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MALIGNANT TRANSFORMATION OF ENCHONDROMATOSIS TO CHONDROSARCOMA IN A CASE OF MAFFUCCI SYNDROME

Pathology

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ABSTRACT

Malignant transformation of benign bone tumors is rare. Although chondrosarcoma may develop in patients with multiple enchondromatosis, this event rarely occurs in the hand. We report a case of chondrosarcoma (CS) in a 26-year-old patient secondary to multiple enchondromatosis in bilateral hands. It is crucial to diagnose malignant change in enchondroma to plan the appropriate management, keeping in mind the possibility of local recurrence and metastasis of secondary CS.

KEYWORDS

Amputation, Chondrosarcoma, Enchondromatosis, Hand, Maffuci syndrome

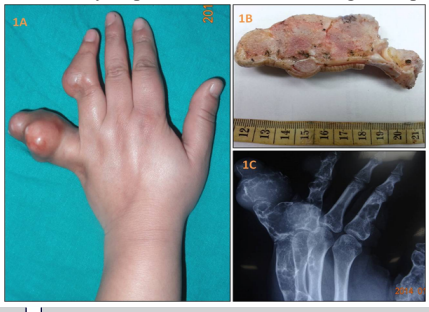
Introduction

Malignant transformation of benign bone tumors is rare, and the actual incidence has not been established. [1] Enchondromas are the most common benign cartilaginous bone tumours arising in the medullary cavity of small bones of the hand. In contrast, chondrosarcomas commonly occur in the pelvis, proximal femur and humerus.[2] Secondary CS are rarely encountered , constituting 1% of all malignant bone tumors.[1] This transformation is more commonly observed in cases of enchondromatosis than in solitary enchondroma, which is extremely rare in hands.[2] The incidence of malignant transformation in enchondromatosis is between 5% and 25% while solitary osteochondroma shows the incidence to be ranging from 0.4% to 2%.[3,4] In Maffucci syndrome, cutaneous, soft tissue, or visceral hemangiomas are found in addition to multiple enchondromas. Benign chondrogenic tumors of bone require long term follow-up to understand the clinical course and detect malignant transformation at the earliest.

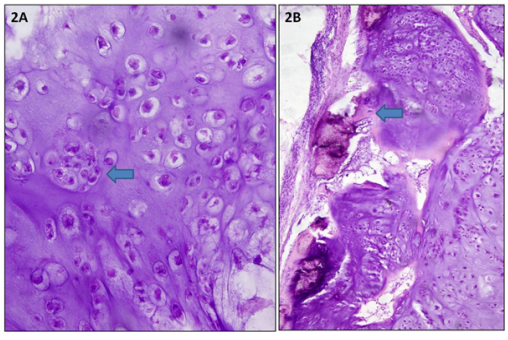
Case history

A 26-year-old female patient presented with complaints of progressively increasing multiple swellings in bilateral hands (Figure 1A). These swellings had been present since 4 years of age and patient experienced no spontaneous pain or tenderness, which explains her delayed presentation to the clinic. She also complained of swelling in the right foot. She had no remarkable past and family history.

On local examination, the ring and little finger showed multiple large swellings leading to deformity of hand. The swelling measured 10 cm in little finger, non-tender, hard in consistency and fixed to the underlying bone. Overlying skin was stretched out and intact throughout. Radiography showed a lytic lesion in the 5th phalangeal bone, causing erosion of bone along with cortical destruction and soft tissue extension (Figure 1B). Because these clinical and radiological findings strongly suggested chondrosarcoma secondary to multiple enchondromatosis, ray amputation of the left little finger was performed.



Grossly the specimen showed a globular mass measuring 10X6X5cm involving the phalanx of 5th left finger. Cut section was white, semilucent and showed lobulated architecture, with destruction of the cortical bone (Figure 1C). Histologically, the tumour was cellular and lobulated with chondrogenic cells dispersed in abundant myxoid background. Occasional clustering of the tumour cells was seen with mild nuclear pleomorphism and binucleated cells (Figure 2A). No significant mitosis was identified. The tumor was breaching the cortex at multiple foci and extending till deeper dermis (Figure 2B). In a focal area at the periphery of the tumour, a classic enchondromatous lesion with a preserved fibrous capsule was evident. A final diagnosis of chondrosarcoma (grade 1) with myxoid areas, arising in a pre-existing enchondromatosis, was made.



Discussion

It is well known fact that the secondary chondrosarcoma may arise in antecedent cartilaginous lesions, like solitary osteochondroma, multiple hereditary exostosis and multiple enchondromatosis.[5] Although enchondromatosis most commonly affect the metacarpal and phalanges, CS secondary to multiple enchondromatosis primarily affects the pelvis, shoulder girdle, distal femur and proximal tibia [1] There have been only a few cases of CS of the hand secondary to multiple enchondromatosis reported in the English literature (Table 1)[5-8] However, in a study of 18 cases of CS of the hand, Palmieri[7] reported that four were the result of enchondromatosis. Martínez Villén *et al* reported a 53-year-old patient with secondary CS in the ring and little fingers.[8] The average age of the secondary CS patient is 35 years, which is younger than patients with primary tumors. [1]

Table 1: Reported Cases of Chondrosarcoma of the Hand Resulting from EnchondromatosisYearAuthorGender/AgeSitePainDisease progression (y)TreatmentFollow up1977Block and Burton[6] 25/MRing, little fingerNo7Ray amp7 Mo NR1984 Palmieri[7] 73/MThumbYesUnknownExcision8 y NR 81/FIndex Yes Unknown

Ray amp2 y NR 62/MRingNoUnknownRay amp4 y R 59/MIndexYesUnknownRay amp3 y NR2003 Goto et al[5]27/MLittle No10Ray amp6 mo NR 76/MRingNo2Amputation1 y NR2007Altay et al[1]24/MUnknown fingerUnknown10Ray amp14 mo NR2004 Martínez Villén et al[8]53/FRing, littleNo28Ray amp12 mo NR2010 Muramatsu et al[2]78/MRing, littleNo60Ray amp1 y NR**2017Present study26/FRing, littleNoUnknownRay amp1 yr NR**R: recurrence; Ray amp: ray amputation; NR: no recurrence; mo: month

The rate of CS in conjunction with Olliers disease is estimated to be 20 to 50%. [2] Similar to Olliers disease, Maffuci syndrome may show malignant transformation and the risk appears to be even higher than Olliers. [1] Altay *et al* reported one of three patients with Maffucci's syndrome developing secondary CS. [1] With Maffucci syndrome, cutaneous, soft tissue, or visceral hemangiomas are found in addition to multiple enchondromas. [1] In the present case, hemangiomas of the lower extremities were noted in addition to enchondromas.

The signs of malignant transformation of a benign enchondroma are increase in size of the lesion, onset of pain or tenderness, and cortical destruction associated with soft-tissue invasion. [5] In the present case, there was no pain or tenderness though there was increase in size.

The distinction between low grade CS and enchondroma on histology is very subtle and histological approach was proposed by Mirra *et al* based-on tissue patterns. [9] The important histological features of chondrosarcoma consist of a myxoid stroma and permeation of bone through which the tumor fills the marrow cavity and frequently entraps pre-existing bony trabeculae. Ogose *et al* also proposed histopathological criteria for the diagnosis of CS, including hypercellularity, double-nucleated cells, myxoid change, necrosis, permeation, and soft tissue extension.[10] The distinction is of therapeutic importance, because an enchondroma may respond to local curettage but a chondrosarcoma needs wide excision or amputation.[5] In the absence of documentation of pre-existing enchondroma on

Year	Author	Gender/ Age	Site	Pain	Disease progression (y)	Treatment	Follow up
1977	Block and Burton6	25/M	Ring, little finger	No	7	Ray amp	7 Mo NR
1984	Palmieri7	73/M	Thumb	Yes	Unknown	Excision	8 y NR
		81/F	Index	Yes	Unknown	Ray amp	2 y NR
		62/M	Ring	No	Unknown	Ray amp	4 y R
		59/M	Index	Yes	Unknown	Ray amp	3 y NR
2003	Goto et al5	27/M	Little	No	10	Ray amp	6 mo NR
		76/M	Ring	No	2	Amputation	1 y NR
2007	Altay et al1	24/M	Unknown finger	Unknown	10	Ray amp	14 mo NR
2004	Martínez Villén et al8	53/F	Ring, little	No	28	Ray amp	12 mo NR
2010	Muramatsu et al2	78/M	Ring, little	No	60	Ray amp	1 y NR
2017	**Present study**	**26/F**	**Ring, little**	**No**	**Unknown**	**Ray amp**	**1 yr NR**

biopsy, the diagnosis of secondary CS relies on long medical history and radiologic changes of pre-existing lesion.

Despite the low grade of secondary CS in enchondromatosis, it exhibits local invasion, recurrence or distant metastasis, most commonly to the lung. [2] As CS is radioresistant and chemoresistant, surgery is the only form of treatment. [2,7-8]Roberts and Price reported 19 patients with CS of the proximal phalanges or metacarpals of the hand. [6] Amputation of the ray or digit was curative in 13 cases. In contrast, recurrence was observed in four cases which underwent local curettage. Because wide excision of the tumour with finger salvage is impractical in the hand, amputation or disarticulation of the digit or ray is the choice of surgery, which provides a reasonably functional and cosmetically acceptable hand.[7] Ahmed *et al* reported that patients with secondary CS who undergo no treatment or inadequate treatment die within ten years after diagnosis. [3]

Although the possibility of malignant transformation in benign chondrogenic tumors is rare, it is encountered in cases of multiple enchondromatosis particularly if associated with a syndrome like Maffuci or Olliers disease. Curettage of enchondromas at an early age is recommended to prevent malignant change. Early recognition and prompt surgical intervention of secondary CS is necessary for successful clinical outcome.

Acknowledgement: None

Figure Legends:

Figure 1a: Swelling in the fourth & fifth digit. 1b: Globular mass in the phalanx of left fifth digit. Cut surface lobulated, white, semi lucent.1c: X ray: (Posteroanterior view of the hand) Lytic lesion in the 5th phalangeal bone, with cortical destruction.

Figure 2a: Grade 1 chondrosarcoma: moderately hypercellular lesion showing cartilaginous matrix with plump crowded (arrow), hyperchromatic tumour cells (x200, H&E) 2b: Permeation of bony trabeculae by tumor cells (arrow) (x100, H&E)

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