Case 6738

Diaphyseal Aclerosis
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Section: Musculoskeletal system
Case Type: Clinical Cases
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Patient: 60 years, male

Clinical History:
Acute pain in the right ankle.

Imaging Findings:
A 60 year old male patient attended the Emergency Room complaining about acute onset of pain in the right ankle, following a mild trauma during his morning walk. Radiologic evaluation of the right ankle revealed no fracture, though multiple osteochondromas of the distal fibula and tibia were identified (Figure 1). The patient had a known history of diaphyseal aclerosis (DA). Physical examination of the knees and ankles revealed asymptomatic masses at the distal femurs, as well as the proximal and distal fibulas and tibias. Radiographs of the left ankle (Figure 2) and both knees (Figures 3, 4) revealed multiple exostoses with significant bone deformities. The patient had been aware of these lesions since his childhood, whereas his father was also known to have similar skeletal outgrowths. No chronic pain or other significant information were mentioned in the clinical history, apart from limited range of movement in the left knee joint. Radiologic evaluation with plain films obtained 7 years earlier revealed no change.

Discussion:
DA or Hereditary Multiple Exostoses is an inherited autosomal dominant disorder with incomplete penetrance in which multiple osteochondromas are seen throughout the skeleton. The estimated prevalence of DA is 1:50 000 to 1:100 000 in Western populations. Approximately two-thirds of affected individuals have a positive family history whereas the remaining cases are attributed to new mutations. The specific genetic abnormalities have been detected within three distinct loci on chromosomes 8, 11 and 19 designated respectively as EXT1, EXT2 and EXT3 [1,2]. Virtually all patients are diagnosed by the age of 12 years [2]. Osteochondromas are developmental anomalies resulting from the fragmental separation of the epiphyseal growth plate cartilage. Subsequent growth and enchondral ossification result in a subperiosteal osseous excrescence either sessile or pedunculated, with a cartilage cap that projects from the bone surface. The stalk of the osseous protuberance is in direct continuity with the underlying cortex and medullary canal. Upon skeletal maturity growth ceases [2].

In DA, any bone preformed in cartilage (enchondral ossification) may develop osteochondromas, with the exception of the calvaria. Specific sites include the scapula and ribs (40% of cases), humerus (50%–98%), elbow (35%–40%), wrist (30%–60%), hands (20%–30%), pelvis (5%–15%), hips (30%–90%), knees (70%–98%), ankles (25%–54%), and feet (10%–25%)[2]. Complications include osseous deformities, fractures, short stature, limb length discrepancy, pseudo-Madelung deformity, neurovascular compromise, bursa formation and sarcomatous transformation (chondrosarcoma or, rarely, osteosarcoma)[2].

Plain radiography remains the examination of choice in the evaluation of osteochondromas which are depicted as well marginated pedunculated or sessile bony excrescences. Continuity with the cortex and spongiosa of the parent
bone is pathognomonic. Pedunculated exostoses in tubular bones tend to be round or pointed and extend from the metaphyses toward the diaphyses, away from the joint. Metaphyses often become irregularly expanded and club-shaped. The cartilage cap may contain punctate, flocculent, comma-shaped, arclike or ringlike calcifications [2,6]. CT is complementary in the assessment of osteochondromas in the pelvis, shoulder, or spine. Using three-dimensional CT imaging, reconstructions can be formatted in various planes [2]. Ultrasonography is valuable in the diagnosis of bursitis and other complications associated with osteochondromas, such as arterial or venous thrombosis, aneurysm, and pseudoaneurysm formation [2]. MRI is useful for assessing accurate measurements of the cartilage cap which has high signal intensity on T2-weighted spin-echo images. Cap thickness more than 1,5 cm in skeletally mature patients suggests malignant transformation. It is the method of choice for evaluating compression of the spinal cord, nerve roots, and peripheral nerves [2,3]. Scintigraphy with thallium Ti 201 is useful in differentiating malignant transformation from benign osteochondroma in DA [4]. A normal isotopic bone scan virtually excludes the possibility of malignant transformation [5].

**Differential Diagnosis List:** Diaphyseal Aclasis

**Final Diagnosis:** Diaphyseal Aclasis

**References:**


Description: Anteroposterial radiograph of the right ankle: shows multiple steochondromas of the distal tibia and fibula. Note the widening of the distal fibula associated with modeling deformity [6] of the adjacent tibia. The cortex of the lesions is contiguous with the adjacent bone cortex. Origin:
**Description:** lateral radiographs of the right ankle: shows multiple osteochondromas of the distal tibia and fibula.

Note the widening of the distal fibula associated with modeling deformity[6] of the adjacent tibia. The cortex of the lesions is contiguous with the adjacent bone cortex. **Origin:**
Description: Anteroposterial radiograph of the left ankle. Note the dysplastic distal tibia as well as a sessile, broad based osteochondroma pointing away from the adjacent joint leading to the classical "coat-hanger" finding. Origin:
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Description: Anteroposterior radiograph of the right knee shows a sessile osteochondroma of the medial surface of the distal femur with characteristic cartilaginous calcifications, as well as a smaller pedunculated osteochondroma of the lateral surface. There is marked deformity of the proximal tibia and fibula caused by a pedunculated and a sessile osteochondroma accordingly. Origin: