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CASE REPORT

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Asynchronous progressive diaphyseal dysplasia

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Abstract We report the case of a 42-year-old Japanese woman with unusual diaphyseal dysplasia of bilateral femora. Radiographs showed thickening and sclerosis of the cortex with resultant enlargement of the diaphysis, unclear demarcation of the surface of the cortex, and no periosteal reaction. These changes were found on the left femur at the first presentation, and those on the right femur developed within several years. Although this patient partly presented characteristics of Ribbing disease and Camurati–Engelmann disease, the focal involvement of bilateral femora suggested an unknown pathogenesis.

Key words Asynchronous onset · Caffey disease · Camurati–Engelmann disease · Diaphyseal dysplasia · Femur · Ribbing disease

Introduction

Diaphyseal dysplasia is a rare bone dysplasia, which is known as Ribbing disease[1] or Camurati–Engelmann disease.[2] Radiographically, thickening of the cortices of the long bones is observed. Only the cortical bone of the diaphysis is involved and the metaphysis and epiphysis of the same bones are usually normal. The etiology of these diseases is unknown and therefore the optimal treatment for these patients has not been established.

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We report the case of a 42-year-old Japanese woman, whose clinical and radiological features do not completely fit the diagnosis of classical diaphyseal dysplasia. The features of this patient were asynchronous onset, focal involvement of bilateral femora, and a negative family history. The diagnosis of this patient has not been established as yet.

Case report

A 42-year-old woman presented with a 1-year history of pain in the left thigh with insidious onset. The patient had neither a history of traumatic event nor constitutional symptoms of a systemic illness. Her family history was negative. On examination, there was tenderness on the distal anteromedial aspect of the left thigh. The range of motion of bilateral hips and knee joints was full. Radiographs of the left femur showed thickening of the cortex and sclerosis with the resulting enlargement of the diaphysis, unclear demarcation of the surface of cortex, and no periosteal reaction, whereas the right femur was apparently normal (Fig. 1). A ^{99m}Tc-medronate (MDP) bone scan revealed marked uptake in the left femur and very minor uptake in the right femur (Fig. 2A). A T1-weighted coronal image showed an intramedullary lesion of low signal intensity, while a T2-weighted coronal image revealed a high-intensity lesion on bilateral thighs (Fig. 3). The laboratory findings were normal, except for a slight increase of the white blood cell (WBC) count to 15300, a C-reactive protein (CRP) value of 0.52 mg/dl, an alkaline phosphatase (ALP) value of 255 U/l, a lactate dehydrogenase (LDH) value of 255 U/l, a calcium value of 9.5 mg/dl, and a phospholipid value of 3.3 mg/dl. A needle biopsy was performed. The bacteriological examination was negative, but the histological examination revealed nonspecific destructive and reparative changes of the bone without infiltration of inflammatory cells or tumor cells. Antibiotics did not help to alleviate her symptoms.

Four years after the onset, the symptoms had not disappeared, but the right thigh had remained asymptomatic.

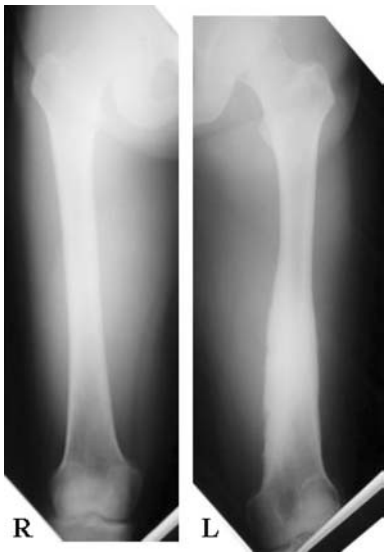

Fig. 1. Anteroposterior radiographs of the femora obtained when the patient was 42 years old. Marked cortical thickening of the left femur was detected

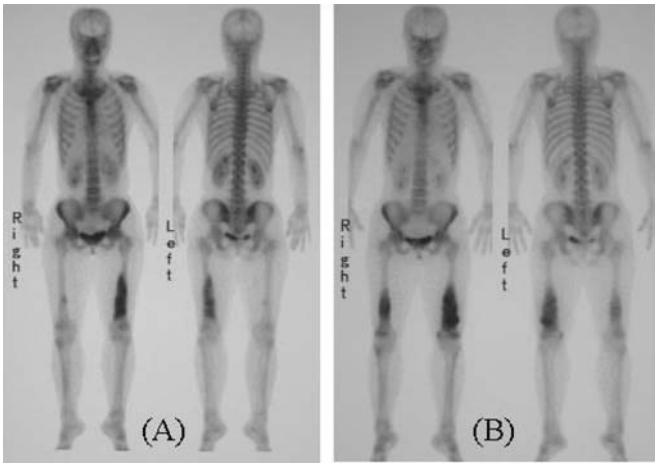

Fig. 2A,B. ^{99m}Tc-Medronate bone scans. **A** At the age of 42 years marked uptake was noted in the diaphysis of the left femur, whereas only very minor uptake was detected in the right femur. **B** At the age of 45 years, asymmetric, increased uptake was observed at the femora while there was no remarkable uptake at other sites

The patient was afebrile and had no signs of inflammation on either thigh. The laboratory examination disclosed slight inflammatory findings similar to those observed at the first presentation. Radiographs of bilateral femora showed thickening of the cortex and sclerosis, the left lesion showed progressive changes, and the right lesion showed a remarkable thickening of the cortex and sclerosis compared with the initial radiographs (Fig. 4). Axial computed tomography (CT) scan showed thickening of the cortex of bilateral femora (Fig. 5A), and a laminar productive change was observed in the left femur (Fig. 5B). A ^{99m}Tc-MDP bone scan revealed increased uptake in both lesions and no remarkable uptake at other sites (Fig. 2B). A needle biopsy of

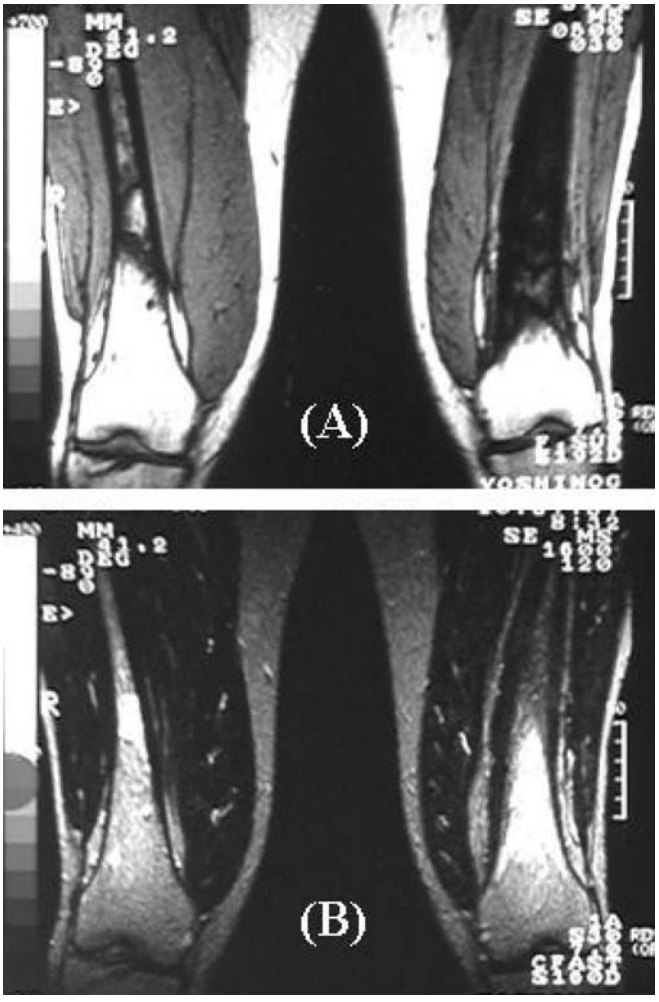

Fig. 3A,B. Magnetic resonance images of the distal thighs obtained at the age of 42 years. **A** T1-weighted coronal image showing an intramedullary lesion of low signal intensity. **B** T2-weighted coronal image showing a lesion of high signal intensity on bilateral femora

the lesion in the left femur was performed again, and the histological examination showed hyperostosis.

The patient was prescribed etidronate disodium, which contributed toward alleviating pain to a certain extent. The patient has been followed up while under treatment with nonsteroidal anti-inflammatory drugs (NSAIDs). She has presented no symptoms related to the lesion on the right femur (Fig. 6).

Discussion

We have described the case of a patient with diaphyseal dysplasia of bilateral femora characterized by asynchronous onset, focal involvement, and a negative family history. Radiographically, thickening of the cortex and sclerosis of bilateral femora were detected. Ribbing disease,[1] Camurati–Engelmann disease,[2] Caffey disease,[3] chronic sclerosing osteomyelitis,[4,5] multifocal periostitis,[6] prostag-

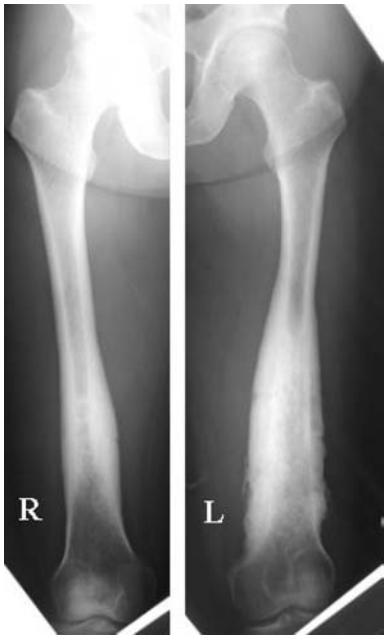

Fig. 4. Anteroposterior radiograph of the femora obtained at the age of 45 years. A remarkable thickening of the cortex and sclerosis were detected in the right femur, while progressive changes were noted in the left femur compared with the initial radiographs

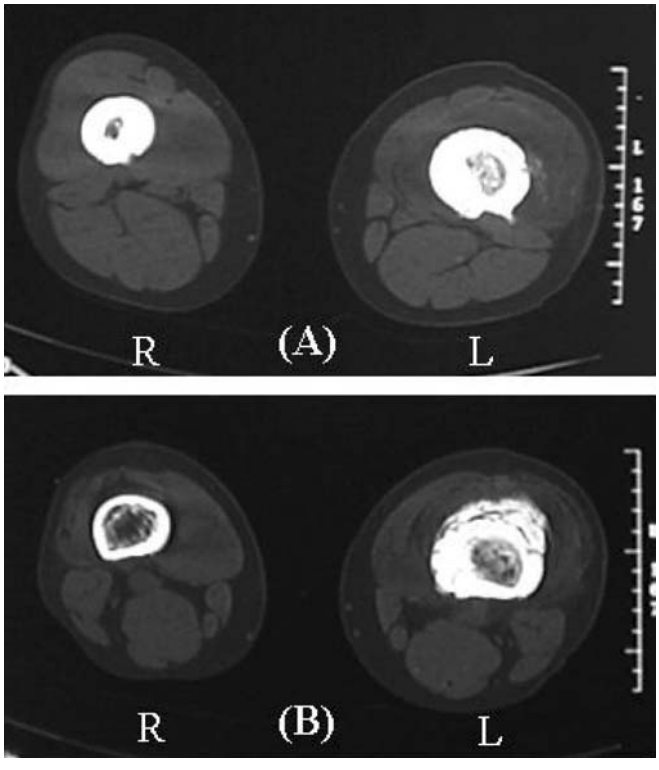

Fig. 5A,B. Axial computed tomography scan obtained at the age of 45 years. **A** Cortical thickening was detected in bilateral femora. **B** Laminar productive change was detected in the left femur on a distal slice

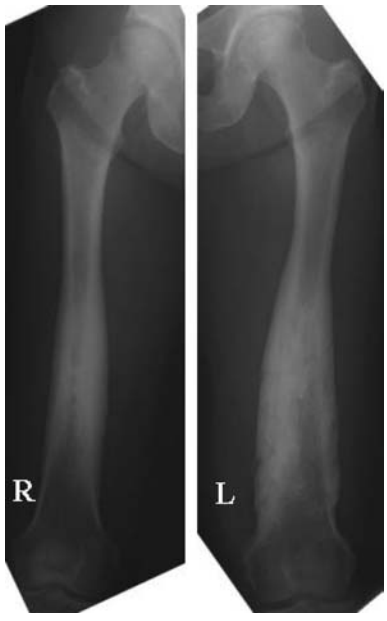

Fig. 6. Anteroposterior radiograph of the femora obtained at the age of 47 years showing progressive changes of the lesions compared with the previous radiographs

landin E-induced hyperostosis,[7] and Paget's disease[8] were considered for the differential diagnosis.

Although Ribbing disease may appear identical to other osteosclerosing diseases, radiographically many clinical differences exist. Camurati–Engelmann disease and Caffey disease present during childhood, whereas Ribbing disease presents in individuals of middle age. Camurati–Engelmann disease is bilateral and symmetric, whereas Ribbing disease is either unilateral or asymmetrically and asynchronously bilateral. Camurati–Engelmann disease affects long bones formed by intramembranous ossification; therefore, the skull is involved almost as frequently as the long bones.[9] Ribbing disease has been reported only in the long bones. The gait and neurologic abnormalities associated with Camurati–Engelmann disease are absent in Ribbing disease. In our patient only the bilateral femora were affected. The onset was at the age of 41 years, and the lesion of the left femur preceded the one in the right femur. Although most of the reported cases of Ribbing disease were siblings,[1,9–11] the course and physical findings of our patient were suggestive of a rare case of sporadic Ribbing disease.

Chronic osteomyelitis of bilateral femora was thought to be unlikely because the patient presented symmetrical lesions. On the other hand, multifocal periostitis, which is also a rare disease, is difficult to distinguish from sporadic cases of Ribbing disease. There was a possibility that our patient might have multifocal periostitis. Prostaglandin E1-induced hyperostosis and Paget's disease were also considered for the differential diagnosis. However, our patient had not been under treatment with prostaglandin; besides, the thickening of the cortex and sclerosis excluded the diagnosis of Paget's disease.

The lesions obscured in our patient have also been observed in patients with palmoplantar pustulosis (PPP) or

pustulotic psoriasis.[12–14] However, she has no skin problems such as acne or palmoplantar pustulosis. The bone scintigraphy showed a slightly increased uptake at the sternoclavicular joints, but she presented no related symptoms. Although treatment with NSAIDs has proved unsuccessful in most cases with PPP or pustulotic psoriasis, our patient was successfully treated with NSAIDs and etidronate disodium.

In summary, we presented the case of a 42-year-old Japanese woman whose clinical and radiological features did not completely fit the diagnosis of classical diaphyseal dysplasia. Although a sporadic case of Ribbing disease is the most likely diagnosis, no definite diagnosis has been established as yet.

References

1. Ribbing S. Hereditary, multiple diaphyseal sclerosis. Acta Radiol 1949;31:522–36.
2. Engelmann G. Ein Fall von Osteopathia Hyperostotica (sclerotisans) multiplex infantilis. Fortschr Geb Roentgenstr 1928;39:1101–6.
3. Caffey J. Infantile cortical hyperostosis; a review of the clinical and radiographic features. Proc R Soc Med 1957;50(5):347–54.
4. Kozlowski K, Masel J, Harbison S, Yu J. Multifocal chronic osteomyelitis of unknown etiology. Report of five cases. Pediatr Radiol 1983;13(3):130–6.
5. Stewart A, Carneiro R, Pollock L, Shaw D. Case report 834: Chronic sclerosing osteomyelitis of the mandible with long bone periostitis. Skelet Radiol 1994;23(3):225–7.
6. Kozlowski K, Anderson R, Tink A. Multifocal recurrent periostitis. Report of two cases. Rofo 1981;135(5):597–602.
7. Yoshida K, Oida H, Kobayashi T, Maruyama T, Tanaka M, Katayama T, et al. Stimulation of bone formation and prevention of bone loss by prostaglandin E EP4 receptor activation. Proc Natl Acad Sci USA 2002;99(7):4580–5.
8. Hadjipavlou AG, Gaitanis IN, Kontakis GM. Paget's disease of the bone and its management. J Bone Joint Surg Br 2002;84(2):160–9.
9. Shier CK, Krasicky GA, Ellis BI, Kottamasu SR. Ribbing's disease: radiographic-scintigraphic correlation and comparative analysis with Engelmann's disease. J Nucl Med 1987;28(2):244–8.
10. Paul LW. Hereditary multiple diaphyseal sclerosis (Ribbing). Radiology 1953;60(3):412–6.
11. Furia JP, Schwartz HS. Hereditary multiple diaphyseal sclerosis: a tumor simulator. Orthopedics 1990;13(11):1267–74.
12. Davies AM, Marino AJ, Evans N, Grimer RJ, Deshmukh N, Mangham DC. SAPHO syndrome: 20-year follow-up. Skeletal Radiol 1999;28:159–62.
13. Kasperczyk A, Freyschmidt J. Pustulotic arthroosteitis: spectrum of bone lesions with palmoplantar pustulosis. Radiology 1994; 191(1):207–11.
14. Hyodoh K, Sugimoto H. Pustulotic arthro-osteitis: defining the radiologic spectrum of the disease. Semin Musculoskel Radiol, 2001;5:89–93.