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E-QUID: ANSWER / *Pediatric imaging*

Mac Cune-Albright syndrome. Answer to the e-quid ''Painful lameness in a child''☆

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Case report

A 5-year-old girl consulted for painful intermittent lameness of the right leg that has been evolving for 2 weeks. The clinical examination revealed tumefaction at the root of the thigh and the presence of three pigment spots (Fig. 1). Upon questioning, the recent occurrence of vulvar bleeding was discovered. An X-ray of the right hip (Fig. 2) and a MRI of the pelvis (Figs. 3) were performed.

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☆ Here is the answer to the case ''Painful lameness in a child'' previously published in the nº 3/2013. As a reminder we publish again the entire case with the response following.

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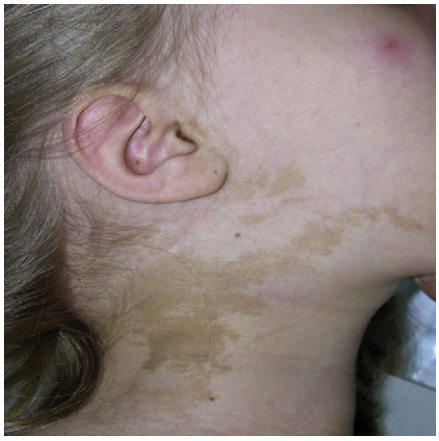

Figure 1. Photo of the neck.

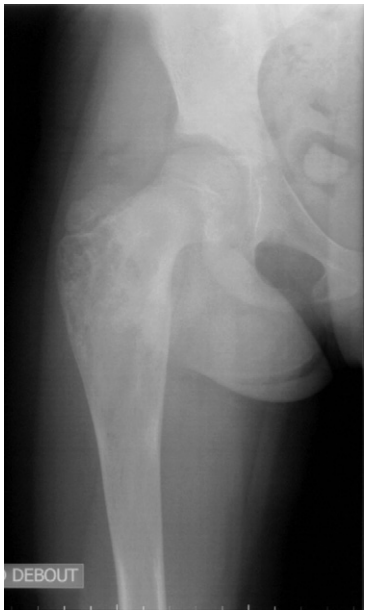

Figure 2. Front X-ray of the right hip.

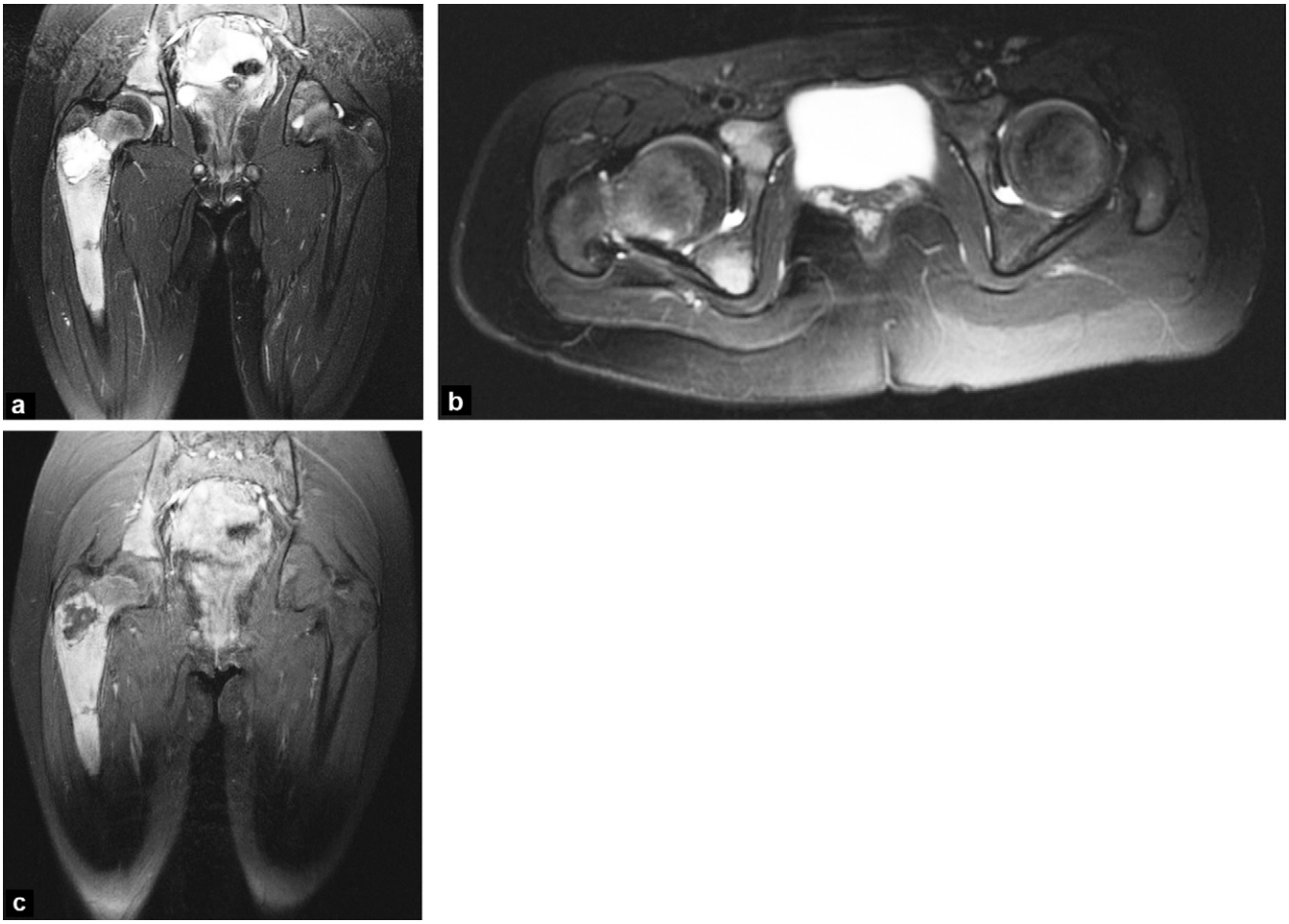

Figure 3. MRI of the pelvis. (a) Coronal section T2 weighted with saturation of the fat signal. (b) Axial section T2 weighted with saturation of the fat signal. (c) Coronal section T1 weighted after injection of Gadolinium and with saturation of the fat signal.

What is your diagnosis?

After reading the case report, what diagnosis would you choose from the following proposals:
- metastatic neuroblastoma;
- fibrous dysplasia;
- aneurismal cyst;
- chondroma;
- Mac Cune-Albright syndrome.

Diagnosis

Polyostyotic fibrous dysplasia associated with early puberty and the presence of *café au lait* spots in a fern-like pattern included in a Mac Cune-Albright syndrome.

Complementary examination

Pelvic ultrasound (Fig. 4).

Comments

The X-ray of the right hip (Fig. 5) shows an osteolytic lesion of the upper end of the femur with zones of condensation in unpolished glass and an enlargement of the trochanter.

Cortical thinning is associated, extending to the diaphysis. In the pelvic MRI (Fig. 6), the metaphyseal-diaphyseal femoral lesion demonstrates two distinct contingents: a proximal section, with serpiginous outlines, in hyposignal T1, hypersignal T2 distinct not enhanced by the Gadolinium and a distal osteoid area of intermediate signal in T2 and enhanced in a homogenous manner after injection of Gadolinium. There are other locations on the right acetabulum on both sides of the cartilage in Y (Fig. 6b).

Discussion

Fibrous dysplasia is a non hereditary congenital bone disease. The prevalence is difficult to estimate due to the many asymptomatic forms. It is estimated at less than 1/5000. The genetic anomaly responsible is a mutation of the gene coding for the α sub-unit of the stimulating G protein (Gs) located on chromosome 20q13 [1]. All bones may be affected. Isolated, or monostotic impairment, is the most common. It affects the ribs (45%), the femur (15%), the tibia (12%), the maxillary bone (10%), the top of the skull, the mandible, the humerus and the ulna [2]. Classically, a unilateral arrangement of the lesions is found in the polyostotic form. The first symptoms occur in childhood. The age of discovery ranges from 5 to 30 years of age.

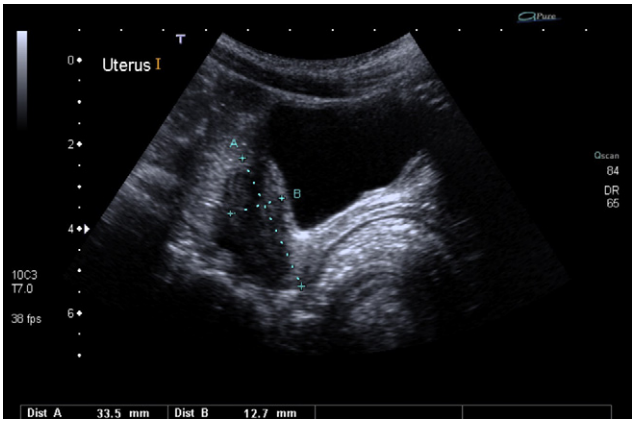

Figure 4. Pelvic untrasound. Sagittal section. Pubic uterus, large for the age of the patient.

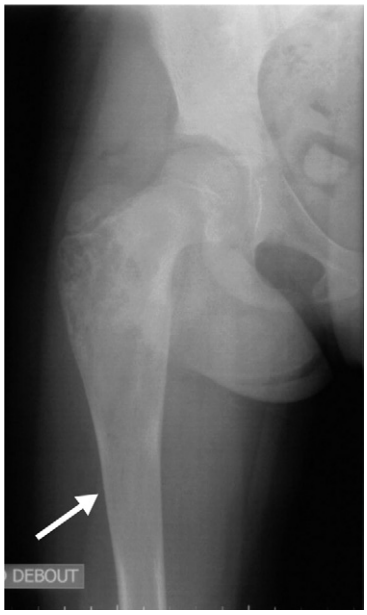

Figure 5. Front X-ray of the right hip. Unpolished glass appearance associated with cortical thinning (arrow).

The lesions are often asymptomatic and the discovery by chance. The non specific symptoms are dominated by chronic bone pain [3]. Headache is classic in the cranio-facial forms. Any acute pain may indicate fracture complications. Due to its expansive nature, fibrous dysplasia sometimes provokes bone deformations. For example, shepherd's crook deformity of the proximal femoral location is classic. Cranio-facial impairment may account for neurosensory complications, diplopia by oculomotor compression being most common.

The diagnosis may be based on imaging. Three radiographic aspects are described: homogenous clarity, smoke curl appearance or condensed unpolished glass appearance. The usual description is that of a well circumscribed, blowing, radiotransparent, lytic lesion, developed to the detriment of the bone marrow. In the extensive forms, this expansive lesion is at the origin of cortical remodelling or even thinning. The benign nature is indicated by the presence of a dense, typically thick, peripheral border. The infiltration of long bones is diaphyseal—metaphyseal.

The asymptomatic locations may be found by bone scintigraphy (since the still active lesions demonstrate tracer uptake) or, even better, in fully body MRI, in order to limit the exposure in the child. In the scan, unpolished bone condensation is classic [4]. Lesional calcifications may be present. Moreover, the scanner is more sensitive for the detection of cortical erosion, bone fissures and fracture

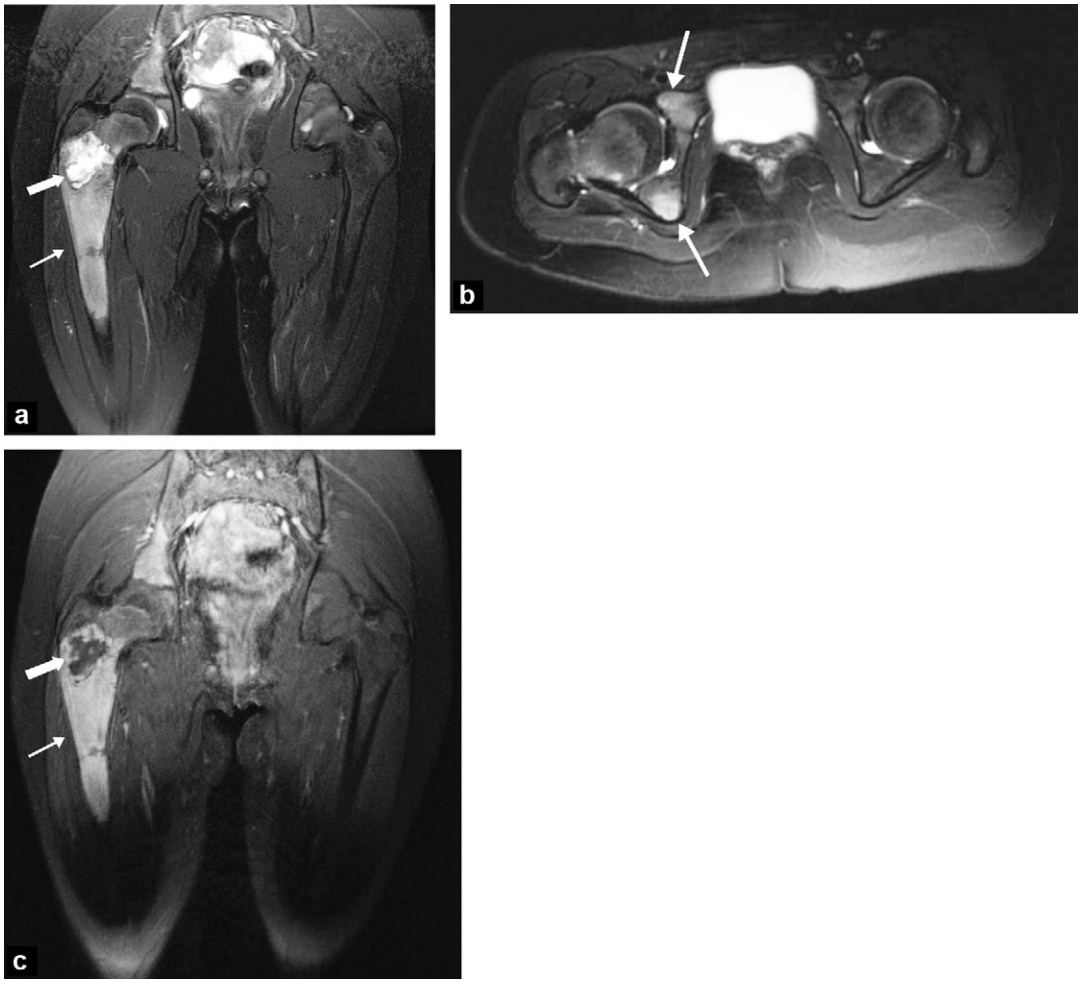

Figure 6. MRI of the pelvis. It consists of a mixed form of fibrous dysplasia with a cystic proximal contingent and a distal osteoid matrix. (a) Coronal section T2 weighting with saturation of the fat signal. The cystic component is in distinct T2 hypersignal (full arrow). The osteoid component is more intermediate (thin arrow). (b) Axial section T2 weighting with saturation of the fat signal. Note the polyostotic aspect of the fibrous dysplasia with other locations on the right acetabulum (white arrows). (c) Coronal section T1 weighting after injection of Gadolinium and with saturation of the fat signal. The non-enhanced cystic contingent is distinguished (full arrow) of the osteoid contingent that takes up the contrast (thin arrow).

complications. The absence of periostyle appositions and infiltration of the soft tissue is an argument in favour of the benign nature of the lesions. In the cranio-facial forms, the scanner reveals pseudo-cystic rearrangements and sclerotic lesions at the base of the skull.

The MRI signal varies according to the hystologic nature, the degree of mineralisation, the presence of bone framework or haemorrhagic rearrangements [5]. No aspect is pathognomonic. The most mineralised seats appear hyperintense in T1 and of intermediate signal in T2. The enhancement is often central as well as peripheral in one third of the cases. In polyostotic fibrous dysplasia, the radiologic elements suffice for the diagnosis. A biopsy is only indicated in doubtful cases, to make sure of the benign nature of a single lesion of tumoral appearance. Fibrous dysplasia is a disease of the osteoblast stem cells with anomalies in the proliferation and differenciation of the osteogenic cells. Normal bone is replaced by a pathological connective matrix filling the bone marrow and taking on the appearance of a Chinese calligraphy. The spongy bone is resorbed, leading to progressive cortical thinning. This bone resorption accounts for the growth potential of the lesions. There is also an excess of osteoclasts.

The mutation affects the osteoblast precursors as well as a great many endocrine gland cells [6]. Hormone receptors coupled with Gs proteins (LHr, FSHr, TSHr, GHRHr, ACTHr) lead to the stimulation and production of gonad, thyroid or autonomous pituitary hormones. Endocrine disorders, associated with polyostatic fibrous dysplasia and *café au lait* skin spots indicate a Mac Cune-Albright syndrome [1]. The outline of the pigment spots is irregular, finely cut and classically described as a fern-leaf pattern and are darker and less abundant than the ''coffee with milk'' of the neurofibromatosis. The endocrine impairment is related to autonomous endocrine activation. Early pseudopuberty was the first endocrine disorder described in the Mac Cune-Albright syndrome although other endocrine disorders are possible (hyperthyroidism, acromegaly, hyperparathyroidism, hypercorticism). The lesions become quiescent at puberty. The prognosis is functional, related to the pain and the bone deformations. Regular clinical monitoring suffices in the asymptomatic forms. The bisphophonates have proven

their efficacy on the pain and the reduction of the number of locations. Surgery only has a limited place in the complicated forms (fractures, neurological compression) [7]. Finally, care of the Mac Cune-Albright syndrome includes a specific treatment for the endocrine disorder.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

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