Maffucci's syndrome combined with dedifferentiated chondrosarcoma

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Summary. We report a rare case of Maffucci’s syndrome combined with dedifferentiated chondrosarcoma in the right shoulder girdle developing from pre-existing enchondroma. In this case, magnetic resonance imaging was useful in diagnosing dedifferentiated chondrosarcoma before surgery. T2-weighted imaging was used to distinguish between the cartilaginous component and the dedifferentiated one. Histologically, there was enchondroma in the humerus and grade 2 chondrosarcoma in the scapula. Further, the dedifferentiated tumor had three mesenchymal elements: osteosarcoma, malignant fibrous histiocytoma, and fibrosarcoma. This histological heterogeneity may be due to mesodermal dysplasia of Maffucci’s syndrome.

Maffucci’s syndrome, a rare, congenital, nonhereditary mesodermal dysplasia, is characterized by multiple enchondromas and soft tissue hemangiomas. The incidence of malignant tumors associated with the disease is high, with more than 50% of these being malignant transformations of enchondromas to chondrosarcomas [11]. This report details the case of a 72-year-old woman diagnosed as having Maffucci’s syndrome combined with dedifferentiated chondrosarcoma in the right shoulder girdle developing from pre-existing enchondroma.

Dedifferentiated chondrosarcoma was first fully described in 1971 by Dahlin and Beabout [5]; however, dedifferentiated chondrosarcoma associated with multiple enchondromas is rare. Three dedifferentiated chondrosarcomas occurring in patients with multiple enchondromas (Ollier’s disease) were described by Dahlin and Unni [6]. To our knowledge, dedifferentiated chondrosarcoma arising in an enchondroma in Maffucci’s syndrome has not previously been reported.

In this case, magnetic resonance imaging (MRI) was used to distinguish between the cartilagenous component and the dedifferentiated one. Histologically, the dedifferentiated lesion had three elements: osteosarcoma, malignant fibrous histiocytoma, and fibrosarcoma.

A 72-year-old woman presented with a 6-month history of right shoulder pain. During the first 4 months, she was treated conservatively at Shinminato City Hospital for periarthritis of right shoulder joint. At that time, radiographs showed no evidence of any malignant change in a pre-existing enchondroma (Fig. 1a). Thirty years previously her right small finger had been amputated because of recurrent enchondroma.

A diagnosis of Maffucci’s syndrome was made based on the presence of multiple enchondromas in the phalanges, metacarpals, radius, ulna, humerus, and scapula of the right upper extremity and multiple subcutaneous hemangiomas in the right hand and elbow (Fig. 1b). The woman had had no history of malignancy prior to this time.

A physical examination showed remarkable swelling of the right shoulder and severe limitation of the range of motion of the shoulder joint. The skin had a slightly erythematous change, and the mass, located in the posterior lateral aspect of the shoulder, was firm, immobile, and tender to palpation.

Radiographs of the right shoulder demonstrated an expansile, destructive lesion involving the humerus and scapula (Fig. 2). A pulmonary metastasis was observed in the radiograph of the chest. A radionuclide bone scan showed markedly increased uptake in the scapula, humerus, and soft tissue component of the shoulder girdle tumor; however, the bone scintigram did not permit specific location of the uptake. No other sites demonstrated abnormal uptake. Computed axial tomography (CAT) showed a low density area and spotty calcification in the scapula and humerus, but did not allow distinction between extraosseous extension of the lesion and normal muscle.

Magnetic resonance imaging (MRI) demonstrated the existence of two components in the tumor. The tests were performed with a 1.5-tesla system (Signa; General Electric Medical Systems) with the right upper extremity placed in a surface coil. On the T1-weighted images [repetition time (TR) = 500 ms; echo time (TE) = 20 ms] by the spin echo method, both the intraosseous lesions in the humerus and scapula and the extraosseous lesion posterior to the shoulder girdle showed low signal intensity similar to normal muscles. However, the T2-weighted image (TR = 2500 ms; TE = 80 ms) demonstrated increased signal intensity in the lesion of the humerus and scapula compared to normal fatty marrow. The intraosseous tumor was arranged in a lobular pattern. Further, the signal intensity of the scapular lesion was higher than that of the humeral lesion. The extraosseous lesion showed even and low signal intensity relative to the intraosseous lesion in the T2-weighted image. Partial discontinuity of the cortex of the humerus and scapula was also observed (Fig. 3).

After complete clinical evaluation, the patient’s condition was diagnosed as malignant transformation of enchondroma to chondrosarcoma with dedifferentiated components. Wide excision was performed according to the Tikhoff-Linberg procedure [12] to relieve the severe pain. Reconstruction was achieved using a cement-
ed Neer prosthesis (Fig. 4). Four weeks after surgery, the patient died of respiratory failure due to multiple pulmonary metastases. Pain relief as a result of surgery was satisfactory.

**Pathological findings**

The intraosseous lesion was a semitranslucent, partially calcified and lobulated, cartilaginous tumor. A small area of cortical destruction was seen at the dorsal aspect of the humerus and scapula. A gray, fleshy, anaplastic tumor abutted the cartilagenous component. Enchondromas were present in the humerus, high grade chondrosarcoma in the scapula, and a highly malignant spindle cell sarcoma in the extraosseous lesion (Fig. 5). Using the classification system described by Evans et al. [8], the condrosarcoma in the scapula was grade 3 as more than two mitoses were observed in each high power field. There was also a small of enchondroma in the lower focus edge of the lesion in the scapula.

Closer examination revealed that the dedifferentiated lesion had three mesenchymal elements, consisting of osteosarcoma, fibrosarcoma, and malignant fibrous histiocytoma (Fig. 6). Small foci of malignant osteoid formation seen were near the edge of enchondroma in the humerus. Most of the fibrogenic area showed a “storniform” or “herring-bone” pattern. There were histiocytic cells which showed grooving or indentation of nuclei in a small area of this fibrogenic tumor. The dedifferentiated tumor was classified as a fibroblastic osteosarcoma according to the definition by Dahlin and Unni [6].

The pulmonary metastatic lesions were found to be malignant spindle cell sarcoma. There was no evidence of osteoid production by the spindle cells.

**Discussion**

The syndrome of enchondromatosis with hemangiomatisos was first described by Maffucci in 1881 [13]. In 1942, Carleton et al. [4] reviewed 20 cases of this syndrome and named it Maffucci’s syndrome. Lewis and