomies seen on the lower extremities

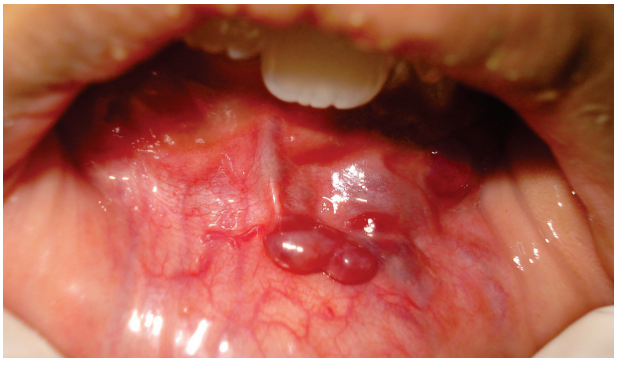

Figure 2 - The bluish-red nodular swelling measuring approximately 5mm in diameter seen lateral to labial frenum on left side in the lower labial mucosa obliterating the depth of the vestibule

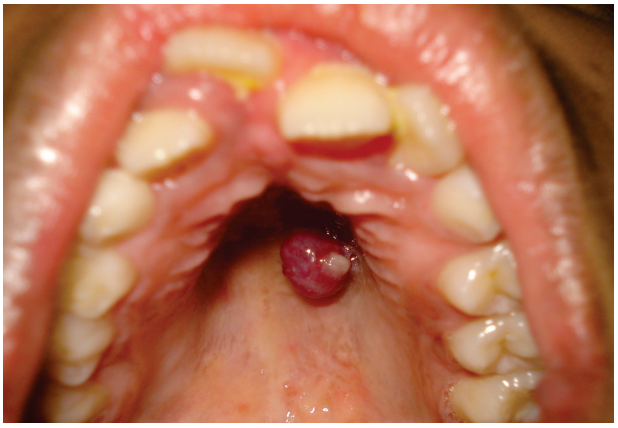

Figure 3 - A well defined bright red nodular swelling approximately 5mm in diameter seen in the anterior hard palate region

After correlating all these features, a diagnosis of Maffucci's Syndrome was made. The patient was advised excision of the labial and palatal lesions, which were surgically removed. On light microscopy (Figure 4), the excised specimen exhibited a background stroma that was fibrous and scanty with numerous plump proliferating endothelial cells walling the lumen, engorged with red blood cells. A mild chronic inflammatory response is seen mainly in the form of lymphocytes and plasma cells confirming the diagnosis of haemangioma. The healing after the excision was uneventful, but the patient could not report back for the follow up appointments and communicated via telephone that there was no evidence of any lesions on the mucosa two years after the excision.

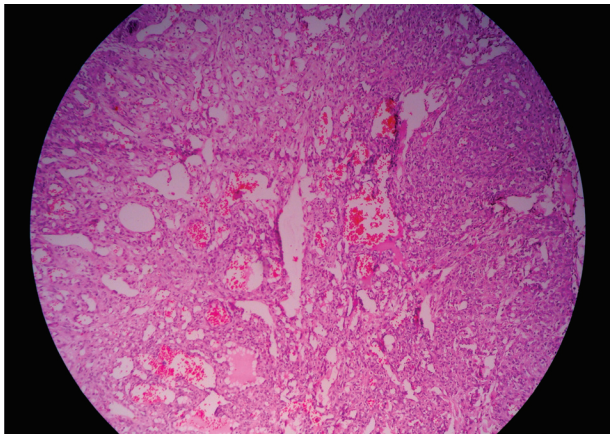

Figure 4 - Photomicrograph depicting proliferating endothelial cells forming the lumen filled with RBC (Red Blood Cell) (Haematoxylin and eosin stain, 10x Magnification)

Discussion

Multiple enchondromas of long bones with haemangioma involving oral mucosa is termed as Maffucci`s Syndrome (2, 3). Maffucci first described this condition in 1881, this condition was earlier known as Kast's Syndrome (3). This condition usually manifests around the age of five years (3). No definite sex predilection has been implicated. Patients affected are normal at birth and the lesions develop after five years of age and by puberty, they are invariably evident and either detected on routine clinical examination or maybe a presenting complaint. The age at the onset of the disease, affected sites, and the clinical features in our patient were seen to be in concordance with most of the reported cases. The soft tissue haemangiomas are mainly of the cavernous type. Oral manifestations present typically as haemangioma, with the generally favoured site being the tongue although there have been reports of lesions involving the cheeks, lips and soft palate (5, 6). In the present case soft tissue haemangioma was the predominant feature as compared to previous reports where bony abnormalities in long bones were reported. Malignant transformation occurring in the skeleton and vascular lesions has also been reported (2). The haemangiomas most often arise in early infancy, but the patient may notice it only after it grows to a larger size and cause an aesthetic or functional concern (6). In our case, the patient had noticed the vascular swellings just six months prior to presentation; however we feel that they may have been present for a while before that, until it affected his smile and made him seek consultation.

Management of Maffucci's Syndrome mainly relies on early identification and subsequent treatment depending on the nature of the lesion. Bony abnormalities affecting long bones may require treatment ranging from bony ostetomies to corrective surgeries depending on the extent of bony involvement. Various treatment options are available for managing soft tissue haemangioma ranging from injection of sclerotic agent, radiation therapy and surgical excision (1). In the present case, surgical excision of the soft tissue hemangiomatous lesion on lip and palate was carried out with favourable results. Due to the paucity of the reported cases and in particular those treated with excision, it not clearly known whether the resected lesions recur often or not.

In conclusion the above described case highlights the need for the dental clinician to be aware of such oral manifestations of Maffucci's Syndrome and be prepared to competently provide comprehensive oral care according to the patient's needs.

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