

The role of bone scan in the diagnosis of Jaffé-Lichtenstein-Uehlinger syndrome

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Abstract

The case of a 12-year-old girl with Jaffé-Lichtenstein-Uehlinger syndrome is presented. A bone scan pattern exhibits clinical symptoms, X ray images and histological findings are described in a case of polyostotic form of the fibrous dysplasia. A biopsy with histology was performed in consideration of bone scan findings and a confirmed final diagnosis.

Key words: Jaffé-Lichtenstein-Uehlinger syndrome, bone scintigraphy, X-ray images, clinical symptoms, histology

Introduction

Fibrous dysplasia (FD) is a non-neoplastic developmental defect affecting a single bone (monoostotic form) or several bones (polyostotic form). The disease was described for the first time as separate entity by Lichtenstein in 1938. FD belongs to the group of tumour like diseases and osteochondrodysplasias of unknown aetiology. It usually is diagnosed in childhood or adolescence. The polyostotic form may be associated with endocrine condi-

tions such as precocious puberty, acromegaly, hyperthyroidism or Cushing's syndrome. The combination of the bone changes with precocious puberty and enhanced skin pigmentation is referred to as Albright's syndrome (1937) [1–3].

Fibrous dysplasia occurs when bone marrow cells are affected by somatic activating mutations of the gene encoding the α -subunit of the stimulatory G protein, $Gs\alpha$. The mutation has a mosaic distribution and results in locally increased stimulation of adenylyl cyclase and overproduction of cyclic adenosine monophosphate (cAMP). In bone mesenchymal cells, the downstream effects of the mutation include increased production of the c-fos protein and interleukin. At the bone tissue level, FD is characterised by dysplastic lesions that consist of abnormal fibrous tissue in the marrow space intertwined with poorly oriented, irregular trabeculae of woven bone [4]. The prognosis of the disease is favourable in the majority of cases irrespective of the development of complicating deformities or fractures. Malignant transformation occurs in approximately 1% [1].

A case report

A 12-year-old girl was referred with a history of right ankle distortion. Enlargement of sternal end of the left clavicle (Fig. 1) and right external ankle pain and oedema (Fig. 2) were found during a complex clinical investigation. In hematological and biochemical tests only high level of sedimentation rate considered to be significant. A bone scan using Tc-99m hydroxydiphosphonate (HDP) (Fig. 3, 4) and X rays of painful sites suggesting an osteomyelitis (Fig. 5, 6) were performed. A whole body bone scan (Fig. 7) showed more foci of increased radioactivity, aside from the lesions in the left clavicle and right foot corresponding to clinical findings, also the foci of increased activity in the spine, left knee and the right wrist. A biopsy of the left clavicle with histology confirmed the final diagnosis (Fig. 8). The girl was readmitted seven months later with similar difficulties. A follow-up bone scan described new foci of increased activity (Fig. 9).

Bone scintigraphy is a useful method in multifocal bone disease. It can detect lesions earlier and it can help in the assessment

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Figure 1. Enlargement of sternal end of left clavicle found during complex clinical investigation.

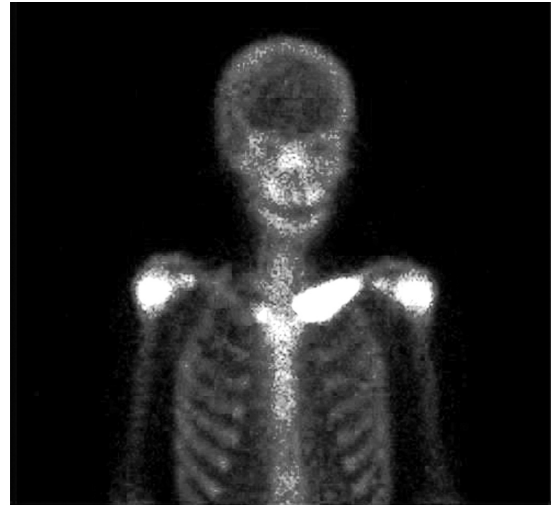


Figure 4. Bone scintigraphy — increased uptake of tracer in the whole left clavicle.

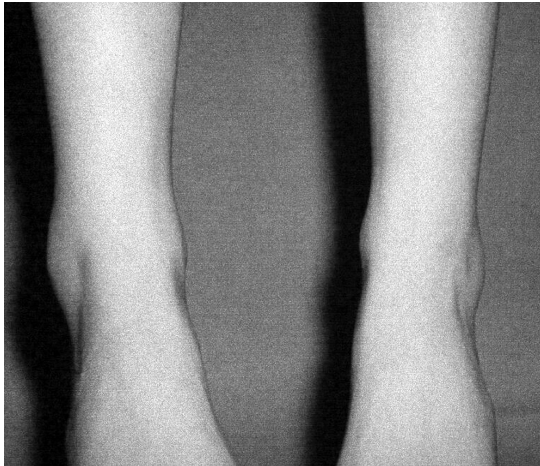


Figure 2. A 12-y-old girl with a history of right ankle distortion. Oedema and pain of the right external angle.



Figure 5. A small defect of the bone tissue in the area of metaphysis next to the border-epiphyseal growing zone of the distal end of right fibula detected by X ray.

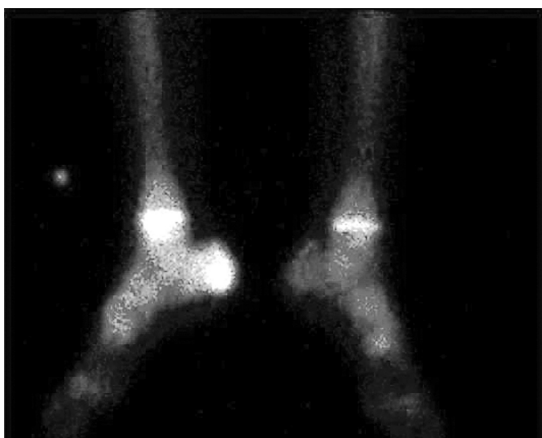


Figure 3. Bone scintigraphy with Tc-99m MDP - the focus of increased radioactivity in the localisation corresponding to the bone tissue's defect of fibula on X ray. Another focus of increased uptake of tracer was seen in calcaneus.

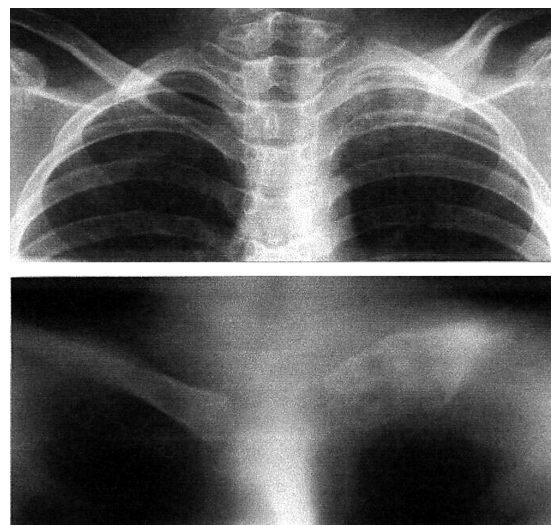


Figure 6. X ray of left clavicle — „bubble” cystoid transformation and smooth edge sclerosis.



Figure 7. Whole body bone scan — more foci of increased activity in the left clavicle, right ankle, right calcaneus, vertebrae Th-5, -6, -12, L-2 and in the left knee and the right wrist.



Figure 9. The follow-up of bone scan seven months later — new foci in the right scapula and the head of right humerus. The foci in the spine and in right wrist did extend. The focus in the left clavicle was larger and increased activity survived in the left knee only in the area of proximal tibia. The findings of right foot remained the same.

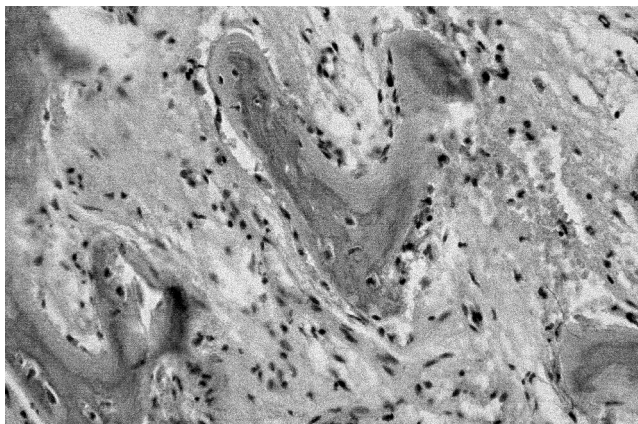


Figure 8. Histological findings — cellular fibrotic tissue with characteristic „V“ shaped trabeculae of fibrotic bone, which is not well matured everywhere. Osteoblasts were on the surface of trabeculae.

of the appropriate site of a biopsy. It is also very important in follow-up investigations to monitor the development of the disease.

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