

## Seropositive Rheumatoid Arthritis with Very Unusual X-ray Findings

Laith Alamliah, Mohamed Alkahlout, Abdulrahim Siam, Syed Alam, Abdul-Wahab Al-Allaf  
Rheumatology Department, Hamad General Hospital, Doha, Qatar

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### ABSTRACT

We described the case of a 23-year-old Nepalese man with seropositive rheumatoid arthritis and abnormal x-ray findings, found to be due to a very rare bone disease: Camurati Engelmann disease or progressive diaphyseal dysplasia (PDD). This is the first case reported in the Gulf area, although approximately 300 cases have been described worldwide. These patients usually present with limb pain and easy fatigability. Our patient first presented with bilateral, symmetrical inflammatory polyarthritis involving the knees, ankles and wrists but sparing the hands and feet. The diagnosis of PDD in our case was based on the classic radiological findings and a bone scan.

### LEARNING POINTS

- Rheumatoid arthritis is a common condition with typical radiological findings.
- Any unusual radiological finding should be carefully assessed and explained.
- In our case the unusual findings were due to progressive diaphyseal dysplasia.

### KEYWORDS

Camurati Engelmann disease, rheumatoid arthritis

### CASE REPORT

A 23-year-old Nepalese man presented with bilateral symmetrical joint pain and swelling involving his knees, ankles and wrists, but sparing the small joints of his hands and feet, and associated with significant early morning stiffness. There was no history of skin rash, fever or shortness of breath and no back, chest or abdominal pain.

On examination, the patient had marked swelling of the knees, ankles and wrists with tenderness. There were large knee effusions bilaterally (Fig. 1). The small joints of the hands and feet were not swollen or tender, but the hands and feet including the fingers and toes were enlarged with clubbing. There were no oral ulcers, eye redness, or back or sacroiliac joint tenderness.

### Investigations

CBCs, renal, liver and bone profiles were all normal. The inflammatory markers were elevated (ESR 51 and CRP 56), and the positive anti-CCP antibody was present. Synovial fluid analysis revealed non-inflammatory fluid with a WBC of 355 with lymphocyte predominance. X-rays showed diffuse irregular cortical bone thickening and mild fusiform bony enlargement in the diaphyseal region of the long bones, with sparing of the epiphyses, which is characteristic of PDD (Figs. 2–4). A bone scan showed an isolated increase in radiotracer uptake involving the diaphyseal regions in a symmetrical fashion (Fig. 5).

The patient was started on oral methotrexate (MTX) with folic acid in addition to oral prednisolone 5 mg daily. However, response to treatment was minimal with persistent joint pain and swelling.



Figure 1. Bilateral knee swelling with effusion



Figure 2. Femur X-ray showing generalized symmetrical mild fusiform enlargement and cortical thickening involving the diaphysis of the right and left femur (red arrows), sparing the epiphyses.



Figure 3. X-ray of the right leg showing symmetrical enlargement and cortical thickening involving the diaphysis of the tibia and fibula (red arrows), sparing the epiphyseal region, which is classic of progressive diaphyseal dysplasia (Camurati-Engelman's disease). Similar changes were found in the other leg the synovial membrane and structural alterations of the tibia, talus and calcaneus with several chondromatosis bodies in the anterior part of the tibiotalar joint



Figure 4. Right knee showing symmetrical enlargement and cortical thickening involving the diaphysis of the visualized bones, with knee effusion. the anterior part of the tibiotalar joint

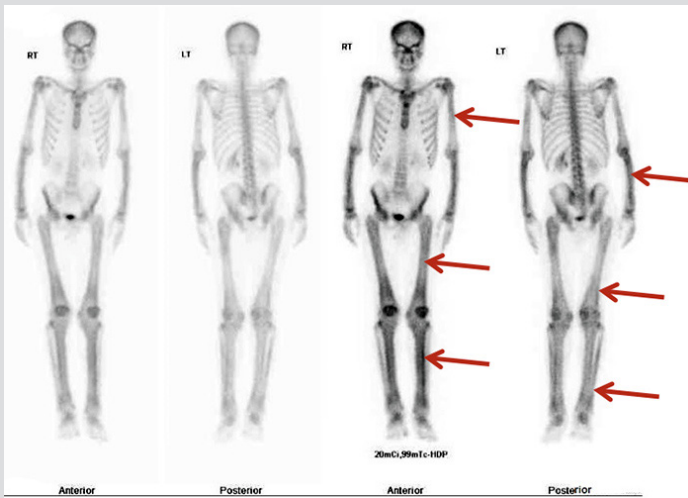


Figure 5. A bone scan showed symmetrically increased radiotracer uptake involving the diaphyseal regions of the long bones of the upper and lower limbs (lower more than upper extremities). The cortex is differentiated from the medulla of the involved shafts of the long bones, but the ends of the long bones (metaphyso-epiphyseal regions) are not affected.

## DISCUSSION

PDD is very rare<sup>[1,2]</sup>. It is most likely caused by increased periosteal bone formation with decreased endosteal resorption in the long bones, which is evidenced by thickening of the diaphyseal area together with a narrowed medullary canal<sup>[3]</sup>. The disease is associated with mutations in the TGFB1 gene<sup>[5]</sup>, but not all patients with the disease have a mutation. Therefore, it was proposed that the disease be classified as type I (MIM 131300) when a mutation in TGFB1 is identified and as type II (MIM 606631) if the mutation is not found<sup>[6]</sup>. Although most cases are familial, some sporadic cases have been reported<sup>[7-9]</sup>. The classic radiographic findings include symmetrical hyperostosis and endostosis of the diaphysis of the long bones, and to a lesser extent of the metaphysis. The lower extremities are usually more often involved than the upper extremities<sup>[9]</sup>. An isotope bone scan usually shows uptake in the diaphysis, which finding can also be found in some members of the affected family with no clinical or radiological indication of the disease<sup>[10]</sup>. Our case demonstrated all the classic radiographic and bone scan findings for PDD.

In the majority of cases, inflammatory markers should be within normal limits, but occasionally can be raised<sup>[11,12]</sup>, as in our patient. In this case, the presence of inflammatory arthritis, which is not typical for PDD, should be further explored. Our patient clearly has seropositive RA, which is a much more common condition. We do not think that there is an association between PDD and rheumatoid arthritis (RA) as a rare disease such as PDD could incidentally co-occur with another common condition such as RA.

The management of PDD and RA is totally different. Treatment for PDD includes anti-resorptive agents such as pamidronate, steroids, aspirin and NSAIDs. They are used mainly for pain, with variable success, but have no effect on the radiographic and isotopic features of the disease<sup>[13-16]</sup>. Our patient was treated with MTX and folic acid for his RA with only a modest response. We think that the partial response could be related to the treatment of RA with MTX. However, the low, non-inflammatory range of synovial fluid WBCs raised the possibility that this effusion could be due to PDD, which has not been previously reported.

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