PACHYDYSOSTOSIS OF THE FIBULA

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We report four patients with unilateral bowing of the lower leg, affecting only the fibula. The bone is too long with anterolateral curvature of the distal third. Because of its regressive course and the absence of cutaneous involvement, this newly described entity can be distinguished from other forms of bowing of the leg.

Unilateral bowing of the lower leg is not infrequent, but generally the tibia and fibula are both involved. We report on a form of bowing affecting only the fibula which is enlarged and elongated. We have found four similar cases suggesting that this type of dysostosis represents a separate entity for which we propose the name ‘fibular pachydyosostosis’ (from the Greek work ‘pachy’, meaning broad) since enlargement of the bone is a characteristic feature.

PATIENTS

Case 1. A two-month-old girl was referred to our clinic with bowing of the left lower leg first noticed at the age of ten days. The pregnancy was unremarkable except for cerclage of the cervix. The child was born normally at term, weighed 3.06 kg and measured 50 cm in length. The parents were not consanguineous and had no physical abnormality. A brother born three years later was normal. The only abnormality noticed on clinical examination was bowing of the lower third of the left leg with a lateral convexity. There was no shortening of the affected limb and no limitation of joint movement. No cutaneous abnormality was seen (Fig. 1a).

Radiological examination at the age of seven weeks showed severe anterolateral curvature of the lower third of the fibula and an enlargement of this region, especially visible on the lateral views. The lower end of the tibia was slightly deviated medially (Fig. 1b). There was also a doubling of the femoral and tibial cortices which is normal at this age. A CT scan was performed on suspicion of an invading fibroma but there was no abnormality of the surrounding soft tissue. The patient has been followed up regularly, and a gradual regression of the deformity observed. When the patient was last seen at the age of 4.9 years, only the slightest anterolateral convexity remained. The position of the foot was neutral and the growth of the left lower limb normal. She measured 114 cm and weighed 21 kg. Radiographs confirmed the slight deformation of the two bones and showed persistent enlargement of the middle and lower thirds of the fibula (Fig. 1c).

Case 2. A four-week-old boy was referred with congenital bowing of the right lower limb. The pregnancy had been normal and the baby weighed 3.9 kg and measured 52 cm at birth. He had an exchange transfusion for neonatal jaundice. He is the only child of healthy non-consanguineous parents. Clinical examination was normal except for bowing of the right lower leg and slight varus of the foot. Radiological examination showed bowing of the fibula with a dorsal convexity and enlargement of the bone with somewhat irregular contours (Fig. 2a). The CT scan revealed no abnormality of the surrounding soft tissue. The patient has been seen regularly and there has been progressive remodelling. At the age of 8.4 years the leg was almost normal. Radiological examination showed minimal persistence of bowing and a slight enlargement of the lower third of the fibula (Fig. 2b).

Case 3. A four-year-old boy was referred to us with congenital elongation of the fibula noticed at birth because of bowing of the left lower limb with dorsolateral convexity. The pregnancy had been uneventful except for treatment with progesterone. The parents are normal and non-consanguineous. At the age of four years there was no deviation of the lower leg but a slight enlargement of the lower third of the fibula. Radiological examination at the age of three weeks had shown bowing and enlargement of the bone very similar to the preceding
Case 1. Figures 1a and b – Bowing of the left lower leg in a three-month-old girl. Note the curvature of the fibula. Figure 1c – The deformity has regressed after five years.

Case 2. Bowing and enlargement of fibula at one month (a). At nine years of age (b), the leg is clinically almost normal.
Case 3. Radiographs taken one month after birth (a), and at four years of age (b).

Fig. 3a

Fig. 3b

case (Fig. 3a). At the age of four years these features had regressed (Fig. 3b).

Case 4. An 18-month-old girl was referred with bowing of the lower leg noticed at the age of 16 months. She is the second child of healthy and non-consanguineous parents. Her elder brother is normal. Clinically, growth was normal (weight 11.8 kg, height 80 cm). The left lower leg was deformed by outward bowing with no shortening of the limb. The patient had no other abnormality except for camptodactyly of the fifth finger, also found in her father. There was no skin abnormality. Radiological examination showed bowing of the fibula with proportional elongation and enlargement of the lower third (Fig. 4a). At the age of 16 years, all the features had regressed (Fig. 4b).

DISCUSSION

Congenital bowing of the lower leg is a relatively frequent abnormality. Apart from deformation secondary to the absence of the fibula, two types can be distinguished. The first with an anterolateral convexity is frequently accompanied by radiological changes in the tibia with the appearance of a lytic lesion at the apex of the convexity. This form has a rather unfavourable prognosis due to the risk of pseudarthrosis (Dal Monte et al 1987) and its consequent difficulties for surgical treatment. It can also be a symptom of neurofibromatosis (Hunt and Pugh 1961), evidence for which requires a systematic search; the diagnosis can be difficult at birth as skin lesions may not be visible.

A second type of bowing with a dorsal convexity affects the lower third of the two bones. The prognosis is
favourable and spontaneous regression usually occurs with growth (Pappas 1984; Carlioz and Langlais 1986). Neither of these types of bowing correspond to the clinical picture of our patients. Unlike the first type, our cases showed more dorsal bowing and a favourable progression. We could find no sign of neurofibromatosis at birth nor subsequently. The second type of bowing resembles our observations more closely, but the apex of the curve was more distal in our cases. Moreover, fibular elongation or enlargement have never been described. These deformations of the fibula could suggest fibrous dysplasia (Stewart, Gilmer and Edmonson 1962), but the congenital manifestation of the lesion, the elongation of the bone, the regressive course, the isolated involvement of the fibula and the absence of any cutaneous signs, allow this hypothesis to be excluded. Initially, we also considered the possibility of an invading fibroma, but the CT scan, and again the favourable course, preclude this diagnosis.

We have not been able to find a previous report of an analogous case. A new syndrome with multiple congenital malformations, an abnormal (serpentine) fibula close to our description was described by Exner in 1988 but the fibula had more undulations, and other malformations were present (polycystic kidney and facial dysmorphism). We think that our clinical picture is different and, appearing in four cases, would seem to constitute a separate entity. Awareness of this relatively benign dysostosis will help avoid confusion with more severe disorders.

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REFERENCES