Interesting Image
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Polyostotic Fibrous Dysplasia in a Six-year-Old Boy

Altı Yaşındaki Erkek Çocukta Poliostotik Fibröz Displazi

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Abstract

Fibrous dysplasia (FD) is a rare congenital benign bone disease that manifests as a defect in the bone remodeling process, affecting the function, differentiation, and maturation of osteoblasts. This process is located in the bone marrow, where the normal marrow tissue is replaced with immature bone islands and fibrous stroma. The etiology is unclear so far, but it is known to be connected with a point mutation of the gene that encodes Gs α protein at the time of embryogenesis, and because of that, all of the affected somatic cells become dysplastic. It is important to determine whether the mutation occurred earlier in the process of embryogenesis so that there will be more mutant cells and the disease will appear in a more severe form. The clinical presentation of FD is variable, so there are plenty of potential differential diagnoses. The most common include Paget disease, non-ossifying fibroma, osteofibrous dysplasia, aneurysmal bone cyst, adamantinoma, giant cell tumor, fracture callus, and low-grade central osteosarcoma.

Keywords: Fibrous dysplasia, bone scan, SPECT

Öz

Fibröz displazi (FD), osteoblastların işlevini, farklılaşmasını ve olgunlaşmasını etkileyen, kemiğin yeniden şekillenmesi sürecinde bir kusur olarak ortaya çıkan, nadir görülen, iyi huylu konjenital bir kemik hastalığıdır. Bu süreç, normal kemik iliği dokusunun olgunlaşmamış kemik adaları ve fibröz stroma ile yer değiştirdiği kemik iliğinde cereyan eder. Etiyoloji net değildir, ancak etiyolojinin embriyogenez sırasında Gs α proteinini kodlayan genin nokta mutasyonu ile bağlantılı olduğu bilinmektedir ve bu nedenle etkilenen tüm somatik hücreler displastik hale gelmektedir. Mutasyonun embriyogenez sürecinde daha erken meydana gelip gelmediğini belirlemek, daha fazla mutant hücre oluşması ve hastalığın daha şiddetli bir şekilde ortaya çıkması açısından önemlidir. FD'nin klinik prezentasyonu değişkendir, bu nedenle çok sayıda potansiyel ayırıcı tanı vardır. En yaygın olanları Paget hastalığı, ossifiye olmayan fibroma, osteofibröz displazi, anevrizmal kemik kisti, adamantinom, dev hücreli tümör, kırık kallus ve düşük dereceli santral osteosarkomdur.

Anahtar kelimeler: Fibröz displazi, kemik sintigrafisi, SPECT

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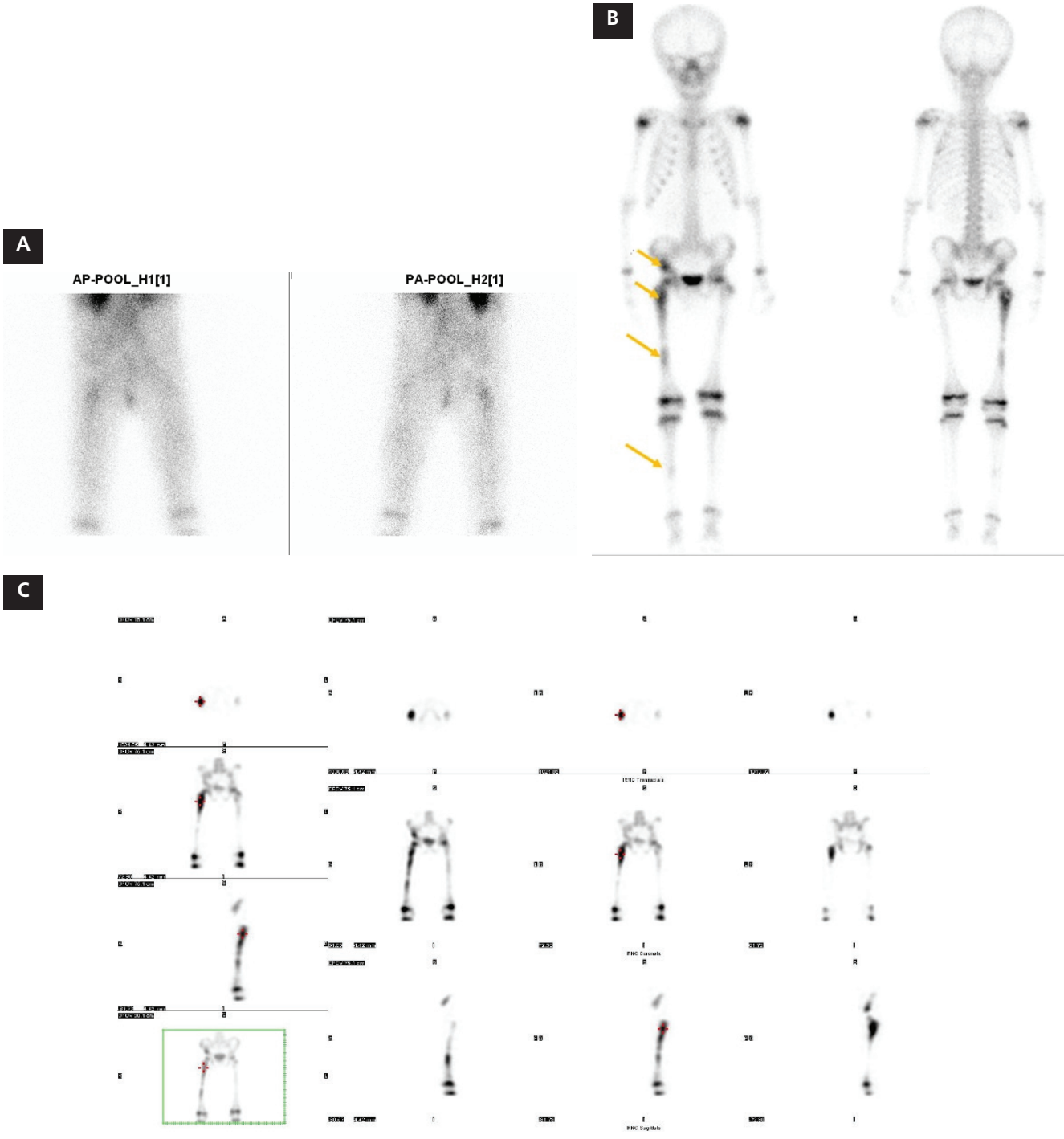

Figure 1. A We present a case of a 6-year-old boy complained of a right hip pain, especially at night and on exertion. He was diagnosed with a fracture of the subtrochanteric region of the right femur. The bone scan performed after an intravenous application of Tc-99m-methyl diphosphonate (MDP) revealed increased vascularization of the right hip in the early pool phase image (A). Whole-body images showed intensive uptake of Tc-99m-MDP in the right iliac bone and right femur (proximal diaphysis), with not very intense accumulation in the middle 1/3 of the right femur and right tibia (B). Single-photon emission computed tomography (SPECT) images confirmed the uptake in the iliac bone and the femur (C).
Fibrous dysplasia (FD) accounts for 7% of the benign bone tumors, primarily affecting young adolescents and young adults, with no gender predilection gender predilection. It may appear as monostotic (affecting one bone), polyostotic (multiple bones), or craniofacial FD (skull and facial bones alone).

Common sites of skeletal involvement are long bones, ribs, craniofacial bones, and the pelvis (2). The polyostotic form is found in 20-30% of cases. It presents earlier, typically in childhood (mean age of 8 years) with two/thirds being symptomatic by the age of 10. In early childhood, lesions are metabolically active and expand during linear growth. The lesions typically become static in size after puberty, and metabolic activity may decrease throughout adulthood (3). The polyostotic form of the disease is often accompanied by an endocrine disorder such as McCune-Albright syndrome. This kind of syndrome combines FD with other extraskeletal features, such as "café-au-lait" skin macules with characteristic distribution, gonadotropin-independent sex steroid production in girls and women or autonomous testosterone production in boys and men, thyroid lesions, growth hormone excess, neonatal hypercortisolism, and renal phosphate wasting (4). The bone scan shows increased uptake throughout life, but the uptake becomes less intense as the lesions mature. Some characteristic findings within lesions of FD are a bar-shaped pattern, whole bone involvement, and a close match between the size of the lesion on radiographs and the size of the area of uptake. The lesions show increased uptake of the tracer, but its intensity is variable, so sometimes false-negative results are detected. The mechanism of different degrees of Tc-99m-MDP metabolism of FD is unclear. As we know, FD is developmental failure in the remodeling of the primitive bone to a mature lamellar bone. Fibroblasts are the predominant proliferating cells in FD lesions, and the different degrees of 99mTc-MDP metabolism among them may be due to the difference in the vascular supply or the number of proliferating fibroblasts or their metabolic turnover (7).

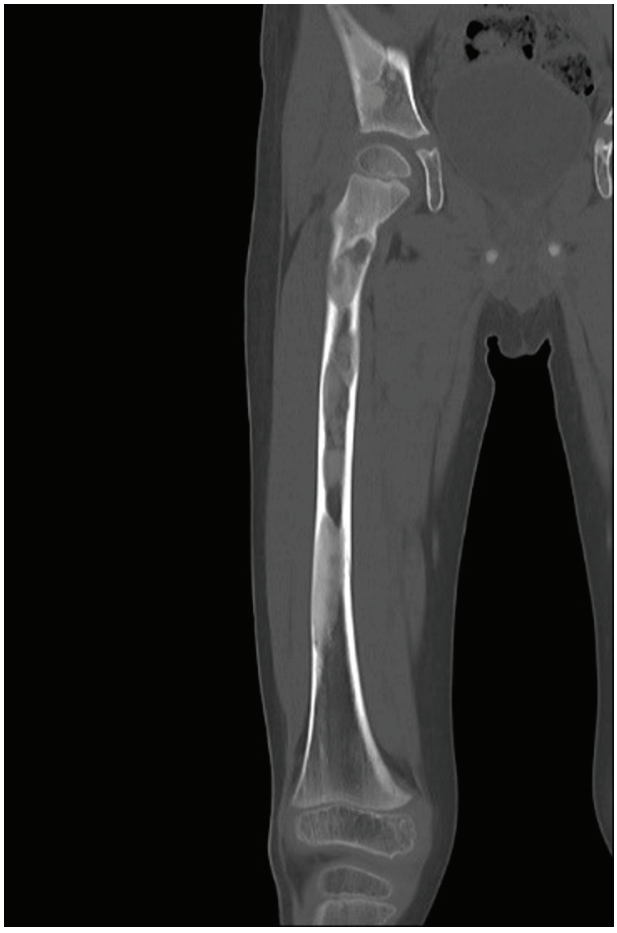

Figure 2. Computed tomography image of the right femur. The coronal plane showed a changed skeletal structure of the diaphysis of the right femur with present multifocal ground-glass opacities and cystic components.

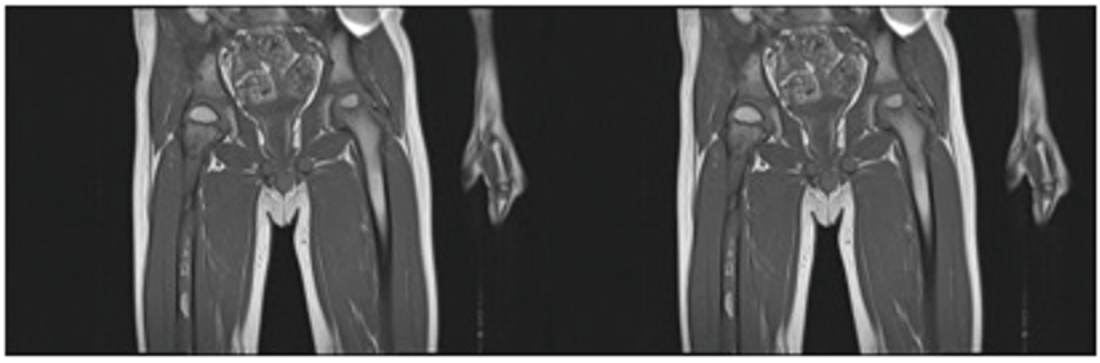

Figure 3. Magnetic resonance imaging (MRI) in 2019. There were multiple circumscribed lesions with ground-glass opacity in the right iliac bone, the superior ramus of the right ischiatic bone, and the right femur without involving the head. In the region of the right femoral neck and the intertrochanteric region, predominantly cystic and sclerotic components were delineated in the lesions. No cortical disruption or the cortical expansion was noted. There were signs of bone expansion in the inter- and subtrochanteric regions with cortical thinning but no disruption of the cortex.

Radiolucent ground glass matrix, which is smooth and homogeneous, is a typical FD lesion on radiography. Sometimes these lesions appear as completely radiolucent (cystic) lesions or sclerotic lesions, but mixed forms are also described. They have well-circumscribed thick sclerotic margins around the radiolucent lesion that can be interrupted, the so-called rind sign (5). MRI reports in patients with FD have been quite challenging, considering the high variability in the tumor signal on the MR examination..

Laboratory findings were as follows: C-reactive protein: 27.4 (<6 mg/L); white blood cells: 20.1 (4-9x109/L); alkaline phosphatase: 173 (36-126 U/L). These inflammatory markers are non-specific for FD, except for the alkaline phosphatase that can be high in patients with metabolic bone disease. Three months later, he underwent surgery with resection of the right fibula that was implanted on the site of the proximal diaphysis of the right femur.

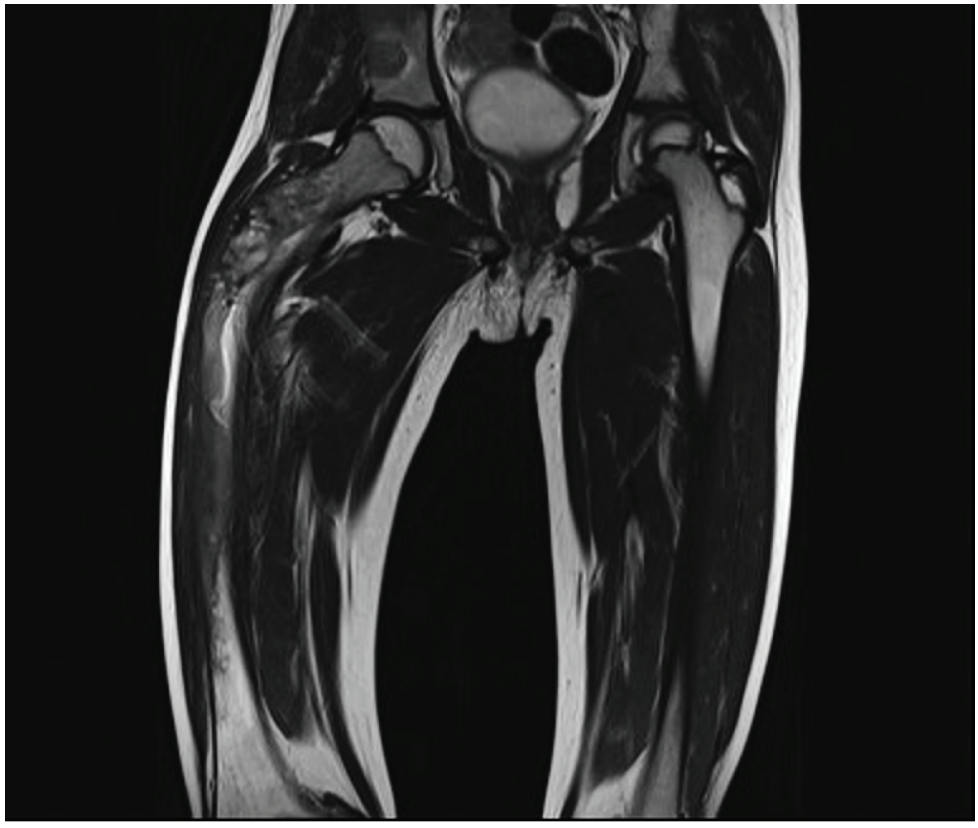

Figure 4. MRI images in 2021 there are predominantly fibrous and cystic lesions in the intertrochanteric region with mild bone expansion and endosteal scallopings. Changes in the skeletal structure in the basicervical, intertrochanteric, and proximal diaphyseal regions.
For confirming the diagnosis, biopsy is not always indicated. If there is a clear radiological finding for FD, only regular observation is needed. Follow-up radiographs should be made every six months to verify that there has been no progression. Bisphosphonates are the therapy of choice for pain relief and bone strengthening. Surgery is needed in rare cases only when complications such as fractures or deformities are present (8).

Ethics

Informed Consent: Written informed consent was obtained.

Peer-review: Externally peer-reviewed.

Authorship Contributions

Concept: N.M., Design: N.M., S.B.K., Data Collection or Processing: N.M., S.B.K., Analysis or interpretation:S.S., T.M., Literature Search: D.T-T., T.M., Writing: D.T-T., S.S., N.M.

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