Pediatr Radiol (2003) 33: 7–10
DOI 10.1007/s00247-002-0660-4

CASE REPORT

Benjamin Joseph
Renjit A. Varghese

Congenital distal humeral dysplasia: a case report

Received: 2 September 2000
Accepted: 30 November 2001
Published online: 12 April 2002

B. Joseph (✉) · R.A. Varghese
Department of Orthopaedics,
Kasturba Medical College, Manipal 576119,
Karnataka State, India
E-mail: bjoseph-vip@zetainfotech.com
Tel.: +91-8252-70013
Fax: +91-8252-70062

Abstract Congenital dysplasia of the humerus is very rare. It is characteristically seen in omodysplasia and has also been reported as one of the associated features of Larsen's syndrome. We report a 4-year-old girl with bilateral humero-ulnar dysplasia, with dislocation of the elbows, facial dysmorphism, ball-and-socket ankles and foot deformities. Although the elbow dysplasia is similar to that seen in Larsen's syndrome, other pathognomic features of Larsen's syndrome were absent. The changes seen in the elbows in this patient are also different from those encountered in omodysplasia. We believe that this condition may be a distinct form of skeletal dysplasia hitherto undescribed.

Keywords Humerus · Dysplasia · Larsen's syndrome · Omodysplasia

Introduction

Congenital dysplasia of the humerus is very rare [1]. Dysplasia of the distal humerus has been reported as being one of the associated features of Larsen's syndrome [2, 3] and is also a characteristic feature of omodysplasia [4, 5, 6]. We report a 4-year-old girl who presented with facial dysmorphism, bilateral humero-ulnar dysplasia with dislocation of the elbows, ball-and-socket ankles and deformities of the feet, without evidence of Larsen's syndrome or the classic changes of omodysplasia.

Case report

A 4-year-old girl presented with deformity of the elbows and feet, which was noticed by her parents since birth. She was the fourth child of a non-consanguineous couple. The pregnancy and delivery were normal. All three older siblings were normal and there was no family history of any similar deformity or disorder.

Clinical examination revealed short stature, facial dysmorphism (flattened facies with a depressed nasal bridge and hypertelorism; Fig. 1) and a high arched palate. There was no ligamentous laxity. The spine was normal. The musculature of the upper limbs was poorly developed and the limbs appeared tubular. Fixed flexion deformity of 50° of the right elbow and 40° of the left elbow were present. Further flexion to 110° was possible bilaterally. The shoulders, forearms, wrists and hands were clinically normal. In the lower limbs, the hips and knees were normal. There was planovalgus deformity of both feet with forefoot abduction (Fig. 2). Both Achilles tendons were tight. The great toe was smaller than the second toe in both feet. Serial radiographs of the elbows from birth were available, revealing dysplasia of the distal humerus with the medial side more involved than the lateral (Fig. 3). The ulna was dislocated anteriorly and, on the most recent radiograph (Fig. 3d), an abnormal ossific centre was noted in the anterior aspect of the arm just proximal to the dislocated ulna. The radial head was in normal alignment to the capitellum, which itself appeared to be tilted anteriorly. At the ankle, a ball-and-socket type of joint was noted bilaterally (Fig. 4). No extra ossification centres were noted in the carpal bones or the calcaneum. The spine (Fig. 5) and pelvis were normal.

Discussion

The facial dysmorphism and elbow changes noted in this child appear to be similar to those seen in Larsen's

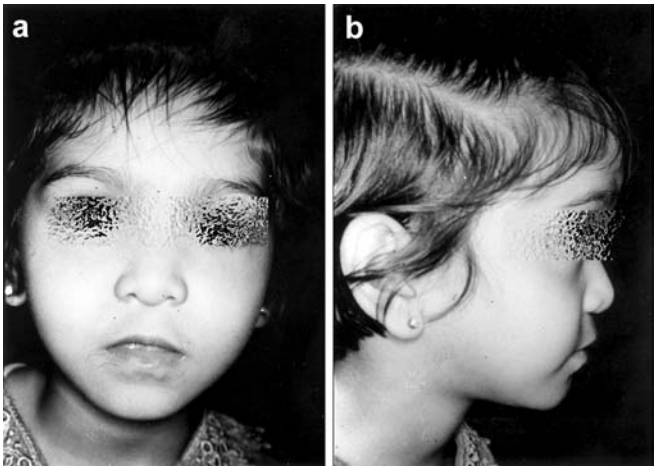

Fig. 1a, b Facial features of the child. The flattened face with hypertelorism and depressed nasal bridge are evident

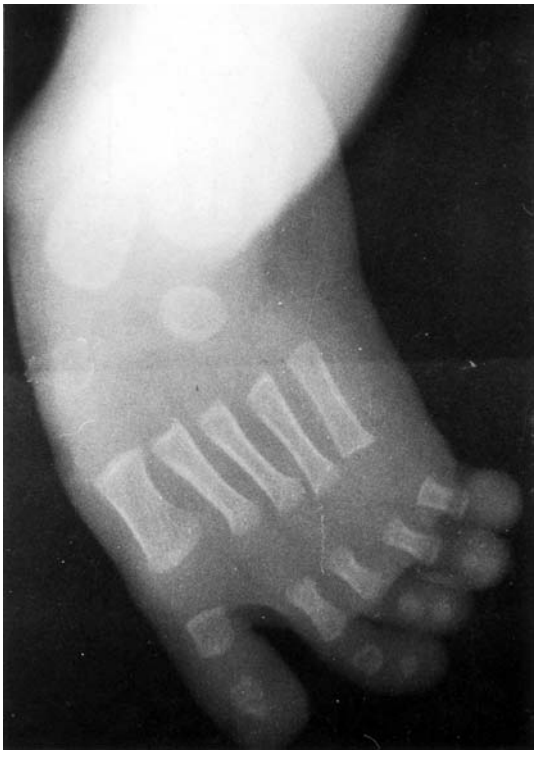

Fig. 2 AP radiograph of the foot showing absence of accessory tarsal bones. Abduction of the forefoot is evident

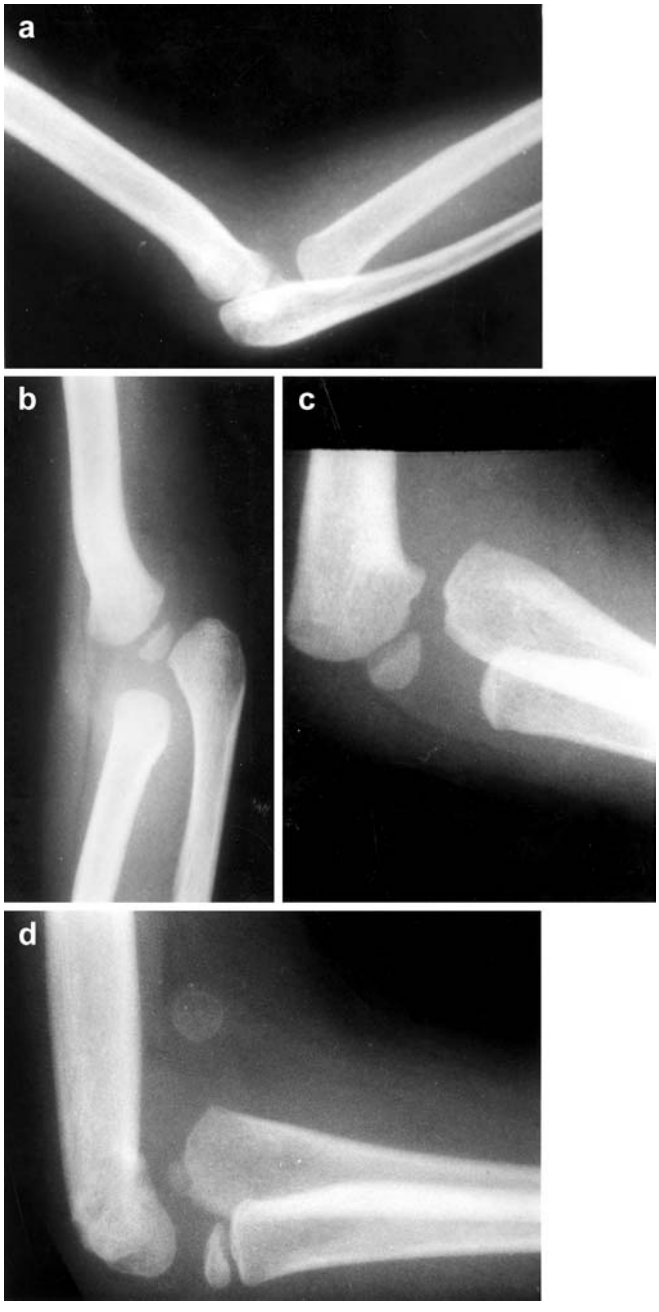

Fig. 3a–d Serial radiographs of the elbow. **a** Lateral view at 1 year of age. **b** AP and **c** lateral views at 2 years of age. **d** Lateral view at the age of 4 years showing anterior dislocation of the ulna, abnormal angulation of the capitellum and an abnormal ossification centre

syndrome [3]. The similarities in the elbow include dysplasia of the distal end of the humerus, anterior angulation of the capitellum, presence of abnormal ossicles around the elbow joint and anterior dislocation of the humero-ulnar joint. However, the other pathognomic features of Larsen's syndrome, which include joint laxity, multiple joint dislocations [2, 3, 7, 8, 9, 10, 11, 12, 13], spatulate thumb [2, 3, 7, 8, 9, 13], cylindrical fingers [2, 3, 7, 8, 9, 11, 12, 13], spinal anomalies [2, 3, 7, 8, 9, 10, 12, 13], and abnormal ossification centres around the wrist and feet [2, 3, 7, 8, 9, 12, 13, 14], were all absent in this child. Thus, although the child had some features resembling

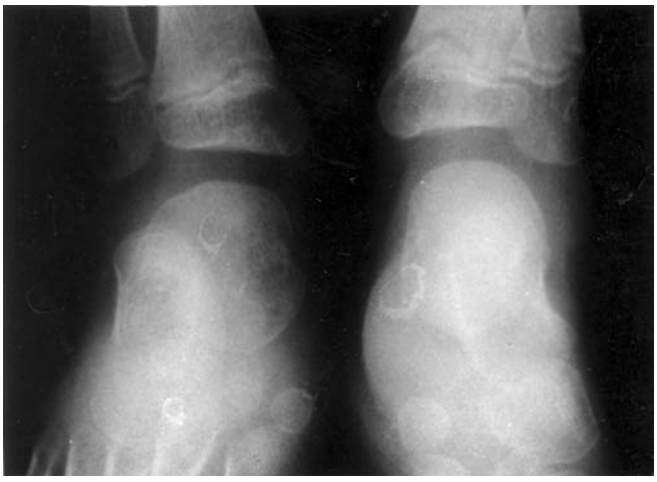

Fig. 4 AP radiograph of the ankles showing ball-and-socket type joints

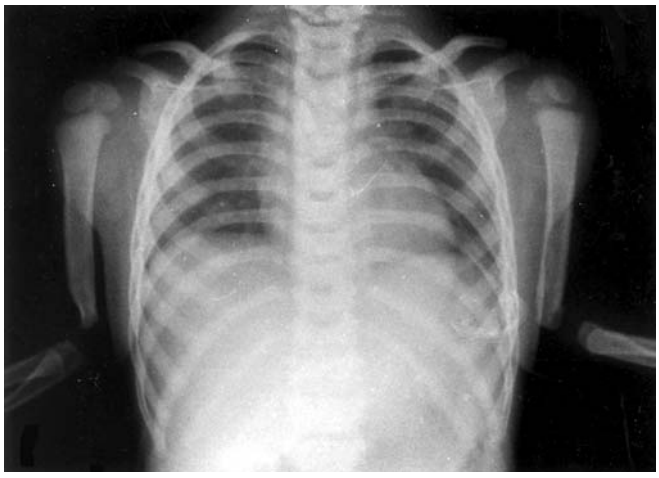

Fig. 5 Radiograph of the chest and upper limbs showing distal humeral dysplasia with no spinal involvement

Larsen's syndrome, she could not be placed under this umbrella because many of the pathognomic features of this syndrome were absent.

The other possibility was that we were dealing with a form of omodysplasia, first reported by Maroteaux et al. [6]. The autosomal dominant form of omodysplasia is characterised by defective growth of the distal humerus with anterolateral dislocation of the radial head. The findings in our patient differed from those seen in omodysplasia in that the ulna was dislocated but the radial head was not. The additional ossicle noted in this patient and also seen in Larsen's syndrome has not been described in omodysplasia. Clearly, our patient had no similarity to the autosomal recessive form of severe micromelic dysplasia in which dysplasia of the distal humerus is also seen [4, 5, 6].

Review of the serial radiographs of the elbows from birth (Fig. 3) enabled us to accurately record the evolution of the bony changes that occurred with growth. At birth, the relationship of the distal humerus with the radius and ulna could not be clearly ascertained. At 1 1/2 years of age, dysplasia of the distal humerus was noted and this progressively increased with age. The medial aspect of the distal end of the humerus was more involved as compared to the lateral side. The capitellum was displaced more anteriorly and the ulna more medially. By the age of 4 years, the ulna was dislocated anteriorly and the capitellum angulated further anteriorly, although the axis of the radio-capitellar joint was maintained. An abnormal ossicle was noted anterior to the junction of the middle and distal third of the humerus (Fig. 3d). This accessory ossicle was not evident at birth but ossified only after 3 years of age. Similarly, the changes in the ankle joint suggestive of a ball-and-socket ankle are subtle (Fig. 4). However, this is the extent of changes one can expect at 4 years and the full-blown features of a ball-and-socket ankle are likely to become apparent with future growth [15].

Despite the fixed flexion deformities of both elbows and the striking radiographic changes, the child had little functional disability in the upper limbs. She was able to perform most of the activities of daily living without difficulty. We therefore refrained from any active intervention for the elbows. For the deformities of the feet, bilateral tendoachilles lengthening was undertaken. At the time of the last follow-up, the feet were found to be plantigrade.

We are reporting this case because we believe that this form of congenital distal humeral dysplasia with facial dysmorphism and foot deformity has not been described in the English literature. Although some features encountered in this patient are distinctly different from those reported in omodysplasia, this may be another form of the syndrome. The other more remote possibility is that the skeletal changes in this child might be one end of the spectrum of Larsen's syndrome. The serial radiographs of the elbow joints provide insight into the evolution of the bony changes that occurred with growth in this condition, which we believe is valuable for future reference.

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