
Case Report

Maffucci's Syndrome with Oral Manifestations

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Maffucci's syndrome is a rare congenital mesodermal dysplasia combined with dyschondroplasia and hemangiomatosis. About 150 cases have been reported till now. Maffucci's syndrome is often combined with other neoplasms. Herein, we report a patient with Maffucci's syndrome and hemangiomas on the dorsum of the tongue, which is rare in this syndrome.

Archives of Iranian Medicine, Volume 12, Number 4, 2009: 421 – 423.

Keywords: Chondroma • hemangioma • Maffucci • oral manifestations

Introduction

Maffucci's syndrome is a rare congenital nonhereditary mesenchymal dysplasia manifested by enchondromatosis (dyschondroplasia) and soft tissue hemangiomas, which has a high incidence of malignant transformation. Till now, about 150 cases have been reported in the literature.^{1,2}

Maffucci's syndrome affects the skin and skeletal system with multiple enchondromas and hemangiomas occurring during the first two decades of life.^{3,4}

Although the distribution of the osseous and soft tissue alterations is similar, the sites of enchondromas and hemangiomas need not coincide. Involvement of the viscera and mucous membranes by hemangioma is a rare finding in this syndrome.^{5,6}

A review of the literature shows that about 5% to 10% of the reported cases have some involvement in the head and neck region.⁷ Oral manifestations are limited to occasional hemangiomas.⁷

The management of skin and oral lesions are surgical and depends on the size and location of

the lesions. Severe affection of the involved site may lead to amputation.⁵

We report a case of Maffucci's syndrome with hemangiomas on the dorsum of the tongue, which is rare in this syndrome.

Case Report

A 23-year-old man presented with chief complaint of two painless nodular masses on the dorsum of his tongue, which interfered with mastication and swallowing. He was suffering from multiple enchondromatosis in his left hand with deformity and shortening.

The bony lesions of the hand had appeared at the first year of his life and its shortening was caused by previous corrective surgery.

Multiple round- to oval-shaped cartilaginous nodules were noticed in his affected hand. Some small blue cutaneous painless nodules were found in the left hand too (Figure 1).

Because the tumors on the patient's tongue interfered with swallowing and mastication, he was operated on to excise the tumors in the summer of 2006. He is now free of the tumor after one year.

The tongue's lesions were two pieces of purple-brown, oval, and elastic soft tissue with polypoid surface measuring 1.5×2.5×2 cm, which enlarged recently (Figure 2).

Radiographic findings of the patient's hand revealed phleboliths within the hemangiomas.

Two excisional biopsy samples were taken

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Accepted for publication: 30 May 2008



Figure 1. Multiple enchondromatosis with deformity and shortening of the affected hand.

from the tongue's hemangioma and hand's enchondroma. The evaluation of the tongue's biopsy sample indicated a benign vascular lesion composed of tortuous and dilated spaces with flattened endothelial lining. These spaces were filled by red blood cells. No evidence of malignancy was found.

Microscopic evaluation of the hand's enchondroma showed mature lobules of hyaline cartilage in addition to foci of myxoid degeneration, calcification, and enchondroma ossification, which were the characteristic features of the disease.

Discussion

Multiple enchondromas (dyschondroplasia, Ollier's disease) with co-existing soft tissue hemangiomas is termed as Maffucci's syndrome.^{8,9}

Maffucci first reported this condition in 1881. Carletone et al. proposed the eponym "Maffucci's syndrome" in 1942.⁴

No familial pattern, sexual, or racial predilection is detected in this syndrome.^{4,8}



Figure 2. Hemangiomas on the dorsum of the tongue.

The disease usually appears around the age of five years. Of the cases, 25% are congenital and 78% before puberty. Forty-five percent of the symptoms are manifested before six years of age.¹⁰

Enchondromas are benign cartilaginous tumors that can appear in any parts of the body but are usually found on the distal phalanges and long bones. These bony abnormalities are usually asymmetric and can cause secondary fractures.⁴ Our patient was a typist and did not have any complaints with his hand's function.

Soft tissue hemangiomas in Maffucci's syndrome are mostly located in the subcutaneous tissues and appear as blue nodules which can be emptied by manual compression.^{5,10} Both cavernous and capillary hemangiomas have been seen in this syndrome but the cavernous type is more common.^{4,8,7}

The age at the onset of the disease, affected sites, and the clinical features in our patient were similar to the most of reported cases.

This syndrome is often combined with other neoplasms. Malignant transformation can occur in both enchondromas and hemangiomas.¹ Chondrosarcoma is the most common neoplasm in Maffucci's syndrome,^{4,5,11} although malignant transformation of the skeletal lesions to fibrosarcoma and of the vascular lesions to hemangiosarcoma, hemangioendothelioma, or lymphangiosarcoma had been reported previously.⁵

Malignancies other than musculoskeletal ones have also been reported in Maffucci's syndrome. The overall prevalence of malignancies associated with Maffucci's syndrome is 23% to 100% in different studies.^{5,6}

Although malignant changes are almost common in this syndrome, there was no evidence of malignant changes in our patient at the time of diagnosis and after a six-month follow-up.

Management of Maffucci's syndrome aims at the relief of symptoms and early detection of malignancies. Operative procedures for the skeletal lesions, such as corrective osteotomy and lengthening of the arm and leg, have been reported.^{5,6} Sclerotherapy, irradiation, and surgery for vascular lesions have been described.^{5,6} In our case simple excisional surgery on the tongue brought out good results.

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