Picture story

A Sri Lankan child with fibrodysplasia ossificans progressiva

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Introduction

Fibrodysplasia ossificans progressiva (FOP) is a severe, rare condition of ectopic ossification with primary involvement of the skeletal muscles, associated with characteristic skeletal abnormalities[1]. A case of FOP was reported in this journal a few years back in an Indian child[2] but this is the first reported case in a Sri Lankan child.

Case report

A 4 year old girl, a product of a non consanguineous marriage, was admitted on 8th April, 2005 with a history of intermittent tender swellings over the back of the scalp, scapulae and neck of 3 months duration without any systemic symptoms. On examination, she had widely spaced teeth, hypoplastic thumbs, clinodactyly of the fifth finger, bilateral hallux valgus and a tender swelling over the back of the right scapulae and neck (Figures 1-3). There were restricted movements of the right shoulder joint. The rest of the systemic examination was normal.

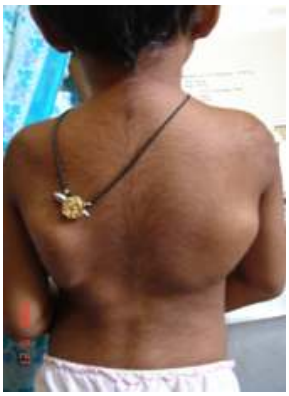

Figure 1 *Swelling over right scapula*

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Figure 2 *Hypoplastic thumbs & clinodactyly*

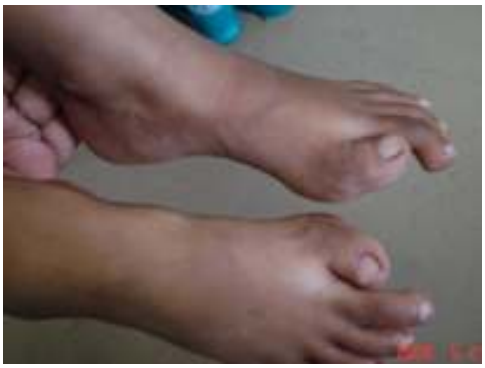

Figure 3 *Bilateral hallux valgus*

Initial investigations, including full blood count and ESR, were normal, and a CT scan of the lesion revealed a soft tissue swelling suggestive of a sarcoma. However, because of the remitting and resolving nature of the symptoms and the presence of dysmorphism, a diagnosis of FOP was entertained.

Discussion

FOP has autosomal dominant inheritance with complete penetrance but variable expressivity, and most cases result from a sporadic mutation[3]. Its incidence is approximately one case per 1.6 million inhabitants[4]. The disorder can occur in both sexes with preponderance towards male. It is mostly sporadic and high paternal age has also been

implicated. The natural history tends towards exacerbation and remission[5].

Pathogenesis is attributed to over expression of morphogenic protein in lymphoblastic cells, leading to defective regulation of endochondral osteogenesis[6]. The gene has been mapped to chromosome 4q27-31[7].

The clinical features include full penetrance for hallux valgus and variable expressivity for fibrodysplasia[5]. The onset of fibrodysplasia could be as early as fetal life or as late as 25 years, but the peak incidence is around 5 years[5]. They present as soft tissue swellings and the most common locations of initial involvement include neck, dorsal trunk, proximal limbs, sternocleidomastoid and masseter muscles[5]. The swelling could be associated with pain and fever. The natural history tends towards exacerbations and remissions, with endochondral ossification appearing within several months to years after the onset of fibrodysplasia[5]. Majority develops symptoms due to ossifications by 15 years[5]. The other associated features include short thumb, clinodactyly of the fifth finger, widely spaced teeth and short femoral necks[5].

Diagnosis depends on clinical features, x- ray evidence of abnormal soft tissue ossifications, ultrasound evidence of synovial enchondromatosis in large joints, tissue biopsy and chromosomal analysis[1].

There is little convincing evidence that any form of treatment alters the progress of the disease. Treatments that have been used include isotretinoin, oral corticosteroids and etidronate without proven benefit[8,9]. Unfortunately, despite treatment most become confined to wheelchair by third decade and succumbs to pulmonary complications in fifth or sixth decade[1].

References

1. Beighton P, editor. Fibrodysplasia ossificans progressiva. In: McKusick's Heritable disorders of connective tissue. St Louis: Mosby; 1993, 501–18.
2. Arulprakash S, Kain R, Chansoria M. A case of fibrodysplasia ossificans progressive. *Sri Lanka Journal of Child Health,* 2005; **34**:23-4.
3. Kaplan FS, Mc Cluskey W, Hahn G, Tabas JA, Muenke M, Zasloff MA. Genetic transmission of fibrodysplasia ossificans progressiva. *J Bone Joint Surg* 1993;**75A**:1214–20.
4. Connor JM, Evans OA. Genetic aspects of fibrodysplasia ossificans progressiva. *J Med Genet* 1982; **19**: 35-9.
5. Jones KL, editor. Smith's Recognizable Patterns of Human Malformations. 5th ed. Philadelphia: WB Saunders, 1997, 492-4.
6. Shafritz AB, ShoneEM, Gannon FH et al. Over expression of an osteogenic morphogen in fibrodysplasia ossificans progressiva. *N Engl J Med* 1996; **335** : 555-61.
7. Feldman G, Li M, Martin S, Urbanek M, Urtizberea JA, Fardeau M et al. Fibrodysplasia ossificans progressiva, a heritable disorder of severe heterotopic ossification, maps to human chromosome 4q27-31. *Am J Hum Genet* 2000; **66** : 128-35.
8. Zasloff MA, Rocke OM, Crofford LJ, Hahn GV, Kaplan FS. Treatment of patients who have fibrodysplasia ossificans progressiva with isotretinoin. *Clin Orthop Rel Res* 1998; **346**: 121-9.
9. Brantus JF, Meunier PJ. Effects of intravenous etidronate and oral corticosteroids in fibrodysplasia ossificans progressiva. *Clin Orthop Rel Res* 1998; **346**: 117-20.