Pediatr Radiol (1998) 28: 117–119

Marcia F. Blacksin
Beth A. Pletcher
Miriam David

Osteogenesis imperfecta with joint contractures: Bruck syndrome

Received: 6 June 1997
Accepted: 17 July 1997

M. F. Blacksin (✉)
Department of Radiology,
University of Medicine and Dentistry of New Jersey, 150 Bergen St., Rm. C320, Newark, NJ 07103-2426, USA

B. A. Pletcher
Center for Human and Molecular Genetics, Department of Pediatrics,
University of Medicine and Dentistry of New Jersey, Newark, New Jersey, USA

M. David
Department of Radiology,
Newark-Beth Israel Medical Center,
Newark, New Jersey, USA

Abstract We describe an Egyptian boy with osteogenesis imperfecta who was born with thumb contractures and bilateral antecubital pterygia. He was seen at 16 months of age with femur and tibial fractures, thoracic vertebral compression fractures, scoliosis and Wormian bones. The findings are consistent with a diagnosis of Bruck syndrome.

Introduction

Osteogenesis imperfecta (OI) is a large family of bone diseases characterized by a defect in type I collagen production. One rare form of OI was described by Bruck as congenital joint contractures and brittle bones in 1897. There are only four reports in the literature of children or families with this disease, and none have been seen in the western hemisphere. We have seen an Egyptian boy with the clinical manifestations of this syndrome, and several new radiographic abnormalities.

Case report

The patient was born to healthy Egyptian parents who deny consanguinity. They arrived for consultation after their 16-month-old son sustained a femur fracture. The patient was born after an uneventful pregnancy and forceps delivery. At birth, he was noted to have thumb contractures and bilateral antecubital pterygia. Chromosome analysis demonstrated a normal 46 X, Y male. He has no siblings.

At 16 months, the child weighed 11.3 kg (50th percentile for age) and his head circumference was 49.5 cm (75–90th percentile for age). Physical exam revealed brachycephaly with downslanting palpebrae and normal sclerae. Dentition was normal in color and number. The facial features were mildly dysmorphic. The patient could not sit unsupported and did not ambulate.

A skeletal survey was performed. The skull was brachycephalic with multiple Wormian bones (Fig. 1 a). The cervical spine demonstrated an incomplete posterior arch of the C2 vertebra, and failure of segmentation at the C2–3 level (Fig. 1 b). Compression fractures of the T5 and T7 vertebral body and a right thoracic scoliosis were noted (Fig. 1 c). Abnormally "thick" and curved acromions were seen bilaterally (Fig. 1 d). There was curvature of the right radius and ulna (Fig. 1 e). Healing fractures of the right distal femur and proximal tibia were seen. Mild flexion contractures were present in both first interphalangeal joints of the hands. The feet were unremarkable.

Laboratory studies revealed normal thyroid and parathyroid hormone levels. Alkaline phosphatase levels were normal, but the calcium level was slightly elevated. This finding was felt to be clinically insignificant with a normal parathyroid hormone level.

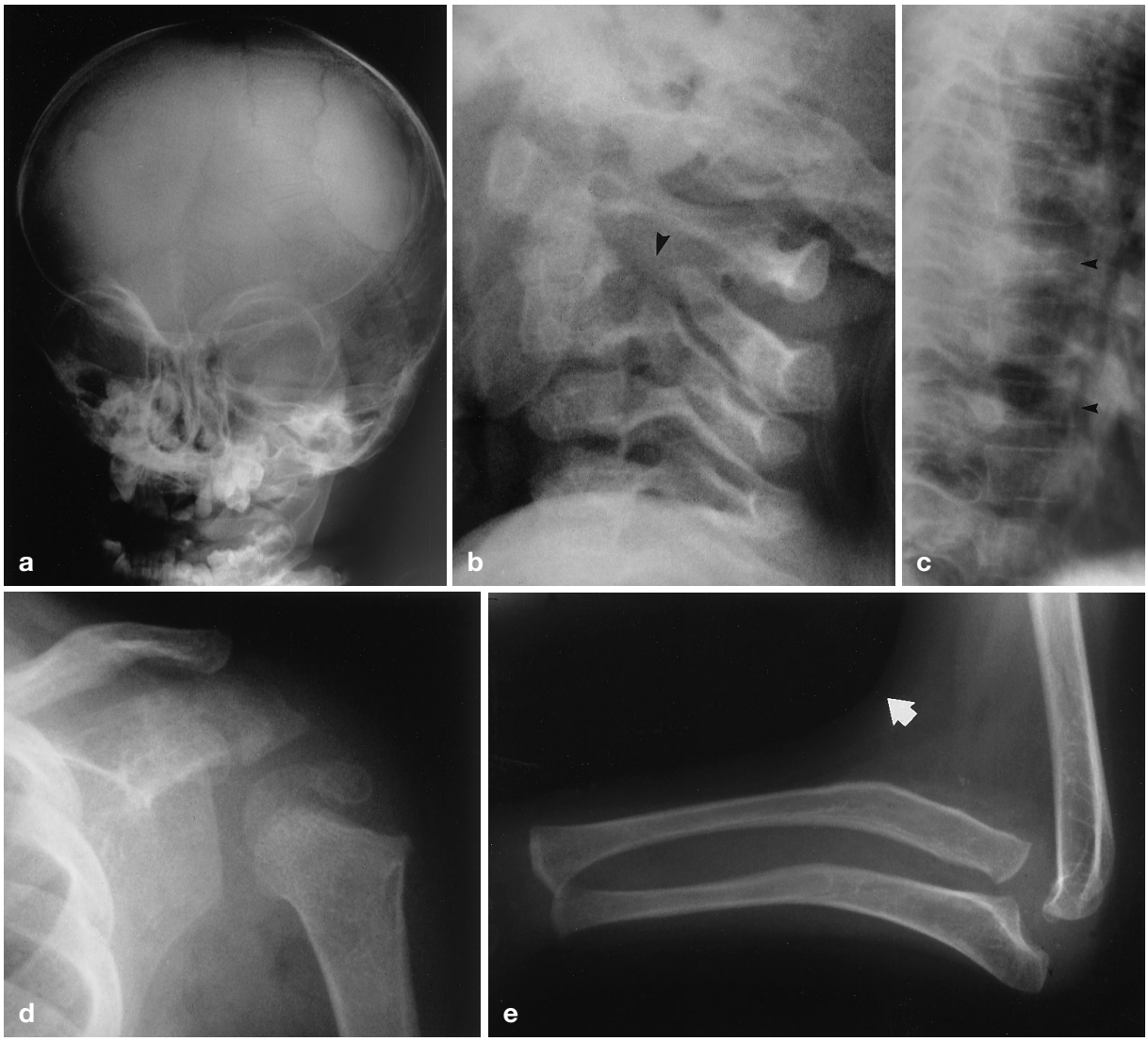

Fig. 1 a–e Skeletal survey in a 16-month-old boy with Bruck syndrome. **a** A frontal radiograph of the skull reveals Wormian bones. **b** A lateral radiograph of the cervical spine shows an incomplete posterior arch at the C2 level *(arrowhead)* with fused C2 and C3 vertebral bodies. **c** A lateral radiograph of the thoracic spine shows compression fractures of the T5 and T7 vertebral bodies *(arrowheads)*. **d** A frontal radiograph of the shoulder reveals a thickened acromion. **e** A lateral radiograph of the forearm shows abnormal curvature to the radius and ulna with an antecubital pterygia *(arrow)*

Discussion

There are only four other reports that describe this syndrome of multiple fractures and joint contractures [1–4]. Initially described by Bruck [1], the underlying disease is now thought to be a subtype of osteogenesis imperfecta. Brenner et al. [2] have done biochemical analysis of the compact bone in a patient with Bruck syndrome and found an underlying defect in fibril formation of collagen type I. There are few reports of the radiographic findings specific to these patients, and many are intially diagnosed with arthrogryposis until they begin to ambulate and fracture [4].

Viljoen et al. [3] has documented five patients in three South African families with this syndrome. The common radiographic finding seen in all of his cases, as well as a case by Sharma and Anand [4], was bilateral equinovarus. This finding was missing from our case and the case reported by Brenner et al [2]. All of the patients seen by Viljoen and Brenner had a scoliosis or kyphoscoliosis, as did our case. Wormian bones were seen in at least 2/5 of Viljoen's patients, Brenner's case and our case. Multiple fractures were also reported in all cases. Though the Wormian bones and kyphoscoliosis are typical of OI, particularly types III and IV, the clubfeet are not [5].

Our patient also revealed findings that were unreported in other cases. The patient possessed bilaterally thick, hooked acromions which may have appeared this way because of repetitive fractures. It is unlikely that this patient could have fractured both acromions, and it is felt that the findings are part of the patient's syndrome. Another atypical radiographic finding was a block vertebral body at the C2–3 level and an incomplete posterior arch at C2. These findings have not been reported with OI, and may be peculiar to this syndrome.

Joint contractures in Bruck syndrome are usually bilateral and symmetrical; they are seen in both the large and small joints. If the clubfeet have resulted from ankle joint and foot contractures, then the ankle is the most common site of contracture. This is followed by the knee and elbows in order of frequency. Osteogenesis imperfecta patients can manifest asymmetrical joint contractures after fracture, but Bruck syndrome patients demonstrate the contractures before fractures are seen [3]. Radiographs done just after the birth of our patient demonstrated no evidence of fractures, but bilateral antecubital pterygia suggested early prenatal contractures.

The phenotypic appearance of patients with Bruck syndrome also differs. In the reports by Viljoen and Brenner, as well as our report, sclerae were white [2, 3]. Sharma reported blue sclerae [4]. The presence of dentinogenesis imperfecta was noted by Brenner, but was absent in our patient and Viljoen's patients [2, 3].

Brenner et al. did electron microsocpy on the cortical bone of their patient and found the typical morphologic characteristics of OI. Their biochemical studies, however, did not show changes typical for patients with OI type III or IV, and they concluded that Bruck syndrome is an atypical OI variant [2]. Given the differences in both the phenotypic and radiographic appearance of these patients, it is not unreasonable to assume that there is some heterogeneity among even those patients considered to have Bruck syndrome. Further biochemical and molecular analysis of patients with contractures and osteogenesis imperfecta will answer these questions in the future.

References

1. Bruck A (1897) Multiple fractures associated with joint ankylosis and muscle atrophy. Dtsch Med Wochenschr 23: 152–155
2. Brenner RE, Vetter U, Stoss H, Muller PK, Teller WM (1992) Defective collagen formation and mineralization in osteogenesis imperfecta with congenital joint contractures (Bruck syndrome). Eur J Pediatr 152: 505–508
3. Viljoen D, Versfeld G, Beighton P (1989) Osteogenesis imperfecta with congenital joint contractures (Bruck syndrome). Clin Genet 36: 122–126
4. Sharma NL, Anand JS (1964) Osteogenesis imperfecta with arthrogryposis multiplex congenita. Indian Med J 53: 124–126
5. Sillence D (1981) Osteogenesis imperfecta: an expanding panorama of variants. Clin Orthop 159: 11–25