

A Unique Case of Maffucci's Syndrome Presenting with Right Iliac Bone Chondrosarcoma

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Abstract: We report a rare case of Maffucci's syndrome in a 40 year old male patient presenting with dedifferentiated chondrosarcoma of right iliac bone developing from previous enchondroma along with multifocal enchondromas and haemangiomas involving various other bones predominantly on right side. In this case X-rays, CT scan and MRI were used to reach the final diagnosis. MRI was of aid to diagnose the dedifferentiated carcinoma even before the surgery. T2WI were used to distinguish between the dedifferentiated and the cartilaginous tumor. Furthermore, the dedifferentiated tumor had three elements-malignant fibrous histiocytoma, osteosarcoma and fibrosarcoma. This histological heterogeneity may be attributed to mesodermal dysplasia of Maffucci's syndrome.

Keywords: Maffucci's syndrome, chondrosarcoma, enchondroma, right iliac bone.

INTRODUCTION

Maffucci's syndrome is rare disease, first described in 1881 with about 200 cases reported up till now [1, 2]. It is a congenital mesodermal dysplasia which is non-hereditary and asymptomatic, manifesting itself during puberty [3].

The incidence of malignant changes is around 25%, changes involving both soft tissue and bones. Very few cases have been reported of dedifferentiated secondary chondrosarcoma [1]. Our case presents a 40 year old male with Maffucci's syndrome and development of secondary chondrosarcoma

CASE HISTORY

A 40 year old male patient presented to the Radiology department with a large swelling in right hip which was painful and causing limitation of movements. Also, there were multi focal swellings in upper and lower limb which were showing progressive growth since last 6 months

The right hip swelling was large, painful and causing restriction in movement whereas swellings at other locations were small and associated with prominent subcutaneous component. There was no previous history of malignancy and no significant family history. (figure1 (a-d)).

On X-ray evaluation, the pelvis with both hips x-ray demonstrated a large, irregularly defined, radiolucent soft tissue lesion causing extensive erosion of cortex of right iliac bone with poor zone of transition and stippled as well as popcorn like calcification. Haemangiomas were also noted in the pelvic cavity.

Expansile, lytic lesions were also noted in phalanges, metacarpals, radius and ulna, tibia fibula and metatarsals as well as phalanges of foot on right side of body with deformity in radius ulna, tibia and fibula. There were haemangiomas noted in the soft tissue with flecks of calcification.

Chest x-ray did not show any metastases. But, there were osteolytic lesions involving multiple ribs on right side (figure 2 a-e).

Based on above x-ray findings a preliminary diagnosis of Maffucci's syndrome was made. Detailed radiological evaluation of pelvis with CT scan and MRI was done.

CT scan revealed large soft tissue lesion involving the right iliac blade extending into the ala of sacrum and ischial bone on right side while there is an enchondroma noted in left iliac bone and haemangioma in pelvic cavity with calcifications. Bone window cuts of CT scan thorax demonstrated multiple enchondroma with stippled and popcorn like calcification in scapula

and ribs on right side. But, could not differentiate muscle involvement from other extra osseous involvement (figure3 a-d).

MRI was of further help in demonstrating two components of the mass. It was performed on 1.5 tesla Philips MRI machine with the lower limb placed on surface coil and T1W and T2W spin echo sequences were taken. T1WI show a lobulated intra-osseous as well as extra osseous mass, hypointense to the bone marrow and iso intense with muscle tissue, however T2WI revealed extra osseous mass showing heterogeneous intensity - hypointense lesion with central hyperintense areas suggesting necrosis while intra osseous lesion is a lobulated predominantly hyperintense mass with partial discontinuity in the cortex of ilium and ischium suggesting type 2 dedifferentiated chondrosarcoma (figure4 a-d).

By the above findings, final diagnosis of dedifferentiated chondrosarcoma (type2) developing in an enchondroma was made.

Excision biopsy confirmed the diagnosis with results as-stratified squamous epithelium with focal ulceration beneath which is fibrocollagenousstroma with chronic inflammatory infiltrate. Focal areas showing cartilaginous matrix having 2 or more cells per lacuna. Nuclei are plump and hyperchromatic. Tumour cells forming osteoid is not seen.

On further histological examination of another specimen demonstrated a dedifferentiated component with mixed features of osteosarcoma, fibrosarcoma and malignant fibrous histiocytosis. Most of the fibrogenic areas show herring bone pattern. The dedifferentiated tumour was classified as fibroblastic osteosarcoma.



Fig-1: Multi-focal soft tissue swelling in images a, b and d involving arm, hand and leg on right side. Image "c" shows large right hip swelling causing it's deformity.



Fig-2: Multifocal osteolytic, expansile lesions with foci of calcifications involving multiple ribs on right side, phalanges and metacarpals of right foot and phalanges of right hand (predominantly middle phalynx of ring finger). Image "d" shows a large osteolytic lesion with calcification and soft tissue swelling involving the right iliac bone and anterior superior iliac spine. Similar lesion can also be appreciated in left iliac bone. Image "e" shows multiple sub cutaneous hyperdense areas likely to be haemangiomas involving the right arm

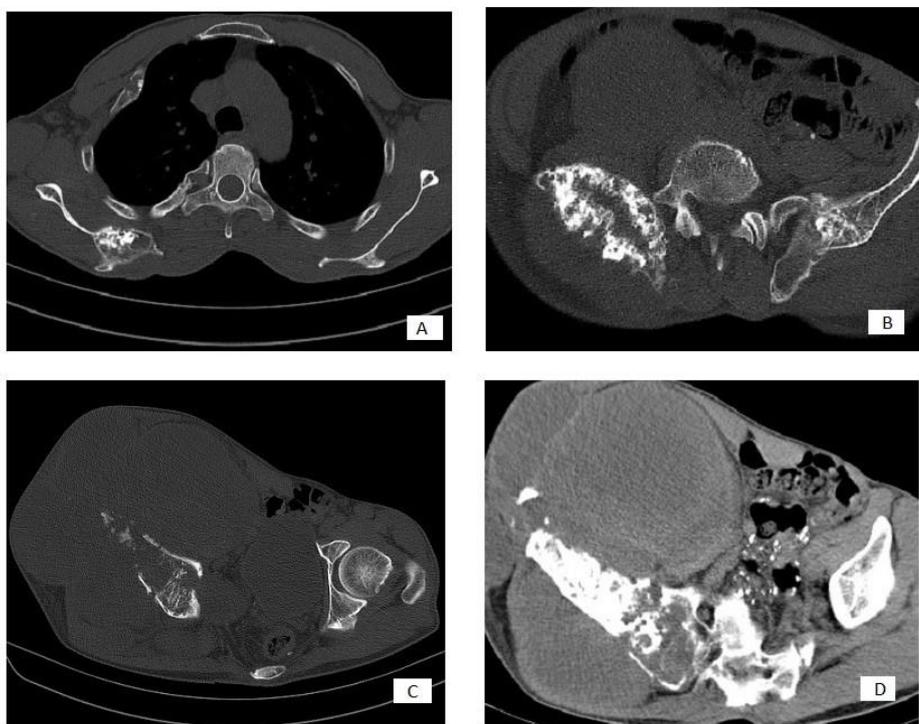


Fig-3: CT images showing multifocal enchondroma with stippled calcifications involving ribs, sternum , right scapula, pedicles of lumbar vertebra and pelvic bones. Fig d also shows the osteolytic expansile tumour causing breach in cortex involving the right iliac bone with surrounding soft tissue involvement and phleboliths in the pelvis.

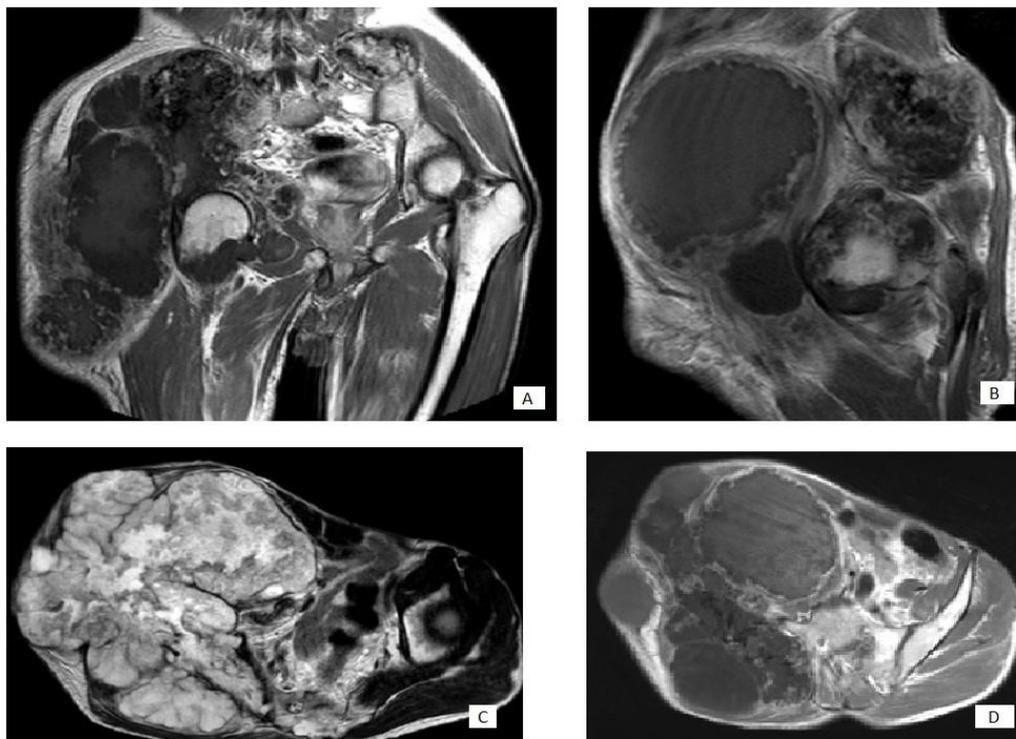


Fig-4: MRI T1/T2/T1CONTRAST images showing a heterogeneously enhancing, lobulated, soft tissue mass lesion with intra as well as extra osseous involvement as well as two components of the tumor-cartilage component and another less differentiated component. It shows mixed intensity on T1 and T2WI, predominantly hypointense on T1WI and hyperintense on T2WI

DISCUSSION

Maffucci's syndrome is a congenital mesodermal hyperplasia with multiple enchondromas, secondary bone deformities and Mesenchymal neoplasias. Mesenchymal neoplasias are in form of soft tissue haemangiomas and may be associated with phleboliths. Association has also been seen with lymphangiomas, lymphangiectasia and pigmentary macules. It was first described in 1881 by Angelo Maffucci. In the 2002 World Health Organization classification Maffucci's syndrome is considered as a subclass of enchondromatosis [5].

The incidence of malignant tumors is high with about 25% undergoing malignant transformation from an enchondroma to chondrosarcoma [6].

Enchondroma is a benign cartilaginous tumor arising from the medulla presenting as a central translucency with a narrow zone of transition. May be associated with punctuate or stippled calcification [4].

Multiple enchondromas are known as Ollier's disease and when it is associated with soft tissue haemangiomas, characterized radiologically as phleboliths, the disorder bears an eponym of Maffucci's syndrome [4].

Etiology is unknown with no sex predilection. Bone changes are caused by congenital defects in endochondral ossification and lead to deficits in final bone growth and bone irregularities. Enchondromas are more often found in hand phalanges, metacarpal bones, foot bones, tibia, fibula, radius and ulna [4]. Distribution is asymmetric, and bone lesions may range from painless edema to pathological fractures. Other bones may be affected, although not as often, as in the case described here, in which ribs were primarily affected [4].

Haemangiomas are usually found along soft tissues as bluish nodules, but may also be found in internal organs and mucous membranes, particularly in the brain, eyes, and gastrointestinal tract [7].

Pathological fractures are found in 26% of the cases; other possible complications are hemorrhage, short stature, pleural effusion and cranial nerve paralysis due to endochondral compression [7]. Our case shows malignant transformation to dedifferentiated chondrosarcoma.

Dedifferentiated chondrosarcoma was first described by Dahlin and Beabout. Radiographically, dedifferentiation is suggested by a sharply demarcated area of aggressive bone destruction associated with an

underlying cartilaginous lesion and the presence of an extra osseous soft-tissue mass.

Three radiographic types of dedifferentiated chondrosarcoma have been described [8, 9].

- Type 1- the radiographic appearance is the same as for a central chondrosarcoma, with the addition of a region with very aggressive bone destruction.
- Type 2-lesions resemble an underlying benign enchondroma but also have destructive changes and/or a large soft tissue mass.
- Type 3- lesions are not distinctive radiographically and present as a very high-grade destructive lesion of bone. The dedifferentiated component can easily be detected by MRI, as a sharply defined osteolytic area of reduced signal intensity adjacent to the hyperintense chondral component corresponding to regions of dedifferentiation. Three distinct MRI patterns have been described-
- In the first pattern, there is clear demarcation between the two regions of high and reduced T2-weighted signal intensity, a so-called biphasic pattern. This pattern typically correlates with a type 1 radiographic lesion.
- In the second pattern, the only MRI evidence of an underlying chondral lesion is the presence of multiple areas of signal void corresponding to matrix mineralization identified radiographically; this indicates the presence of underlying or residual enchondromas, and correlates with a type 2 radiographic lesion.
- In the third MRI pattern, the vast majority of the lesion has a relatively reduced signal intensity compared with typical chondral tissue and a diagnosis of an underlying chondral component could not be made on MRI. In this case, smaller areas of hyperintensity on T2-weighted images could be seen within the main tumor mass. These presumably correspond to tumor necrosis rather than chondral tissue, and correlate with a type 3 radiographic lesion.⁸

CONCLUSION

In my study the major points to remember are:

1. Maffuccis syndrome is a rare disorder presenting with swelling/ pain secondary to malignancy or fracture .
2. Chondrosarcoma is the most common malignancy that secondarily affects the enchondromas occurring in Maffucci's syndrome.
3. Golden point is that Dedifferentiated chondrosarcoma can be diagnosed on MRI by a spectrum of signal intensities on T2WI/FLAIR. We can almost reach to the histo pathological diagnosis with help of MRI.

KEY MESSAGE

Maffucci's syndrome is a rare entity presenting with enchondroma and haemangioma in various parts.

Furthermore, secondary chondrosarcoma is a rarer complication of the disorder. MRI clinched the diagnosis of dedifferentiated type of secondary chondrosarcoma

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