Singapore Med J 2008; 49 (10) : 841

CME Article

Clinics in diagnostic imaging (124)

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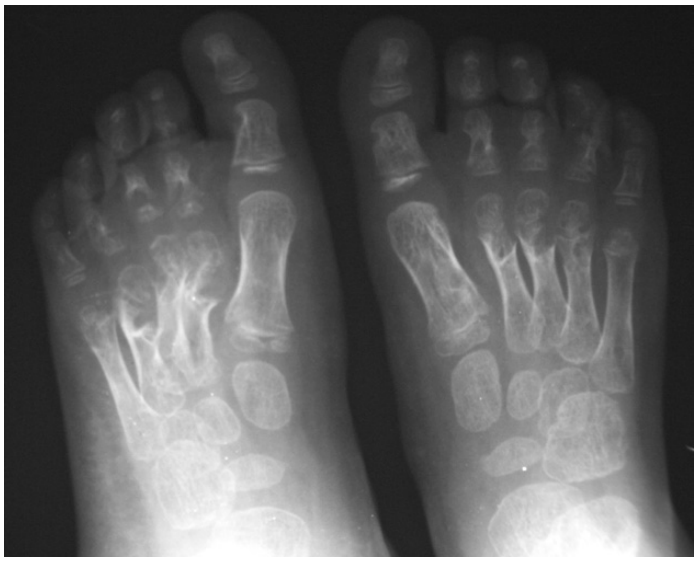

Fig. 1 Anteroposterior radiograph of both feet.

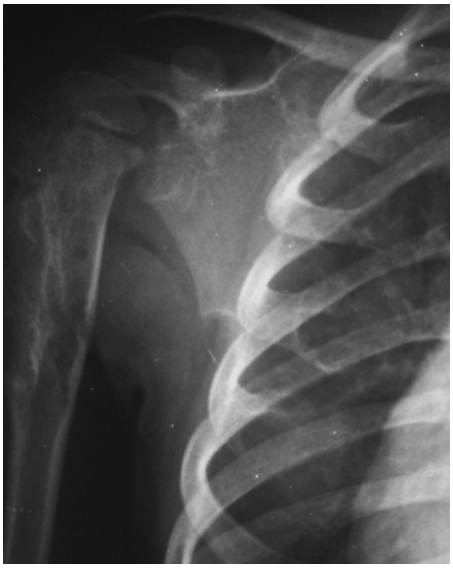

Fig. 2a Anteroposterior radiograph of the right shoulder.

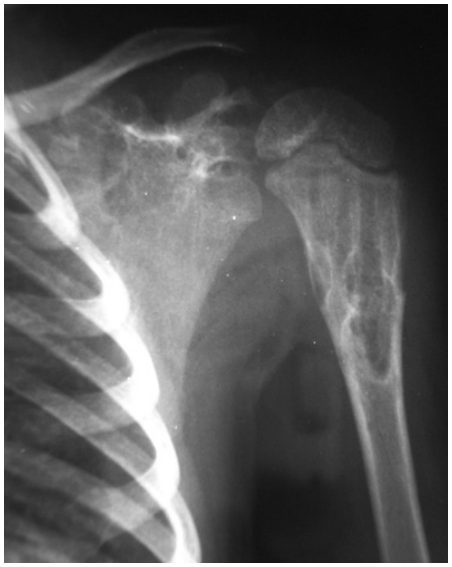

Fig. 2b Anteroposterior radiograph of the left shoulder.

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CASE PRESENTATION

A three-year-old girl was noticed by her parents to have slow-growing swellings over the left foot for the past one year. Similar painless swelling of the upper right arm was also noted. Radiographs of both feet (Fig. 1), and the right and left shoulders (Figs. 2a & b), were taken during the initial visit. What do the radiographs show? What is the diagnosis?

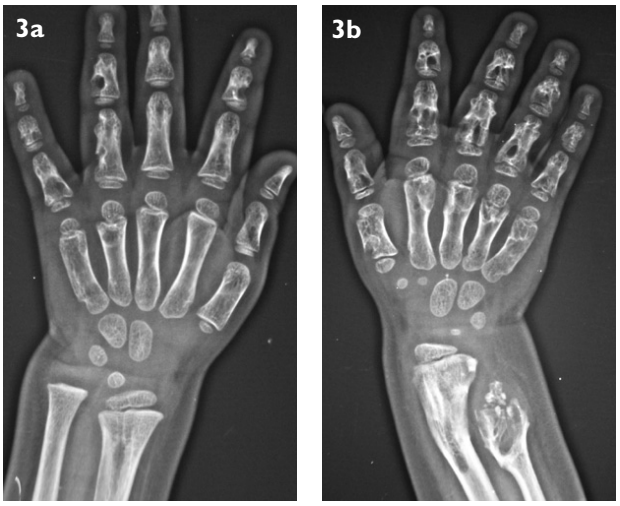

Fig. 3 Anteroposterior radiographs of the (a) left and (b) right hands show multiple well-defined osteolytic lesions with endosteal scalloping, cortical thinning, particularly at the right metacarpals and bilateral proximal and middle phalanges. There is also involvement of the right distal radius and ulna with bowing deformity.

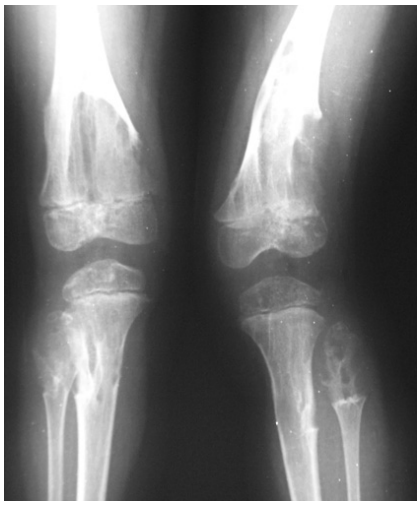

Fig. 4 Anteroposterior radiograph of the both knees shows multiple similar lesions along the metaphyses of both distal femurs, proximal tibiae and fibulae. There are channel-like radiolucent areas in the metaphysis. There is enlargement of the left distal femur. The right femur is bowed, with genu valgus deformity of the right knee.

IMAGE INTERPRETATION

Radiographs of both feet show multiple well-defined lytic lesions arising from the medullary cavity of the tubular bones, particularly at the metatarsals and proximal phalanges. These lesions show bony expansion and endosteal scalloping, with sclerotic rims (Fig. 1). The radiographs of both shoulders (Figs. 2a & b) show multiple similarly well-defined osteolytic lesions in both proximal humeri and both scapulae. Several of the partially-visualised ribs are also involved, with bony expansion. No soft tissue component or soft tissue calcification could be identified.

DIAGNOSIS

Multiple enchondromatosis in Ollier disease.

CLINICAL COURSE

Additional radiographs of the patient were also taken. These included a radiograph of the hands (Figs. 3a & b), both knees (Fig. 4) and the pelvis (Fig. 5). Multiple expanded osteolytic lesions with well-defined sclerotic margins, similar in appearance to the other enchondromas in the feet and humerus, were present. In the subsequent years, the patient sustained several spontaneous, pathological fractures requiring surgical fixation. The patient also had a short stature due to bowing of the femurs. Surgery to realign the growth of both lower limbs has been performed (Figs. 6a & b). Now 18 years of age, the patient is currently confined to a wheelchair.

DISCUSSION

Enchondromatosis, or multiple enchondromas, occur in three distinct different conditions. The most common entity is Ollier disease, a non-hereditary failure of cartilage ossification, resulting in multiple enchondromas that typically affect the metaphyseal ends of bones. It usually becomes evident before puberty and is frequently unilateral, leading to shortening of the limbs.[1] The lesions enlarge with progressive skeletal growth, becoming more evident and characteristic with time. After the cessation of normal growth, the lesions do not increase in size. In Ollier disease, the long bones are more commonly affected than the thoracic vertebrae, flat bones of the skull, and bones in the hands and feet. The lesions cause enlargement, shortening and bowing of the bones.[1] The incidence of malignant transformation has been reported to be approximately 30%–50%.[1]

Maffuci syndrome is also a non-hereditary syndrome that is rarer than Ollier disease. It is characterised by multiple enchondromatosis as well as multiple soft tissue cavernous haemangiomas, and less commonly, lymphangiomas.[2] There is also a higher risk of malignant transformation of enchondromas to sarcomas. Both Maffuci syndrome and Ollier disease are associated with an increased incidence of juvenile granulosa cell tumour of the ovary. Patients with Maffuci syndrome also have an increased incidence of malignancies other than musculoskeletal malignancies, including gliomas, gastrointestinal adenocarcinoma, pancreatic carcinomas and ovarian tumour.[2] The third condition, metachondromatosis, is a hereditary autosomal dominant transmitted trait consisting of multiple enchondromas and osteochondromas.[3] In this distinct syndrome, the multiple exostoses characteristically occur in the digits and long bones, and unlike those in hereditary multiple exostoses, point towards the joints and frequently regress spontaneously. The enchondromatosis most often affect the iliac crests and the metaphyses of certain long bones.[3] All these three conditions are characterised by multiple enchondromas, or enchondromatosis. Enchondromas are hypothesised to develop from rests of growth plate cartilage that subsequently proliferate and slowly enlarge.[4,5] Therefore, any bone formed by enchondral ossification may be affected.

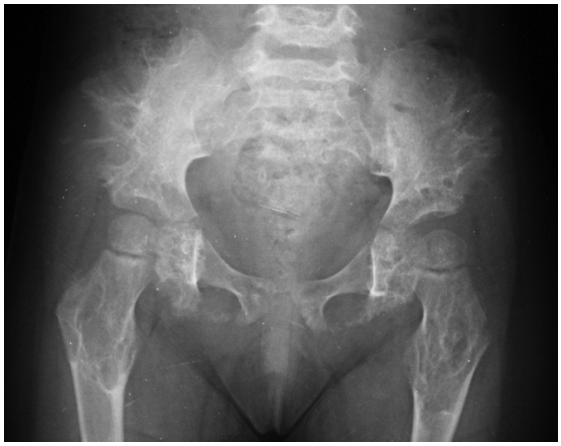

Fig. 5 Anteroposterior radiograph of the pelvis shows similar well-defined osteolytic lesions with endosteal scalloping, cortical thinning of both acetabulae, iliac wings, inferior pubic rami and intertrochanteric region of both femurs.

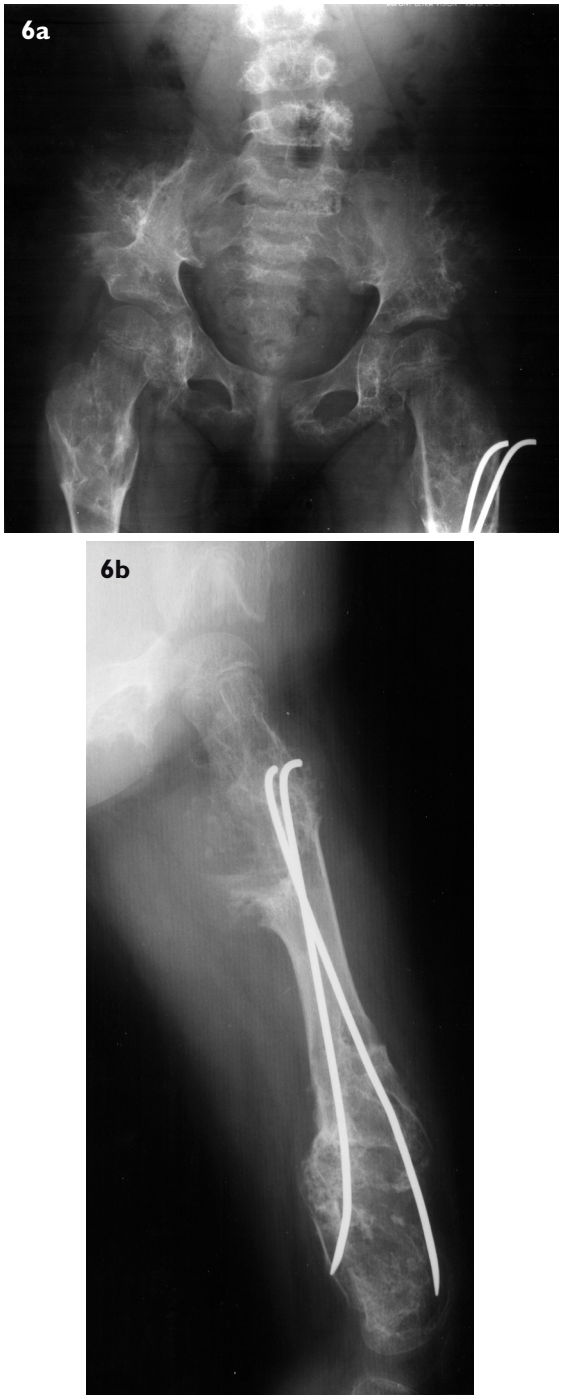

Fig. 6 Follow-up radiographs of the (a) pelvis and (b) left femur show k-wiring of the femoral shaft following corrective surgery.

Enchondroma is the most common bone tumour arising in the bones of the hand.[6,7] Approximately 40% –65% of solitary enchondromas occur in the hands, or less frequently, the feet.[8] It is usually solitary, and occurs most commonly in the small tubular bones of the wrists and hands. The proximal phalanges are the most often affected.[6,7] Less commonly involved sites are the metacarpals and middle phalanges. The ulnar three rays are more often affected than the radial two rays. Lesions of the thumb and in the distal phalanges are relatively uncommon. The carpal bones are a rare location for the lesions.[6,8] Solitary enchondromas occur in the long tubular bones in approximately 25% of cases and are more frequent in the upper extremities than in the lower extremities.[8] Tumours of the ribs, as well as some in the tubular bones, may lead to osseous expansion, designated as enchondroma protruberans. These simulate the appearance of an osteochondroma, or to massive enlargement of the bone.[8] Most patients with enchondromas present with either a painful or painless swelling.[6,7] In some patients, the enchondroma is discovered accidentally during radiographs done following trauma.[6] Enchondromas usually are central tumours that are located in the metaphysis of a long tubular bone, where they may extend into the shaft or epiphysis if the physes is closed, or in the diaphysis of a short tubular bone in the hand or foot.[8] Solitary enchondromas occur most frequently in patients between 10 and 40 years of age.[6]

The radiographs of solitary enchondromas of the hand or foot are usually characteristic.[8] Enchondromas appear as well-defined medullary lesions with lobulated contour, endosteal erosion and ground glass appearance of the matrix (Fig. 7). Expansion of bone with thinning of the cortex may also be seen (Fig. 8). Dystrophic calcifications within the matrix of small cartilage nodules or fragments of lamellar bone are often described as the "rings and arcs" or "flocculent" pattern of calcification (Fig. 9). Occasionally, the calcification is also described as "stippled" (Figs. 10a & b). In the long tubular bones, a centrally- or eccentrically-placed medullary, osteolytic tumour of variable size with or without calcification leading to lobulated erosion of the endosteal margin of the cortex, is most typical.[8] In some cases, channel-like radiolucent areas in the metaphysis are seen, although this finding is more common in Ollier disease.[8] The radiographical abnormalities accompanying an enchondroma in a flat or irregular bone may not be diagnostic.[8]

Cortical destruction or thickening, extensive and deep endosteal scalloping, cortical remodelling, periosteal

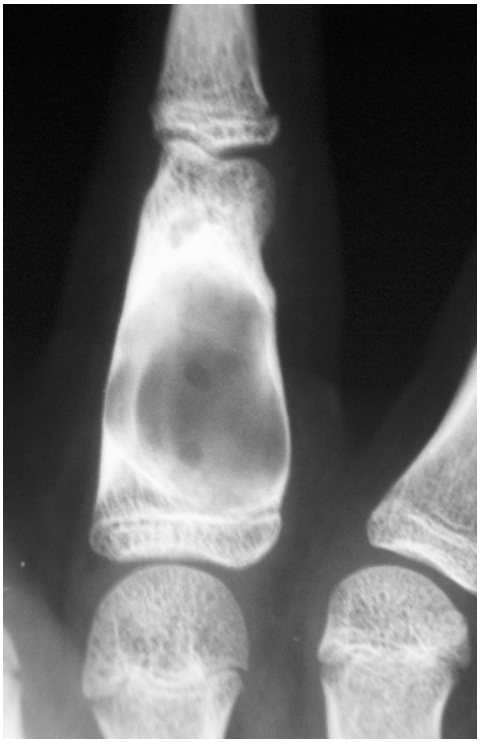

Fig. 7 Anteroposterior radiograph of the right ring finger of a child with a solitary enchondroma shows a well-defined medullary lesion with lobulated contour, endosteal erosion and ground-glass appearance of the matrix in the proximal phalanx.

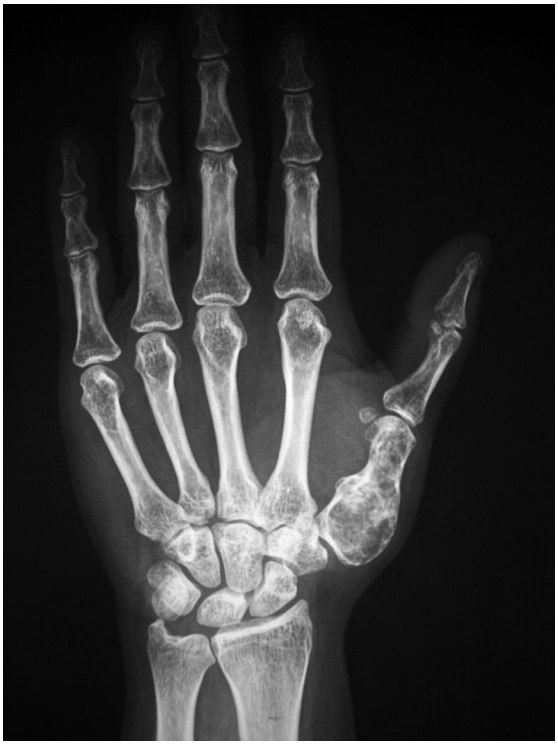

Fig. 8 Anteroposterior radiograph of the left hand of a patient with a solitary enchondroma shows an enchondroma of the first metacarpal with expansion of bone and thinning of cortex.

reaction or soft tissue masses are features suggestive of chondrosarcoma. Pathological fractures are also more common in chondrosarcomas. Enchondromas are more common in the diaphysis and distally in appendicular skeleton.[9] Computed tomography (CT) is superior to radiography in detecting matrix mineralisation as well as in the evaluation of the pattern of calcification. The lobulated margins of the lesions, degree and extent of endosteal scalloping are also better demonstrated on CT. This is particularly so in the characterisation of lesions that occur in the pelvis, or other areas with complex anatomy difficult to evaluate on radiographs.[10] On CT, attenuation values of the non-mineralised tumour components are similar in both chondrosarcomas and enchondromas, being lower than or similar to that of muscle.[9] CT is also useful in the evaluation of the size and presence of any soft tissue component which would favour chondrosarcoma as the diagnosis, although soft tissue component in enchondromas may also occur in association with a fracture and haematoma.

On magnetic resonance (MR) imaging, the non-mineralised component of enchondromas appear as low to intermediate signal intensity lesions on T1-weighted sequences, and intermediate to high signal intensity lesions on T2-weighted sequences.[9,11] Small speckled foci of high signal intensity, often evident on T1-weighted MR images, are postulated to be due to the lobular growth of enchondromas, which leaves intervening residual areas

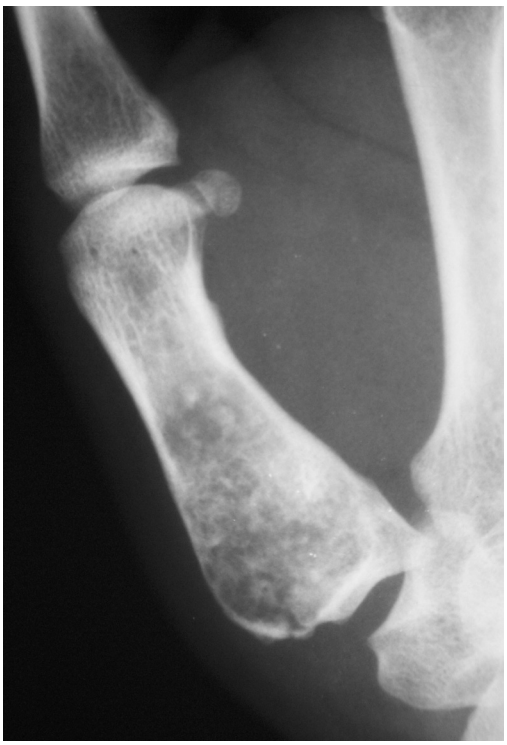

Fig. 9 Anteroposterior radiograph of the left thumb of a patient with a solitary enchondroma shows an enchondroma containing the "ring and arcs" type of matrix calcification. Incidentally, there is also a focal cortical break along the base of the first metacarpal in keeping with a pathological fracture, which may sometimes complicate an enchondroma.

of normal yellow bone marrow [4,9] Low signal intensity septa on T2-weighted MR images are also evident, corresponding pathologically to enchondral ossification or fibrous septations.[9] Following contrast administration,

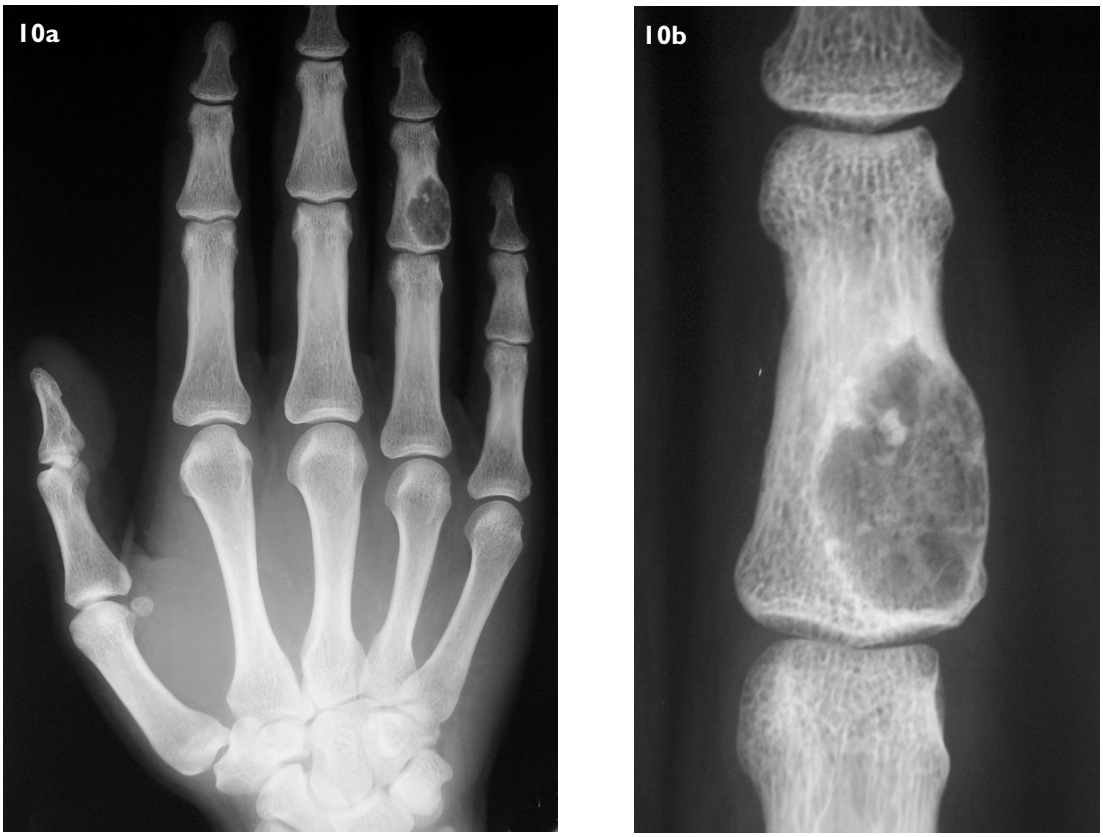

Fig. 10 Anteroposterior radiographs of the (a) right hand, with a (b) magnified view of the middle phalanx of the ring finger of a patient with a solitary enchondroma show an enchondroma with the "stippled" type of matrix calcification.

enchondromas exhibit septal and peripheral rims of enhancement. This pattern of enhancement is also seen in chondrosarcomas. Preliminary studies performed with dynamic MR imaging have suggested early enhancement of chondrosarcoma as a possible useful discriminating feature.[9]

On imaging, possible differential diagnoses include bone infarct, chondrosarcoma, epidermoid inclusion cyst, unicameral bone cyst, giant cell tumour and fibrous dysplasia.[4] Enchondromas may present with pathological fractures (Fig. 9). Malignant degeneration in long bone enchondromas and spontaneous healing may also occur.[5] Treatment of enchondromas includes intralesional curettage, and occasional filling of the bone cavity with bone chips and bone grafts.[7] Enchondromas may recur if nodules of tumour are left behind.[5]

ABSTRACT

A three-year-old girl presented with slow-growing swellings at the left foot and upper right humerus. Radiographs show multiple enchondromas in both feet, proximal humeri and scapulae, as well as at multiple sites in the hands, distal forearm bones and pelvis, in keeping with multiple enchondromatosis in Ollier disease. The clinical presentation and imaging features of enchondromas and the different types of enchondromatosis are discussed.

Keywords: benign hyaline tumour, chondroma, enchondroma, enchondromatosis, Ollier disease

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SINGAPORE MEDICAL COUNCIL CATEGORY 3B CME PROGRAMME
Multiple Choice Questions (Code SMJ 200810B)

	True	False
Question 1. The following statements regarding Ollier disease are true:		
(a) It is the most common type of enchondromatosis.	☐	☐
(b) It is a hereditary condition.	☐	☐
(c) It frequently leads to limb shortening.	☐	☐
(d) It is associated with cavernous haemangiomas.	☐	☐
Question 2. Regarding enchondromatosis:		
(a) Both Maffucci syndrome and Ollier disease are associated with an increased incidence of juvenile granulosa cell tumour of the ovary.	☐	☐
(b) Maffucci syndrome is associated with lymphangiomas.	☐	☐
(c) Metachondromatosis is a hereditary condition transmitted as an autosomal recessive trait.	☐	☐
(d) Metachondromatosis is characterised by multiple exostoses that frequently regress spontaneously.	☐	☐
Question 3. Solitary enchondroma:		
(a) Can occur in any bone formed by enchondral ossification.	☐	☐
(b) Is the second most common bone tumour arising in the bones of the hand.	☐	☐
(c) Undergo malignant transformation in 50% of cases.	☐	☐
(d) Commonly occurs in the carpal bones.	☐	☐
Question 4. Regarding the appearances of enchondromas on radiographs:		
(a) Radiographical features of solitary enchondromas in the hands and feet are usually diagnostic.	☐	☐
(b) They are ill-defined with a lobulated contour.	☐	☐
(c) Dystrophic calcification due to central necrosis can be seen within the tumour.	☐	☐
(d) Channel-like radiolucent areas in the metaphysis can be seen, more commonly in Ollier disease.	☐	☐
Question 5. The following statements regarding imaging of enchondromas are true:		
(a) Pathological fractures indicate malignant transformation to chondrosarcomas.	☐	☐
(b) Cortical destruction or thickening, extensive and deep endosteal scalloping and associated soft tissue masses suggest malignant transformation.	☐	☐
(c) On magnetic resonance (MR) imaging, enchondromas appear as intermediate to high signal intensity lesions on T1-weighted sequences, and low to intermediate signal intensity lesions on T2-weighted sequences.	☐	☐
(d) A possible differential diagnosis on radiographs would be bone infarct.	☐	☐

Doctor's particulars:

Name in full: _____

MCR number: _____ Specialty: _____

Email address: _____

SUBMISSION INSTRUCTIONS:
(1) Log on at the SMJ website: http://www.sma.org.sg/cme/smj and select the appropriate set of questions. (2) Select your answers and provide your name, email address and MCR number. Click on "Submit answers" to submit.

RESULTS:
(1) Answers will be published in the SMJ December 2008 issue. (2) The MCR numbers of successful candidates will be posted online at www.sma.org.sg/cme/smj by 15 December 2008. (3) All online submissions will receive an automatic email acknowledgment. (4) Passing mark is 60%. No mark will be deducted for incorrect answers. (5) The SMJ editorial office will submit the list of successful candidates to the Singapore Medical Council.

Deadline for submission: (October 2008 SMJ 3B CME programme): 12 noon, 25 November 2008.