Congenital osteofibrous dysplasia of the tibia, associated with pseudoarthrosis of the ipsilateral fibula

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ABSTRACT

يصف المؤلف حالة طفل ذكر، حديث الولادة، يعاني من ثدن ليفي عظمي خلقي في عظمة الظنبوب اليمني، وكان مصاحبا لها مفصل كاذب لعظمة الشظية في نفس الساق. تم تجريف الآفة ووضع مكانها طعم عظمي، تم أخذه من عظمة الشظية من الطرف الآخر. نتيجة التشريح المرضي للآفة كانت مطابقة للثدن الليفي العظمي. لم يتم التدخل على المفصل الكاذب في الشظية في نفس الطرف وتمت مراقبته فقط. بمتابعة المريض لمدة سبع سنوات كان هناك شفاء وظيفي جيد للآفة دون أي تنكس. وهذه الحالة تعتبر تقديم جديد للثدن الليفي العظمي الخلقي. وقد تم عرضها للفت الانتباه لهذه الحالة النادرة لكي تؤخذ بعين الاعتبار في التشخيص التفريقي في آفات الظنبوب الخلقية.

We describe an otherwise normal male neonate who presented shortly after birth with rare congenital osteofibrous dysplasia of the right tibia associated with pseudoarthrosis of the ipsilateral fibula. The lesion was curetted, and the defect was packed with a fibular bone graft from the other leg. Histopathological examination was typical for osteofibrous dysplasia. The ipsilateral fibular pseudoarthrosis was observed with no active intervention. Seven years follow-up showed good functional recovery without recurrence of the lesion. The case is a new presentation of congenital osteofibrous dysplasia, and is presented to draw attention to this rare condition that must be considered in the differential diagnosis of congenital lesions of the tibia.

Saudi Med J 2008; Vol. 29 (10): 1507-1509

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Received 27th May 2008. Accepted 18th August 2008.

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Osteofibrous dysplasia is a rare fibro-osseous lesion of bone affecting the tibia and rarely the fibula of children, mainly during the first decade of life.[1,2] The most frequent presentation occurs after minor trauma with symptoms of a swelling of the tibia or fibula that may be painful. Pathological fracture and limp are also other occasional presentations.[3,4] The reported cases of osteofibrous dysplasia in the English literature are countable. A few cases were reported in neonates and congenital cases are extremely rare.[2-8] In the present report, we describe an otherwise normal male neonate who presented shortly after birth with congenital osteofibrous dysplasia of the right tibia, associated with ipsilateral fibular pseudoarthrosis. This case is presented to be added to the previously reported congenital cases, and to draw attention to the unique natural history of this unusual lesion, and its similarity in radiographic appearance to other tumors.

Case Report. An 18-day-old male neonate was admitted to the Orthopedic Department of King Khalid University Hospital, Riyadh, Kingdom of Saudi Arabia because of pathological fracture in the right tibia. The baby was a product of full term, emergency cesarean section due to abruptio placenta with fetal distress for which he was admitted to the neonatal intensive care unit (NICU) for 3 days then discharged in a good condition. One day prior to admission, the mother noticed that he was crying during change of diaper particularly when she touched the right leg, which was not actively moving. The mother denied any history of trauma. Examination upon admission revealed pseudoparalysis of the right lower limb with tender swollen right leg, but no redness, or hotness. There was no fever or skin rash, and all vital signs were normal. General examination showed that the baby was otherwise, normal, active, and sucking well. Blood and urine analysis were normal. Radiological examination of the right leg (**Figure 1**) revealed a big oval lucent lesion at the junction between the upper and middle thirds of the right tibia. The lesion was cortical in location and expanding into the medulla with thinning of the anterior cortex, which was cracked. The margins were well corticated, and there was no associated soft tissue swelling. A narrowed sclerosed segment at the junction of the middle and lower thirds of the ipsilateral

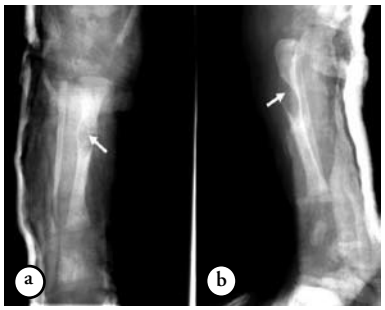

Figure 1 - Anterior posterior **a**) and lateral **b**) views of the right tibia at time of presentation, showing intracortical osteolytic lesion (white arrow) with a crack in the anterior cortex.

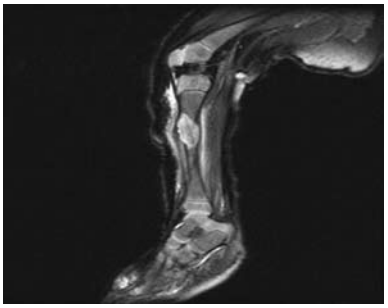

Figure 2 - Sagittal T1 contrast enhanced MRI right tibia, showing a well defined mildly expancile eccentric cortical osteolytic enhancing lesion at right proximal tibial diaphysis, mild overlying periostitis and soft tissue reaction, and narrow zone of transition.

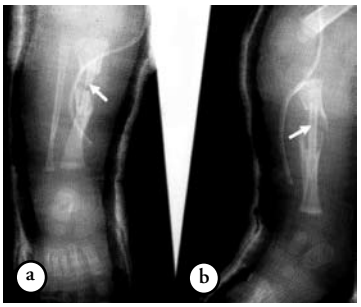

Figure 3 - Postoperative anterior posterior **a**) and lateral views **b**) of the right tibia after excision of the lesion, showing the strut fibular graft (white arrow).

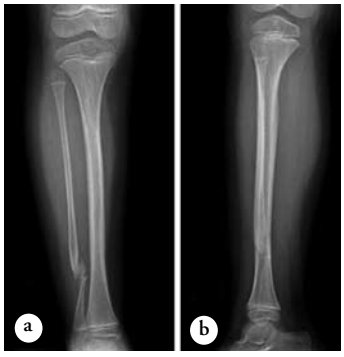

Figure 4 - Anterior posterior **a**) and lateral **b**) views of the right tibia 7 years after excision of the lesion.

fibula was also noticed. This segment had developed spontaneously to frank pseudoarthrosis at the age of 5 years. Reviewing the skeletal survey, which was taken for the baby a few hours after birth upon his admission to NICU showed the same lesion. This indicated that the condition is congenital. Because the lesion looked benign radiologically, a provisional diagnosis was suggested of monostotic fibrous dysplasia and less likely non-ossifying fibroma. Neonatal osteomyelitis was excluded by history, clinical, radiological examination, and blood tests. An above knee back slab was applied to the affected limb and the child was sent home. The mother was asked to observe the temperature and the activity of her baby and to attend the weekly clinic for clinical evaluation. After 4 weeks, a repeat x-ray revealed a slight increase in the size of the lesion with no attempt for healing of the fracture. Clinically, the leg was still tender and swollen. It was decided to curette the lesion and to fill the cavity with a fibular graft from the other leg. Bone and gallium scan as well as MRI (**Figure 2**) were requested prior to surgery. The results excluded infection and supported the same provisional diagnosis. During surgery, the tumor was found soft in consistency with some firm areas, eroding the cortex and extending into the medulla. There was no soft tissue extension. After completing the curettage, the cavity left was so large that using a strut fibular graft was the best available option. The baby tolerated the surgery well, and his limb was immobilized in a back slab. Postoperative x-rays (**Figure 3**) were satisfactory. Histopathological examination showed a benign bone lesion consisting of proliferating fibrous connective tissue that contains numerous stellate and spindle shaped fibroblasts, which are set in a loose connective tissue stroma. In addition, there were bone spicules that were lined by active osteoblasts. At the periphery of the lesion, there was new bone formation with numerous osteoclasts and osteoblasts. The histopathological findings were typical for osteofibrous dysplasia and immunohistochemical stains showed cytokeratin-positive cells, but hyperchromatic epithelial islands characteristic of adamantinoma was not found. Another experienced pathologist from another institute confirmed the diagnosis. The child was regularly followed up until the age of 7 years. The fibular pseudoarthrosis was treated expectantly, and it united spontaneously 2 years after its appearance. The child had normal life style for age, running and playing normally with no complaints from his right leg. In the latest x-rays (**Figure 4**), the right tibia looked slightly thinner, but straight, and equal in length to the other tibia.

Discussion. Osteofibrous dysplasia is a benign, slowly progressive lesion, which generally involves the tibia and rarely the fibula of an infant or a young child. Affections of the ulna, radius, and humerus were reported as well.[1,2,9] Osteofibrous dysplasia is identical in histological appearance to a well-recognized tumor of mandible and facial bones named ossifying fibroma. Indeed, osteofibrous dysplasia appeared formerly in the literature under ossifying fibroma of the long bones.[3,4,7,10] The main differential diagnosis for osteofibrous dysplasia includes fibrous dysplasia and differentiated adamantinoma.[1,3,7] Certain overlapping clinical features (including the location in the tibia and, less often, the fibula) and the morphologic similarities of many areas of osteofibrous dysplasia and differentiated adamantinoma (particularly the presence of cytokeratin-positive cells and scattered epithelioid islands typical of adamantinoma) suggest a more than coincidental association between osteofibrous dysplasia and differentiated adamantinoma.[3,11,12] The clinical course of the reported case did not suggest the diagnosis of adamantinoma at any stage and the histopathological and immunohistochemical studies were clear and conclusive for osteofibrous dysplasia. Osteofibrous dysplasia has been considered as a variant of fibrous dysplasia.[10] However, it can be distinguished from monostotic fibrous dysplasia by its clinical course and its intracortical location, as demonstrated radiographically. Histologically, the trabeculae in osteofibrous dysplasia are lamellar or have a lamellar surface and show osteoblastic rimming with slightly myxoid stroma less heavily collagenized than usually encountered in intramedullary fibrous dysplasia.[1,11,12] In the present case, histopathological examination was the key for the diagnosis of osteofibrous dysplasia.

The case reported here, being congenital, is different in the natural history from ordinary osteofibrous dysplasia. Furthermore, it is different from previously reported congenital cases as well. The first reported congenital case presented with tibia vara and a firm mass that was osteofibrous dysplasia.[4] The second presented with congenital pseudoarthrosis of the tibia and fibula with underlying osteofibrous dysplasia.[8] A third case was reported as congenital fibrous defect of the tibia,[13] its description was very close to osteofibrous dysplasia that was not popular at the time of publishing this case. The case reported here has the 2 pathologies of osteofibrous dysplasia and pseudoarthrosis, but each in a different bone. It can therefore be considered a new presentation of this rare recently recognized congenital association. Previous studies stated that the presence of pseudoarthrosis does not affect the treatment result of osteofibrous dysplasia, which is usually good.[1,3,14] This observation is true in the currently reported case.

Osteofibrous dysplasia is thought to follow a slowly progressive course and to stabilize after skeletal maturity, but it recurs frequently after curettage or subperiosteal resection.[2,3,5,7] Treatment was controversial. Some authors were advocating radical surgery, but others stated that surgery should be delayed as long as possible and should be restricted to extensive lesions or in the presence of pathological fractures.[1,6,9,10] Although the diagnosis of the present case was indefinite at the time of surgery, the radical surgical excision that was performed might have been a good treatment option as the lesion was extensive and was associated with pathological fracture.

In conclusion, although congenital osteofibrous dysplasia is an extremely rare condition, it must be considered in the differential diagnosis of any congenital or neonatal tibial or fibular osteolytic lesion. Careful radiological examination can distinguish congenital osteofibrous dysplasia from the more commonly occurring fibrous dysplasia. Diagnosis of congenital osteofibrous dysplasia must draw attention to the possible association of pseudoarthrosis. Treatment of congenital osteofibrous dysplasia follows the same regimen of treating the lesion when affecting the long bone in children and adolescents.

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