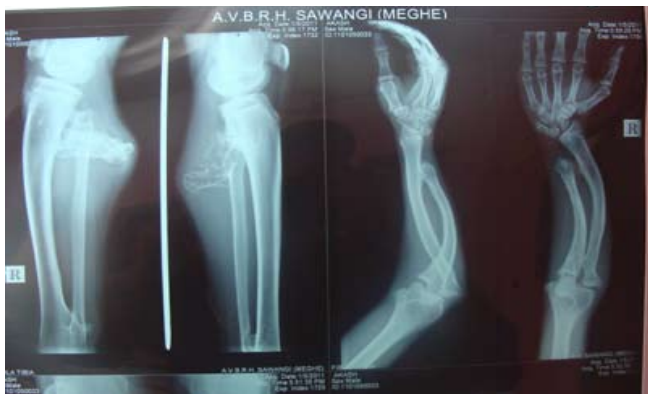
SPOT DIAGNOSIS (IMAGE GALLERY)



MULTIPLE HARD SWELLINGS - MULTIPLE OSTEOCHONDROMAS

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A14 years old boy presented with hard, multiple swellings over the body and right arm deformity. The parents had noticed these swellings early in childhood which increased over the time. The father (right forearm) and the paternal grandmother (right lower limb) had history of similar swellings. On examination, he was a well grown child with a weight of 48 kg and height of 154 cms. Hard bony swellings were noted at both calf area and also right lower thigh area and 5th right rib. There was right forearm bowing deformity with short left middle finger and short right middle toe. Systemic examination was normal. X-ray of the right leg and right forearm is depicted in Figure 1.

What is the diagnosis?

X-ray of the right leg shows pedunculated exostoses from upper tibia going posterolateral causing disruption of superior tibiofibular joint. X- ray right forearm showed bowing of ulna and radius with the shortening of ulna resulting in lower ulna and radial dyostosis and radiocapitular subluxation. In view of clinical picture, family history and X-ray findings, a diagnosis of multiple osteochondromas was made.

Multiple osteochondromas (MO) are also called as Hereditary Multiple Exostoses (HME). Multiple osteochondromas is characterised by development of two or more cartilage capped bony outgrowths (osteochondromas) of the long bones mostly around the knee, followed in frequency by the wrist, the proximal humerus, the proximal fibula and the ribs` with variable involvement of the scapula and the pelvis(1). Exostoses are the result of dysplasia of peripheral aspect of growth plate and are the most common type of benign bone tumor (2). Osteochondromas are not present at birth. They develop and increase in size in the first decade of life, ceasing to grow when the growth plates close at puberty (1). The prevalence is estimated at 1:50,000 and it seems to be higher in males (M:F ratio of 1.5:1) (3). MO is an autosomal dominant disorder with a penetrance of around 96 percent and is genetically heterogeneous. In almost 90 percent of MO patients, germline mutations in the tumour suppressor genes EXT1 or EXT2 are found (4).

The majority are asymptomatic. Remodelling defects caused by disruption of normal epiphyseal growth plate of the long bones lead to limb discrepancy and angular deformities` involved bone may be relatively short especially the ulna with consequent bowing of the forearm and also hand deformity resulting from shortened metacarpals (1). Symptoms may also arise secondary to mass effect causing compression of nerves, blood vessels and rarely spinal cord. Other complications include arthritis, bursa formation, osteomyelitis, muscle impingment and haemarthrosis. The most important complication is malignant transformation of osteochondroma, which is estimated to occur in 0.5–5 percent. Rapid growth and increasing pain, especially in a physically mature person, are signs of sarcomatous transformation (1, 3). Management includes removal of osteochondromas when they give complaints (3,4).

References:

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