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Case Report

Radiology Section

Radiological Pearls in Imaging of Maffucci Syndrome- A Case Report

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ABSTRACT

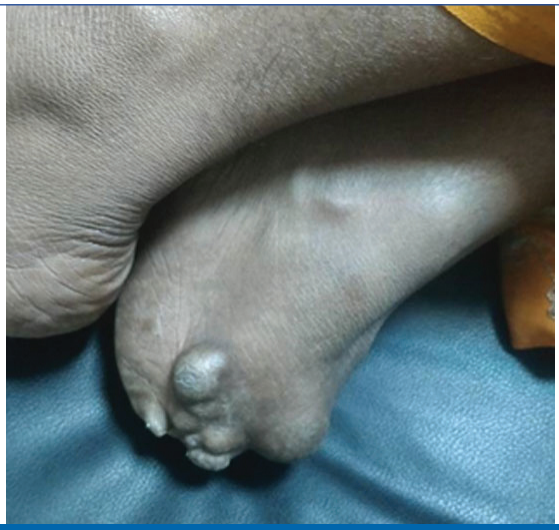
Maffucci syndrome is a congenital mesodermal dysplasia. The disease is characterised by the presence of multiple soft tissue haemangiomas or slow flow venous malformations and enchondromas and was first described in 1881. This report aims to discuss the clinical and radiological findings of Maffucci syndrome in a 29-year-old female patient who presented with swelling in the right foot and foot deformity. The subsequent history taking and different imaging modalities helped to reach the diagnosis. On Ultrasonography (USG), phleboliths were visible and on Computed Tomography (CT), few calcific foci were noted in planter aspect of right foot in subcutaneous and intermuscular plane with soft tissue enhancement. Hence this case report showed that Maffucci syndrome can present as deformity of limb, multiple soft tissue swellings or with cranial nerve palsy which may suggest an intracranial lesion and close follow-up of patients is required as there are high chances of malignant transformation. Patient in this case was referred to the Neurosurgery Department and later on lost to follow-up.

Keywords: Chondrosarcoma, Deformity, Dysplasia, Enchondromas, Haemangiomas, Slow flow malformation

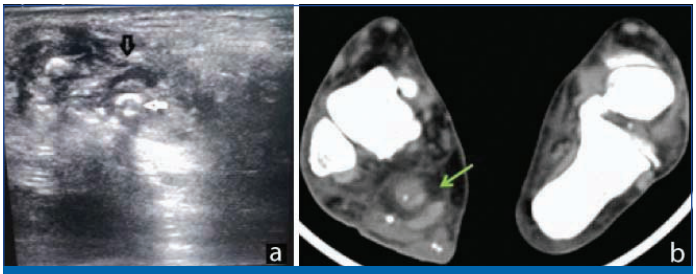
CASE REPORT

A 29-year-old female presented in the Department of General Surgery with swelling and deformity in the right foot for past 5 years [Table/Fig-1]. The toes of the right foot were not seen and foot was deformed. Only small part of the foot distal to the ankle was seen. On planter aspect of visualised foot, few small round soft tissue swellings were present which were soft to firm in consistency, average size measuring 10×10 mm in size. The patient was advised for ultrasound followed by CT Angiography of lower limbs and was referred to the Department of Radio diagnosis. On USG, a multiloculated cystic lesion was seen with presence of few echogenic foci within, suggestive of phleboliths [Table/Fig-2a].

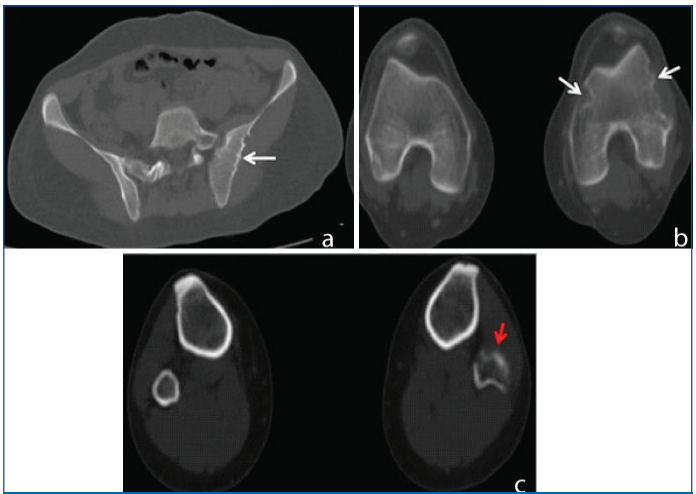
On CT, few calcific foci were noted in planter aspect of right foot in subcutaneous and intermuscular plane with mild enhancing soft tissue in venous phase images [Table/Fig-2b]. The lower limb arteries were normal and no evidence of arterio-venous fistula or aneurysm was seen. Multiple small irregular cortical erosions were also noted postero lateral in aspect of left iliac bone along with mild expansion of the medulla, cortical erosions in medial and lateral aspect of left lower femoral condyles and distal metaphyses of left fibula were also seen [Table/Fig-3].



[Table/Fig-1]: Photograph of the right foot showing deformity and soft tissue swelling.



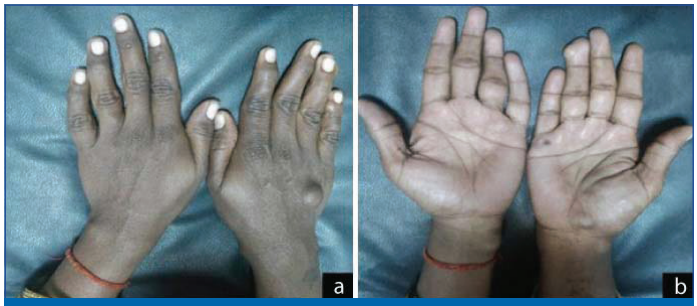
[Table/Fig-2]: a) Ultrasound image of the soft tissue slow flow venous malformation at planter aspect of right foot showing the multiloculated cystic lesion (black arrow) and echogenic foci (phleboliths) within (white arrow); (b) CT Angiography image showing the mildly enhancing soft tissue and calcifications in venous phase (green arrow).



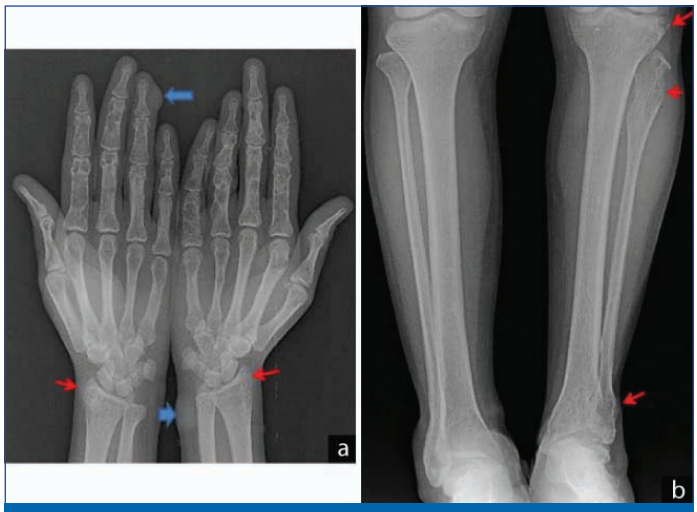
[Table/Fig-3]: CT with bone window images showing enchondromatosis: (a) Mild bony expansion with cortical irregularity of left iliac bone (white arrow); (b) Cortical irregularity of the lower condyle of left femur (white arrows); (c) Cortical irregularity of distal metaphysis of left fibula (red arrow).

Diagnosis of multiple soft tissue slow flow venous malformations with possibility of multiple enchondromas was made. On further probing, patient gave history of surgical excision of one of the soft tissue lesion in left elbow one year back. The histopathology report revealed the presence of dilated vascular spaces lined by

the single layer of endothelium containing red blood cells. There was no positive family history. On detailed skeletal examination, few soft tissue swellings with firm consistency were seen in both hands. Skin overlying the swelling was normal. Radiographs of both hands and feet were done which showed multiple lytic lesions with coarse trabeculae in phalanges of both hands and 4th metacarpal of left hand along with bony remodeling and endosteal scalloping. Lytic areas with mild cortical irregularity were also noted in the head of left fibula, left lateral tibial condyle, left lateral malleolus and bilateral radial styloid processes. Few round to oval soft tissue opacities were also seen in subcutaneous plane in right hand and left wrist with no evidence of calcification. It helped us to reach the diagnosis of multiple enchondromas and soft tissue slow flow venous malformations [Table/Fig-4,5].

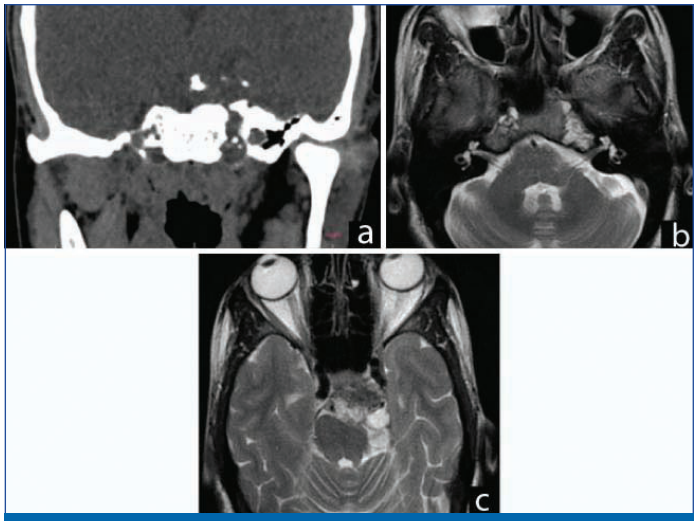


[Table/Fig-4]: Photograph of the patient hands showing soft tissue swellings and slight deformity with deviation of the fingers. Skin overlying the swelling is normal.



[Table/Fig-5]: Multiple enchondromas and soft tissue slow flow malformations. Radiograph of both hands: (a) and feet (b) were done which showed multiple lytic lesions with coarse trabeculae in phalanges of both hands and 4th metacarpal of left hand along with bony remodelling and endosteal scalloping. Lytic areas with mild cortical irregularity also noted in the head of left fibula, left lateral tibial condyle, left lateral malleolus and bilateral radial styloid processes (red arrows). Few round to oval soft tissue opacities are seen in subcutaneous plane in right hand and left wrist with no evidence of calcification (blue arrows).

On further systemic examination, there was mild medial deviation of left eye. Rest of the ocular examination was normal and visual acuity was 6/6 in both eyes. Screening Brain Magnetic Resonance Imaging (MRI) and CT head was done to rule out any intracranial lesion. They showed large lobulated T2 hyperintense lesion involving left petrous apex and clivus with large soft tissue component extending into the left cerebello-pontine cistern, prepontine cistern and left parasellar region causing mass effect on pons. The lesion was seen crossing the midline to opposite side. In Non-Contrast Computed Tomography (NCCT) Head, there were few calcifications noted within the lesion with bony erosions involving left petrous apex and clivus, suggesting the lesion likely to be chondrosarcoma. However, no bony erosion of right petrous apex was seen [Table/Fig-6]. Patient was referred to Neurosurgery Department but was later lost to follow-up. Based upon the above clinico-radiological findings, due to the presence of multiple soft tissue haemangiomas along with enchondromas, diagnosis of Maffucci syndrome was made.



[Table/Fig-6]: Noncontrast CT: (a) shows irregular hypodense mass lesion with few calcifications in bilateral petroclival fissure with bony erosions of left petrous apex. Axial T2 weighted images (b,c) showed large lobulated T2 hyperintense lesion in left petrous apex extending to opposite side with large soft tissue component extending into the left CP cistern, prepontine cistern and left parasellar region causing mass effect on pons suggesting the lesion to be Chondrosarcoma.
T: Time; CP: Cerebellopontine

DISCUSSION

In 1881, Angelo Maffucci had described the syndrome with onset in childhood or adolescence and includes the development of multiple enchondromas along with soft tissue haemangiomas [1]. Similarly, Ollier had described a syndrome of multiple enchondromatosis but without haemangiomas in 1900. Skeletal enchondromatosis can lead to deformity of limbs and can also undergo malignant degeneration [2]. Association with lymphangioma has also been reported in literature. Both Maffucci syndrome and Olliers disease are also known as mesodermal dysplasia syndrome characterised by proliferation of displaced islands of cartilage from the growth plate into the metaphyseal regions, hence also known as dyschondroplasia [2,3].

Both skeletal and soft tissue deformities are seen and skeletal involvement is most marked in extremities especially in hands [4]. Other sites of involvement in reducing frequency are foot, lower leg, femur, humerus, forearm, pelvis, and vertebrae [3]. Multiple types of neoplasia, of both skeletal and soft tissue origin, are reported in literature; but most common is chondrosarcoma and develops in more than 50% of cases in Maffucci syndrome and 40% in Olliers syndrome [2,4].

Radiographs are virtually pathognomonic in Maffucci syndrome, particularly of hands and feet, and show multiple varying sized expansile lytic lesions associated with remodeling and thinning of cortex with endosteal scalloping. There may be presence of matrix calcification in arc and ring pattern as seen in chondroid lesions. Associated deformity of limbs may be present due to interrupted skeletal growth during development [5]. Patwekar SL et al., described this syndrome in a 13-year-old girl who presented with multiple haemangiomas in hands and pre-sternal region, enchondromas in bones of both upper limbs and lower limbs with limb length discrepancy [6]. In the present case, similar radiographic skeletal findings were seen. Chouhan C et al., described radiographs, ultrasound and CT findings in a 30-year-old male patient who presented with multiple enchondromas in bones of right foot and right lower femur along with multiple soft tissue haemangiomas in right lower limb [7]. We also did radiographs, ultrasound, CT Angiography and MRI Brain without contrast to know the extent of lesions in the body.

The intracranial lesion is predominantly seen at the skull base, which is embryologically derived from cartilaginous matrix. The most common site of intracranial chondrosarcoma is spheno-occipital and spheno-petrosal synchondroses or from the parasellar region [2,8]. The malignancy in this syndrome is reportedly seen in

younger age similar to this case where patient is just 29-year-old. The symptoms may not manifest for many years as tumour is slow growing and patient may present with cranial nerve involvement later in life [2]. The present case also showed left 6th nerve involvement. MRI is particularly useful as lesions appear T2 hyperintense and show avid enhancement [9]. Also, disease extent and extensions can be evaluated to provide road map to the surgeons [6,7]. In this case, only screening Brain MRI was done as patient denied contrast examination. Also, the location of the lesion at the petro-clival region, T2 hyperintensity and calcifications in the tumour matrix more likely suggest chondrosarcoma. However, histopathological confirmation could not be done and patient was later lost to follow up.

Imaging of soft tissue swellings can be done via radiographs which may show soft tissue density lesions with calcifications within, ultrasound which may show cystic spaces and echogenic foci to suggest phleboliths and colour Doppler which shows presence of slow venous flow characteristic of cavernous haemangiomas/ slow flow venous malformation, CT can be done to confirm the soft tissue calcifications and enhancement in venous phase images and MRI shows T2 bright lesions and phleboliths as hypointense foci [5,10]. The International Society for the study of Vascular Anomalies (ISSVA) described the nomenclature of vascular anomalies and vascular tumours. The society also enlisted the various syndromic malformations and described the association of spindle cell haemangiomas or slow flow venous malformations in Maffucci syndrome along with enchondromatosis. Spindle cell haemangiomas are either a true neoplasm or a reactive process secondary to thrombus in dilated vessels and may contain phleboliths [11]. They contain thin walled cavernous vessels with occasional thrombi. Slow flow venous malformations present as discrete or multifocal masses with abnormal tubular channels. Thrombi or phleboliths within the lesion may lead to firm consistency in otherwise soft compressible lesion [11]. Histologically, it demonstrates malformed irregular venous channels with muscular walls and internal organising thrombi. Nozaki T et al., described that vascular anomalies associated with Maffucci syndrome are mainly low flow vascular malformations (venous and rarely lymphatic) [12]. According to the latest ISSVA classification, in this case, multiple soft tissue haemangiomas described are reclassified as slow flow venous malformations.

The major differentials in this case were Ollier's disease and Klippel-Trenaunay syndrome. However, Ollier's disease could easily be excluded due to the presence of slow flow venous malformations. Klippel- Trenaunay syndrome includes haemangiomas, varicose veins, and soft tissue and bony hypertrophy. The patients with Maffuci syndrome have shortened or deformed limbs while in Klippel-Trenaunay syndrome, usually excessive growth of the affected limb is present [13]. In the present case, deformity of right foot and absence of varicosities ruled out Klippel-Trenaunay syndrome.

CONCLUSION(S)

Maffucci syndrome is a relatively rare syndrome characterised by multiple soft tissue slow flow venous malformation and skeletal enchondromas and presentation in an adult patient with deformity of limb and soft tissue swellings, should prompt us to rule out this syndrome. Involvement of cranial nerve palsy may be the only first sign to point towards an intracranial lesion in this syndrome.

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