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Case Report

Maffucci Syndrome with Composite Hemangioendothelioma: Two Rare Entities Superimposed Upon Each Other

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ABSTRACT

Maffucci syndrome is a rare non hereditary disorder characterized by multiple enchondromas and hemangiomas. A 23 year old female presented to us with multiple nodular swellings over the left hand and a few bright red soft swellings over the left ear and oral cavity. They were found to be enchondromas and hemangiomas respectively. She also presented with a progressively increasing painless swelling over the left foot which on histology showed the features of composite hemangioendothelioma. It is a recently described unusual vascular tumour of low grade malignancy exhibiting a composite of hemangioendothelioma variants.

Key words: Maffucci syndrome, enchondroma, hemangioma, lymphangioma, hemangioendothelioma.

INTRODUCTION

Enchondromas are commonly occurring benign hyaline cartilage neoplasms of medullary bone which are mostly solitary and asymptomatic. Multiple enchondromas (enchondromatosis) in association with soft tissue hemangiomas is known as Maffucci's syndrome (MS). It was first described by Dr. Angelo Maffucci in 1881. It is rare and a little more than 200 cases have been reported so far in the world. [1] It occurs in all the races with no sex preponderance. Familial occurrence is not reported though occasional reports of disease among siblings have been reported. [2] Composite hemangioendothelioma is an exceedingly rare vascular tumour, with only 29 cases being described in the medical literature so far. [3] In association with Maffucci syndrome, only a single case has been described till date. [4] To the best of our knowledge, this is the first case being reported from India describing an association of Maffucci syndrome with composite hemangioendothelioma.

CASE REPORT

A 23 year old female presented in the dermatology clinic with a red, slightly raised, pruritic, painless, firm swelling over the dorsum of the left foot for 3 months. The swelling had been increasing in size progressively.

She had experienced fracture in the bones of the left forearm following trauma at the age of 5 years. Following this, she started developing multiple painless swellings over the digits of the left hand since the age of 13 years. They started over the little finger, gradually involved the rest of the digits as well, with little finger and the ring finger being more involved and the thumb being completely spared. Subsequently both the deformed fingers were amputated one and a half year ago.

A year ago she started developing skin colored, slightly raised, non itchy, painless lesions over the lateral aspect of left hip. They started to increase in extent and progressed distally to involve the left side of thigh in 2-3 months. Simultaneously, she developed similar lesions over the inner aspect of left leg near the knee region. There was appearance of bright red, raised, asymptomatic nodular lesions over the tongue, inner aspect of the lower lip and helix of the left ear.

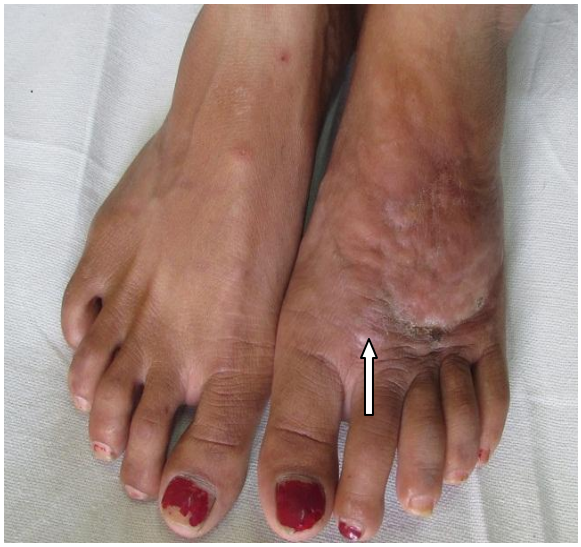

Figure 1: Erythematous to hyperpigmented plaque over the dorsum of left foot- composite hemangioendothelioma.

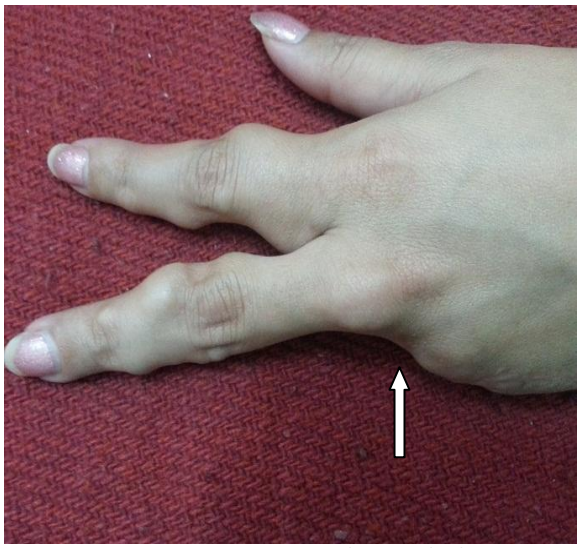

Figure 2: Amputated ring and little fingers of left hand, with bony nodular swellings over index and middle fingers- enchondromas

On mucocutaneous examination, dorsum of the left foot revealed, a single ill to well defined erythematous to hyperpigmented irregular, non-tender plaque of size 8×4cm with variable soft to hard consistency {figure1}. Index and middle fingers of the left hand revealed multiple skin colored, bony hard nodules of variable size 1× 1 cm to 2×2 cm {figure2}. Left lower limb, over the hip, lateral thigh and the medial aspect of knee, revealed multiple skin colored, discrete, soft, non tender papuloplaques of size 0.5 × 0.5 cm to 3×2 cm {figure3}. Lower labial mucosa and the dorsum of the tongue, exhibited two well defined bright red soft nodules of size 1×1×1 cm {figure4, 5}. Helix of the left ear also demonstrated a similar lesion {figure6}.

Routine investigations including complete haemogram and biochemistry were within normal limits. X-rays showed multiple radiolucent lytic lesions in the phalanges of left hand and left foot, favouring the diagnosis of enchondromas {figure7}. Ultrasonography & MRI of abdomen and pelvis revealed no visceral involvement except multiple enchondromas in the left ilium and scanned part of left upper femur. MRI brain revealed no abnormality either. Bone scan illustrated an evidence of increased tracer uptake over the left side of the body, in the scapula, forearm bones, ala of sacrum and pelvic bone, proximal femur and lateral aspect of knee joint {figure8}, suggestive of enchondromas.

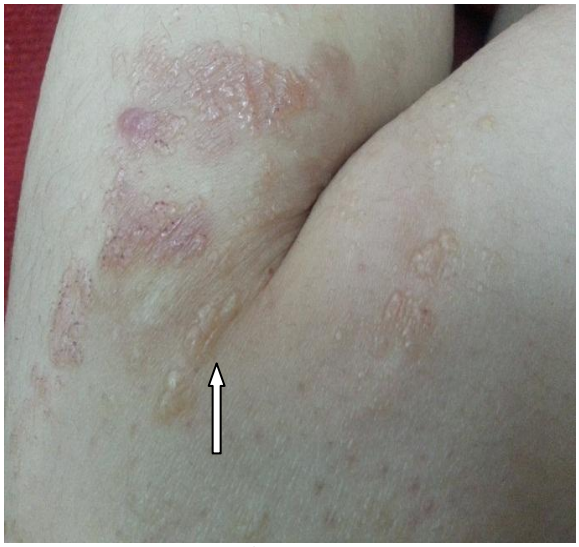

Figure 3: Skin coloured, soft papuloplaques- lymphangiomas.

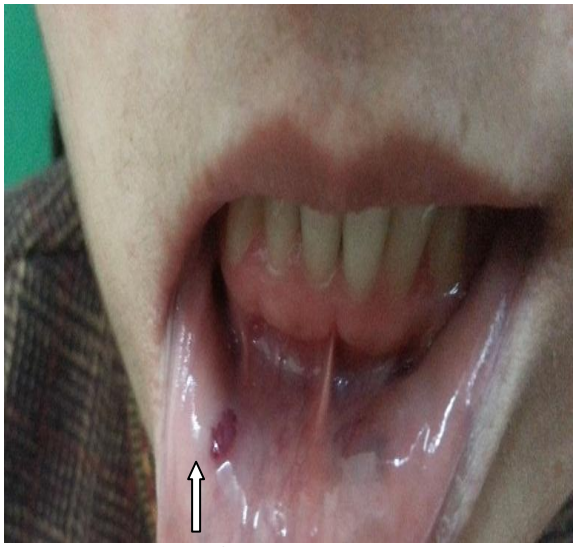

Figure 4: Bright red, soft nodule over the lower labial mucosa-hemangioma.

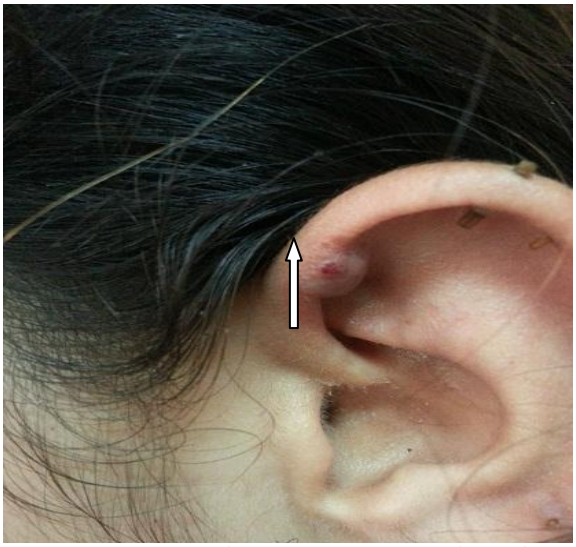

Figure 6: Bright red, soft nodule over the helix of the left ear-hemangioma.

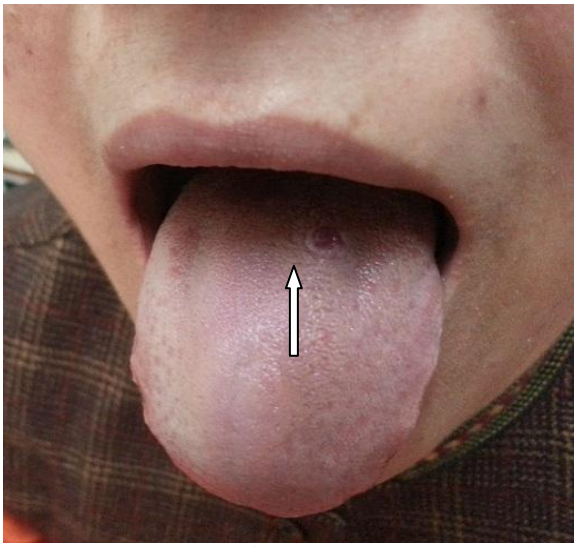

Figure 5: Bright red, soft nodule over the dorsum of the tongue- hemangioma.

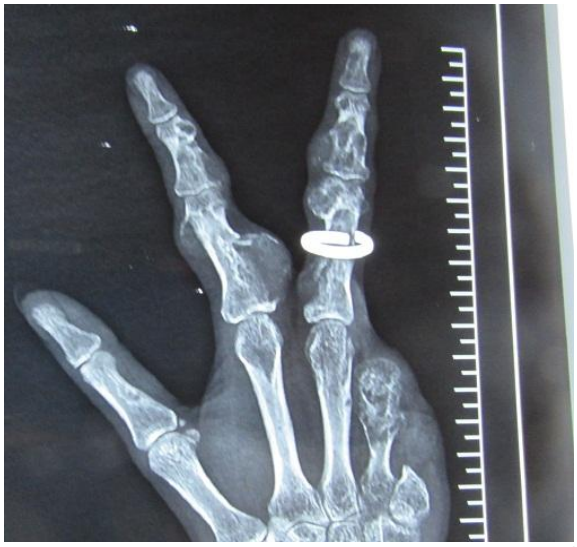

Figure 7: Multiple radiolucent lytic lesions in the phalanges of left hand-enchondromas.

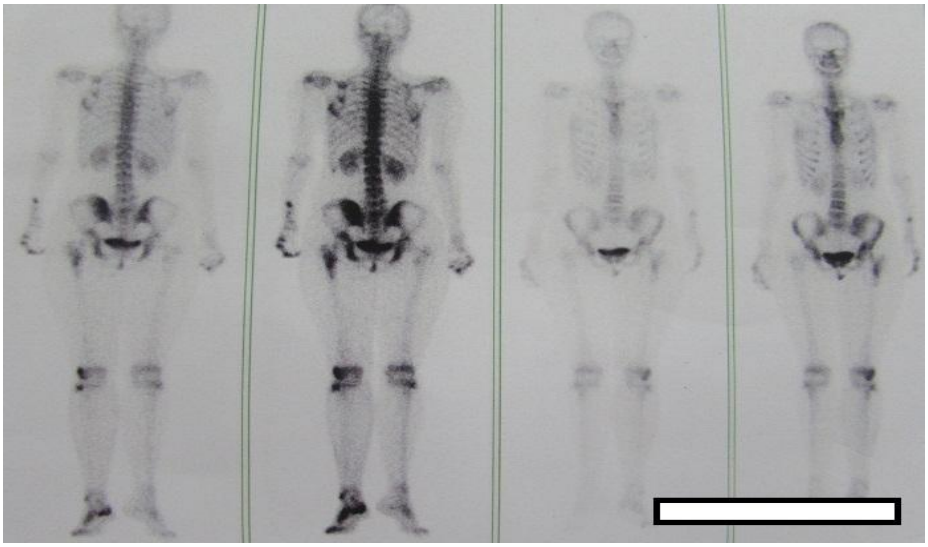

Figure 8: Increased tracer uptake seen over left side of the body, over the scapula, forearm, metacarpophalangeal joints, ala of sacrum and pelvic bone, proximal femur, lateral aspect of knee joint-enchondromas.

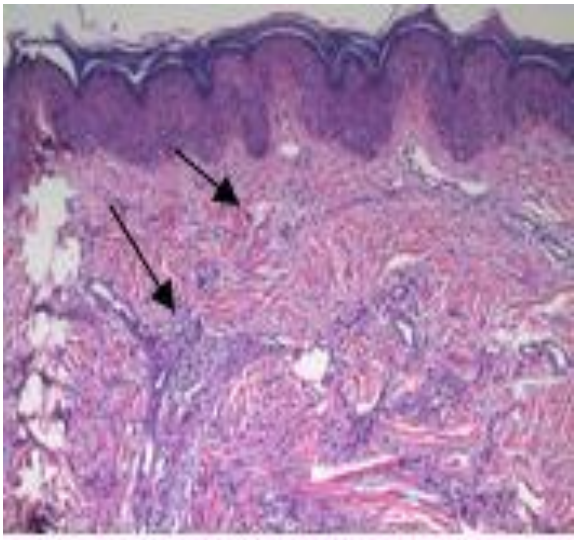

Figure 9: HPE 10x: variously sized and shaped vascular channels lined by neoplastic cells.

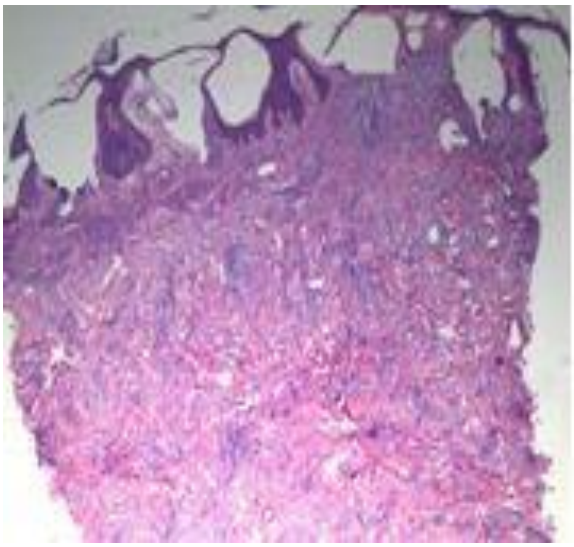

Figure 10: HPE 10x: Cords and nodules of solid proliferation of the neoplastic cells.

Skin biopsy of the lesion over the left foot revealed a non epithelial neoplasm made of numerous variously sized and shaped vascular channels which were lined by neoplastic cells, some of which were elongated and spindle shape while others were plumper and some were even plasmacytoid. Cords and nodules of solid proliferation of these cells were also appreciated. There was no nuclear pleomorphism or mitotic figures. Several dilated channels were seen enlarging dermal papillae resembling a lymphangioma. These finding were consistent with a composite hemangioendothelioma {figure9, 10}. Immunohistochemistry revealed positivity for CD34 and vimentin. Histopathology of the leg lesion revealed cystic spaces and dilated lymphatics in the dermis suggesting the diagnosis of lymphangioma. The patient has been planned for chemotherapy in the department of Oncology.

DISCUSSION

Multiple enchondromas (dyschondroplasia, Ollier's disease) with co-existing soft tissue haemangiomas is termed Maffucci syndrome. Both these conditions tend to involve one side of the body more than the other, [5] as found in our patient. Ollier's disease is thought to belong to the spectrum of the same disease process [2] but the incidence of progressive deformity and malignant transformation is greater in Maffucci syndrome. [5]

Maffucci syndrome comprises multiple enchondromas and haemangiomas. This rare syndrome was first reported by Maffucci in 1881. Maffucci described a thorough autopsy and reported all the main points of the syndrome. Carleton et al proposed the eponym Maffucci syndrome in 1942. [6]

Enchondromas are usually in close proximity to, or in continuity with the growth plate of cartilage. Consequently, they may result from abnormal regulation of proliferation and terminal differentiation of chondrocytes in the adjoining growth plate. A mutant PTH/PTHrP type I receptor (PTHR1) has been detected in human enchondromatosis, that signals abnormally *in vitro* and causes enchondroma like lesions in transgenic mice. [2]

There seems to be no racial or sexual predilection for this syndrome. [1,6,7] The disease manifestations usually appear around the age of 5 years. Twenty five percent of cases are congenital and 78% begin before puberty. Patients are of average intelligence and no mental or psychiatric abnormalities have been noted. The disease seems to develop from mesodermal dysplasia early in life. [6]

Enchondromas are benign cartilaginous tumours that may develop at

any site but are most frequently found in phalanges and long bones. [8] Phalanges of the hand are the preferred site, [5] as seen in our patient. Complications include spontaneous fracture through an area of rarefaction, [8] as found in our patient at the age of 5 years. Chondrosarcomatous transformation in the enchondromas can occur in about 30% of cases. [2]

There is an association with vascular lesions as well- cavernous hemangiomas, phlebectasias and lymphangiomas. [8] Composite hemangioendothelioma in association with this syndrome has previously been described only once in the English literature. [4]

Composite hemangioendothelioma is considered a low to intermediate grade malignant vascular tumour displaying varying combinations of benign and malignant vascular components. [3] It presents on the dorsum of the hands or feet, and on fingers and toes, as single or multiple nodules. It usually affects adults from 21 to 71 years of age (mean 41 years). [9] Histopathological diagnosis requires the presence of at least two hemangioendothelioma variants. Epithelioid and retiform variants are the most commonly observed, but foci of spindle cell hemangioma and angiosarcoma may also be. [4] The tumour can show positive immunostaining for endothelial cell markers (CD31, CD34, and Factor VIII) and spindle cells (vimentin). [3,4,9] Local reccurence of this tumour after initial excision has occurred in eight of the previously reported cases, while metastasis is even rarer, having occurred in four cases so far. [3] The best therapeutic approach and prognosis remain unclear, due to the paucity of cases in literature. The reported high rate of local recurrence suggests excision should extend beyond clinical margins. Adjuvant radiation therapy and chemotherapy may also be considered, although more data are needed to obtain a better understanding of the prognosis and malignant potential of this rare vascular tumour. [3]

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