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Multiple Enchondromatosis (Ollier's Disease)

Geon-Kook Lee, Jung-Ran Kim, Je G. Chi and Eui-Keun Ham

Department of Pathology, College of Medicine, Seoul National University, Seoul 110-744, Korea

=Abstract=Enchondromatosis is a rare disease which accounts of 0.46% of all bone tumors. This report deals with pathologic analysis of 3 cases which were tudied at Department of Pathology for the last 5 years, 1983-1988. There were two females aged 19 years and 32 years and one 17-year-old male. Family history was negative in all cases. Hand bones were most commonly involved. One of three cases had unilarteral distribution.

Clinically they presented with painless mass with or without deformity. Radiologically multiple non-ossifying expansile radiolucent lesions were characteristic. Histologically, they were mildly or moderately cellular cartilaginous masses multilobulated by fibrovascular septa. Occasionally, in the highly cellular areas, there were foci of nuclear pleomorphism, occasional binucleated cells and bizarre nuclei. In one case mosaic ring calcifications were noted. There was no evidence of malignancy in all cases.

Key words: ***Multiple enchondromatosis, Ollier's disease, Bone tumor***

INTRODUCTION

The enchondrom is not uncommon tumor arising primarily inside the bone and is distinguished from ecchondroma or osteochondroma which are exophytic in growth pattern. They occur most commonly as a solitary lesion (Takigawa 1971) but rarely multiple sites are simultaneously involved and have been called as multiple enchondromas, enchondromatosis or Ollier's disease (Dahlin and Unni 1886; Spjut *et al.* 1971; Mirra 1980; Resnick and Niwayama 1988). Incidence of this disease was estimated to be 0.46% of all bone tumors (Dahlin and Unni 1986).

This tumor draws attention in two aspects. One is the risk of malignant transformtion, which is high and estimated about 50% (Spjut *et al.* 1971). Another is frequently associated deformity of bones that are affected by this condition.

Reviewing Korean literatures we could find only one case reported on enchondromatosis (Lee *et al.* 1971). This report dealed chiefly of radiologic findings without detailed pathologic description. The purpose of this paper is to report 3 cases of enchondromatosis and to discuss on the histopathological findings.

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CASE REPORT

CASE 1: The patient was a 19 year-old female presented with painless tumor mass in the left second finger which was found 4-5 years ago. Physical examination was negative except the painless and non-tender mass in the base of the left seond finger. Range of motion of the finger was slightly linmited. Family history was not contributory.

Roentgenogram revealed multiple expansile osteolytic lesions with thin sclerotic margin and cortical erosion in multiple bones of the left side; phalangeal bones of the first, second and third fingers, tne second metacarpal bone, humerus, scapula, the third costochondral junction and proximal phalanx of the first toe. Under the tentative diagnosis of enchondromatosis, curettage and bone grafting was performed in the left second metacarpal bone.

Histopathologically, lobulated cartilage masses were divided by fibrovascular tissue of varying thickness, containing very elongated ramifying vascular channels wih prominent endothelial cells. Macrophages were often seen in collection in areas where perichondrium-like tissue was thickened with telangiectatic vessels. Some of these ovoid cells appeared to be synovial cells and separated the cartilage tissue into several lobules. In areas this tissue blended into proliferting cartilage. Basophilic cartilage matrix varied considerably in amount by different areas. Acellular area alternated with cellular area where chondrocyte of dense eosinophilic cytoplasm were crowded, being encircled by clear lacunae. The nuclei were generally small and monomorphous without nucleoli or mitotic figures. Occasional double nucleated cells were seen. Foci of myxoid change with cells floating in the matrix were seen, where the cells appeared degenerated. In portions the cartilage mass was closely apposed to the woven bone but did not destroy it.

CASE 2: A 17-year-old male presented with shortening and bowing deformity of both hands which developed in early childhood. Physical examination was negative except masses with deformities of fingers of both hands. Family history was negative.

Radiologically, both metacarpal and phalangeal bones showed multiple radiolucent medullary lesions with multilobulated appearance and scattered spotty calcifications. The cortices of these bones were thinned. Destruction and deformities of these bones of both hands were prominent. Curettage and bone grafting were done.

Histopathologically, the tumor masses were found in the medullary cavity, leaving cortical bone and periosteum intact. However, intramedullary expansile masses abutted on the inner table of the cortical bone, showing focal destruction and absorption. There was no bone island entrapped by the cartilage mass. The tumor masses were lobulated. The interlobular element was composed of dense eosinophilic amorphous material rather than firous tissue. Scattered foci of irregular calcification were seen inside and around the masses. The tumor cellularity was mild to mederate, varying slightly in size and shape of the proliferating chondrocyte. The cytoplsm of tumor cells was characteristically eosinophilic. The cells were often stellate and surrounded by ovoid lacunae. The cellularity varied in areas, but plemorphism was not seen. Lacunae with more than one nucleus were occasionally seen.

CASE 3: A 32-year-old female presented with slowly growing mass and discomfort in the left lower calf which developed 7 years ago. Physical examination was negative except non-tender mass and bowing deformity of the left lower leg and masses and deformities of both hands. Family history was not contributiory.

Roentgenogram revealed an irregular oseolytic area with lateral bowing of the left distal tibia and severe deformities and multiple osteolytic lesions of the bones of both hands. Curettage and bone grafting were done in the left distal tibia.

Histopathologically, the tumor masses were fragmented but showed rounded contour in portions. In rounded area thin collagenous connective tissue surrounded proliferting cartilage masses showing densely packed basophilic matrix and scattered chondrocytes. Punctate calcifications were seen abruptly in the tumor masses without association with myxoid degeneration. The tumor cells were fairly uniform in size and shape but binucleated cells were often seen. The cellularity, however, was mild to moderate and varied considerably in areas. A large areas of acellular region were seen in some fragments. There was a focus where the tumor masses were apposed to the trabecular bone with smooth margin.

DISCUSSION

Enchondromatosis is a rare developmental abnormality that presents with ectopic growth of non-ossifying cartilage in the metaphysis and diaphysis of various bones. It represents a dyschondroplasia of bone characterized by failure of normal endochondral ossification, with the production of tumefactive cartilaginous masses (Dahlin and Unni 1986; Spjut *et al.* 1971; Mirra 1980; Resnick and Niwayama 1988). The terms, Ollier's disease and multiple enchondromatosis, have been used synonymously, although Ollier's disease has been applied to the cases, which have strictly unilateral pattern. However, if the phlanges of the hand are involved, there fre-

quently are a few enchondromas in the opposite hand, particularly in the first and fifth digits (Spjut *et al.* 1971). In our cases, hand bones were involved in all cases, long bone in two cases, and flat bone in one case. In two cases bones of both hands were affected. In case 1 unilateral tendency was evident.

In our cases, pathologic specimens which were obtained by excision and curettage were small masses or fragments of lobulated, pinkish blue, semitranslucent, and hard cartilaginous tissue. Covering cortical bone was found in case 2 and minute calcifications were indentified grossly in case 3. Histologically, they were characterized by midly or moderately cellular hyaline cartilages multilobulated by fibrovascular septa and maintained the features of norml cartilage. In case 1 and 2 loose areas were scattered and the hyaline cartilage was not well formed in portions. The chondrocytes constituting the cellular component were small and lay in lacunar spaces. They had round or ovoid, fairly regular inconspicuous nuclei and eosinophilic cytoplasm. Adjacent to the cartilage, some of mesenchyml cells showed chondrocytic differentiation. Occasionally, especially in the highly cellular area, spindle shaped cells and/or slight nuclear pleomorphism were present. There were few bizarre nuclei and a few binucleated cells in portions. In case 2, mosaic patterned ring calcifications were noted in some cartilage lobules. Dense calcifications were prominent findings in case 3.

When long bones are involved in enchondromatosis, severe deformities and stunting of growth are frequent. In our cases, lateral bowing of tibia in case 3 is the only demonstrable deformity. There is a tendency that long bones were largely spared from the involvement. In addition it is known that there is a strikingly increased risk of transformation into chondrosarcoma from one of the chondromas (Cowan 1965). The risk is estimated to occur in about 50% with extensive involvement (Spjut *et al.* 1971). This risk is further increased with involvements of long bone and flat bone rather than involvement of short bones of hands and feet. Differention of enchondroma and multiple enchondromatiosis from well differentiated Grade I chondrosarcoma is therefore an important issue. Pattern of islands of cartilage, enchondroma encase pattern, permeation, infiltration of Haversian system, or bands of fibrosis are some of the differentiating points (Mirra *et al.* 1985). No evidence of malignancy could be found in these cases.

In all cases family history was not contributory. and the symptoms varied; painless mass, or deformity of digit or extremity.

Roentgenographic findings were characteristic; multiple, non-ossifying, radiolucent, expansile, and intramedullary masses in long bones with sclerotic margin and mild or severe cortical erosion. There was no calcification radiologically identifiable, and no extraosseous lesion in all cases. Frequent involvement of small long bones of hands and feet was another outstanding feature.

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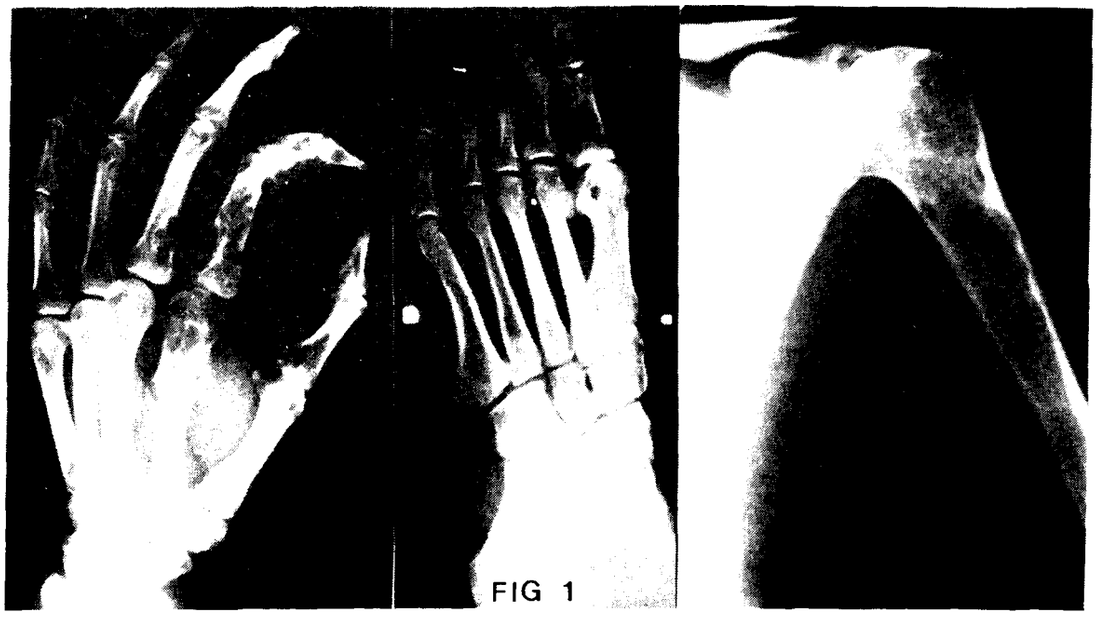

FIG 1

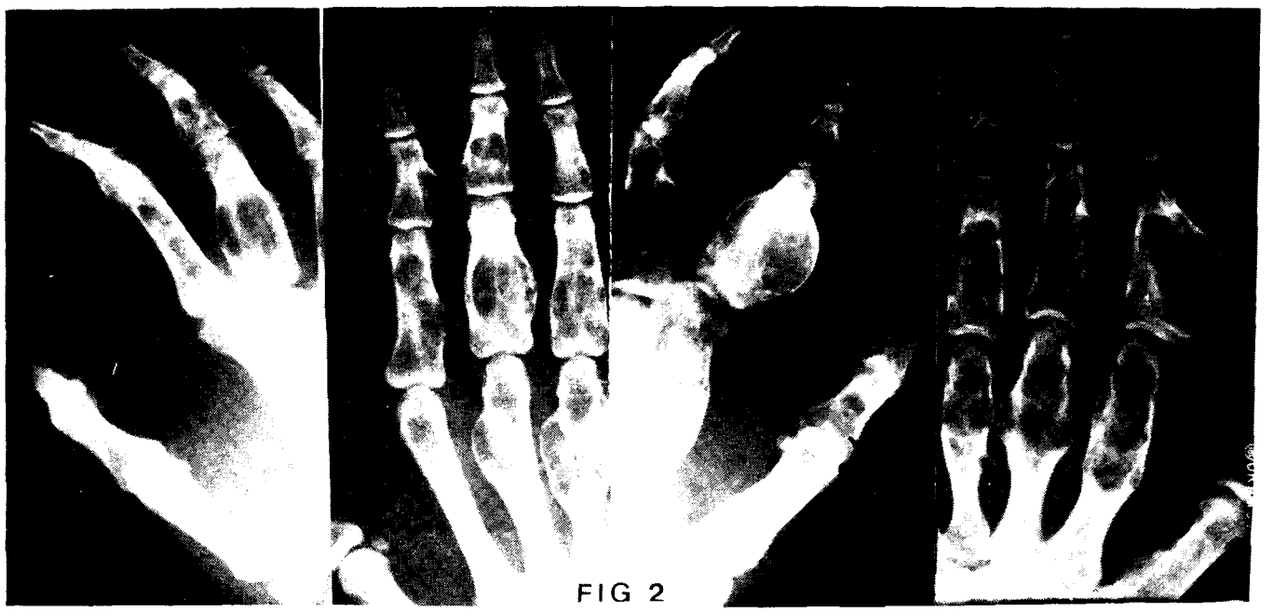

FIG 2

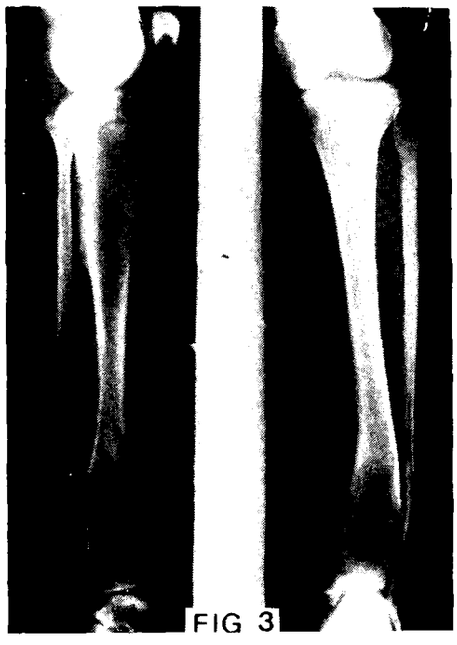

FIG 3

Fig. 1. Multiple radiolucent intramedullary lesions of bone of the left side; the first, second and third phalnages, the second metacarpal bone, proximal phalanx of second toe, and humerus (Case 1).

Fig. 2. Expansile radiolucent masses of both hands. Severe deformity of the left hand is prominent (Case 2). In case 3, involvements of both hands are similar.

Fig. 3. Osteolytic lesions in the right proximl and distal tibia and fibula. Lateral sided bowing deformity is conspicuous (Case 3).

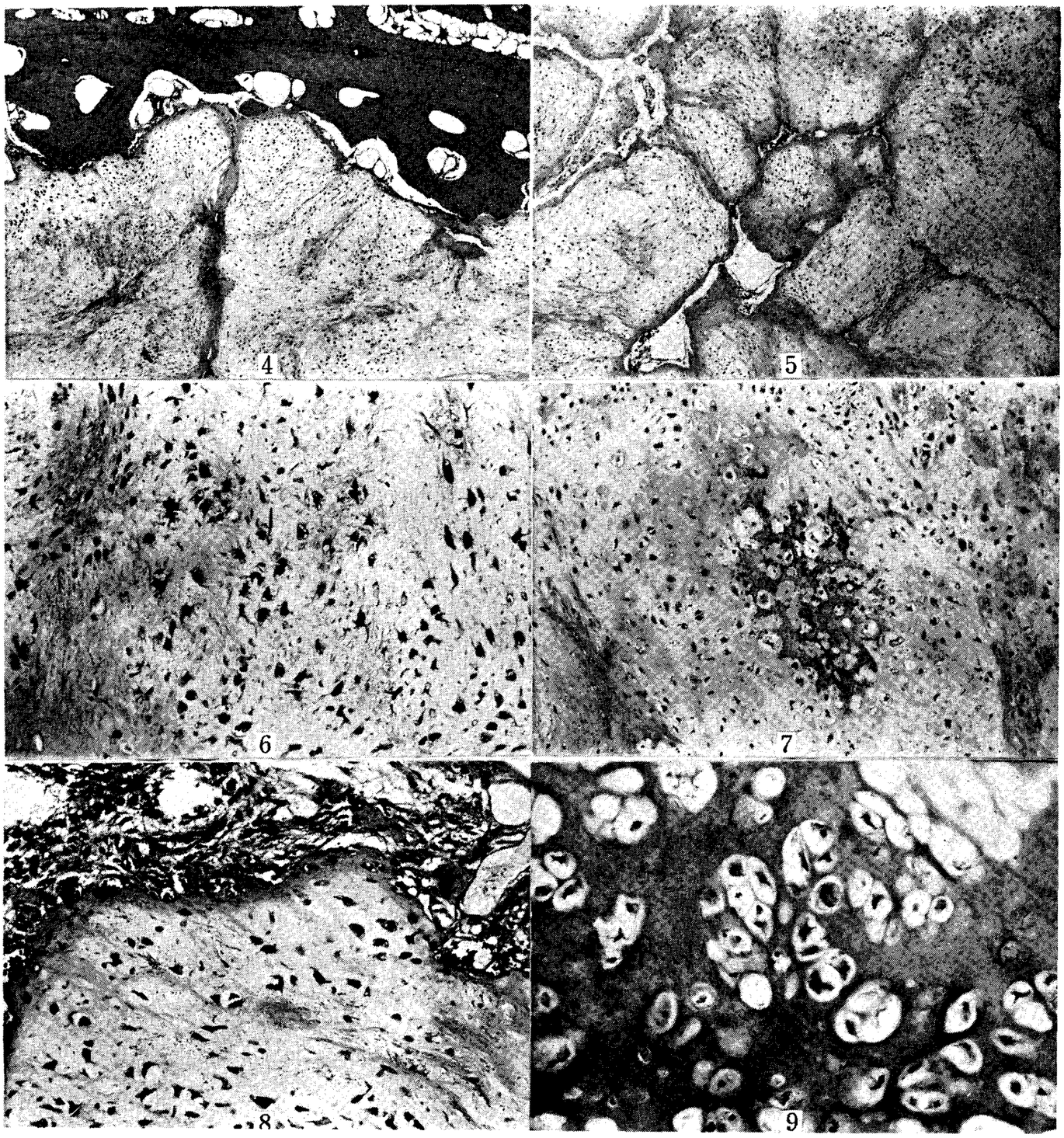

Fig. 4. Cartilaginous mass is closely apposed to the cortical bone. Well formed cartilage islands alternate with loose myxoid areas (Case 2).

Fig. 5. Typical histologic appearance of enchondromatosis. Each island of mildly or moderately cellular cartilage is margined by eosinophilic amorphous material or fibrovascular septa. In central portions of cartilage lobules loose myxoid ares are present (Case 2).

Fig. 6. In more cellular area, nuclear pleomorphism is evident. A few binculeated cells are also noted. Stellate appearance of chondrocyte is prominent (Case 2).

Fig. 7. Characteristic ring calcification is noted (Case 2).

Fig. 8. At the periphery of the cartilage lobule, mesenchymal cells, which appear synovial cell, show chondrocytic differentiation. These cells blend into the cartilage lobules (Case 1).

Fig. 9. Binucleated cells and lacunae with more than one nuclei are demonstrated (Case 1).

＝국문초록＝

다발성 내연골종증(올리어 병)
(3 증례보고)

서울대학교 의과대학 병리학교실

이건국 · 김정란 · 지제근 · 함의근

다발성 내연골종증은 드문 질환으로 전체 골 종양의 0.46%, 양성 골 종양의 2%를 차지한다. 국내 문헌들 중에는 이 질환에 대한 기술이 오직 1예로서, 특히 병리학적 소견에 대한 기술은 없는 상태이다. 저자들은 최근 3예의 다발성 내연골종증을 경험하고 이들 3예에 대한 병리학적 관찰 결과를 보고 한다.

두 예는 여성이고 한 예는 남성이며, 두 예는 10대이고 한 예는 30대였다. 가족력은 모든 예에서 없었으며, 수지골의 이환이 잦았다. 두 예에서 양측 수지골의 이환이 관찰되었고 한 예에서는 올리어의 보고처럼 일측성의 이환이 현저했다.

방사선 소견은 매우 특징적으로, 다발성의 골화되지 않는 방사선 투과성의 팽창성 골수질내 병변들이 피질골들을 침식하면서 경화성 가장자리를 갖고 있었다.

조직학적으로 모든 예들은 혈관성 섬유질에 의해 여러 분엽으로 나뉘는 희박한 혹은 중등도의 세포 밀집도를 보이는 연골 종양의 형태를 가졌다. 특히 세포밀집도가 높은 부분에서는 약간의 핵 이양성과 극소수의 2핵 세포가 관찰되었고, 드물게는 진기한 핵상등이 보이기도 했다. 한 예에서 모자이크양의 환상 석회화가 관찰되었다. 다발성 내연골종증의 진단에 있어 분화도가 좋은 연골 육종과의 감별이 중요한데, 3예 모두에서 악성의 증거는 발견할 수 없었다.