Bruck Syndrome: Arthrogryposis Multiplex Congenita With Osteogenesis Imperfecta

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ABSTRACT

We report a 4 year-old boy who presented in infancy with multiple joint contractures and subsequently with pathological fractures and bowing of the femur when he started to walk. A 6 year-old sister had similar features.

INTRODUCTION

A combination of arthrogryposis multiplex congenita and osteogenesis imperfecta constitute a disorder called Bruck syndrome. This syndrome is extremely rare and only a few cases have been reported in the literature. To the best of our knowledge, it has not been reported in the South East Asian region.

CASE REPORT

A 9-month old boy presented with bilateral knee contractures of 60 degrees and talipes equinovarus of the right foot. A diagnosis of arthrogryposis was made and serial splinting was started. There was minimal improvement and at the age of one year, a release of both knee contractures was done. Three months later, the right foot deformity was corrected by a postero-medial soft tissue release. Despite continued splintage, the left knee contracture recurred and a second release was done at the age of one and a half years. He started to stand and walk only at the age of three years. It was at this time that he sustained a fracture of the upper third of the right femur without any obvious trauma. An x-ray of the femur showed demineralised bone, severe deformity and cystic changes with a pathological fracture at the upper third and a healing fracture at the lower third (Figure 1). He did not have blue sclera but was of short stature and had kyphoscoliosis. Further questioning revealed that he has a six-year old sister with similar problems, though not so severe. When examined, she was also extremely short, had kyphoscoliosis and also contractures of both knees (Figure 2).

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DISCUSSION

Arthrogryposis multiplex congenita and osteogenesis imperfecta are both uncommon conditions. In combination, they are extremely rare and constitute the disorder called Bruck syndrome. The features of arthrogryposis are more obvious at infancy and we did not diagnose the osteogenesis imperfecta component until the patient had a pathological fracture. The left knee contracture also had to be done twice, showing the resistant nature of the soft tissues involved.

Viljoen[1] described five children with symmetrical joint contractures who later showed features of osteogenesis imperfecta when they started walking. He designated it as "Bruck syndrome" after a similar case documented by Alfred Bruck in 1897. Subsequent cases were reported[2,3,4,5] with patients showing flexion contractures, short stature, kyphoscoliosis and multiple recurrent fractures. Some had blue sclera, dentinogenesis imperfecta and also hearing loss. X-rays typically showed osteoporosis, severe deformity especially bowing, and cystic changes at old fracture sites. Brenner[6] did biochemical studies on a patient and found that the underlying defect involved defective fibril-formation of collagen type I leading to an altered mineralisation of bone.

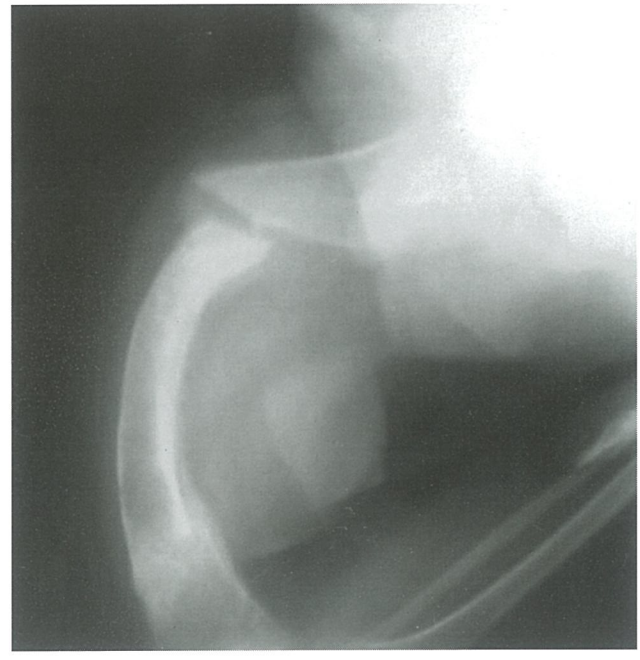

Figure 1. Radiograph showing multiple fractures and bowing of the femur in our patient.

Figure 2. The 4 year-old boy seen here with his 6 year-old sister

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