

Fibrodysplasia Ossificans Progressiva

A 7-year-old boy presented with one year history of progressive multiple painful hard swellings mostly over dorsum of the trunk with restriction of neck, shoulder and hip movements. There was no family history of similar illness. Examination revealed multiple tender bony hard swellings over dorsum of the trunk (*Fig. 1*). There was bilateral short valgus hallux. Clinically, his hearing and other system examination was within normal limits. A diagnosis of Fibrodysplasia Ossificans Progressiva was made.

Fibrodysplasia Ossificans Progressiva is an autosomal dominant disorder of connective tissue, characterized by congenital malformation of the great toes and progressing disabling heterotpic osteogenesis. Typically the age of onset is around 3 to 4 years, with acute episodes of tender warm soft tissue swelling in the neck or upper spine. Exacerbations and remissions highlight the natural course of the disease. Flare-ups occur spontaneously or after minor trauma, biopsy, following intramuscular injections, which follow a specific pattern like dorso-ventral, axio-appendicular, cranio-caudal and proximal-distal gradients, leading on to heterotopic bone formation in the muscles and connective tissue with ankylosis of the joints. Almost all muscles, fascia, tendon and ligaments are involved except tongue, heart, larynx, diaphragm, sphincters, facial and extra ocular muscles. Therapeutic interventions are based



Fig. 1. Ossified swellings of back musculature

on preventive measures. Several drugs like Ethane-1-hydroxy-1-diphosphonate and isotretinoin have been used to counteract flare-ups and have shown equivocal results.

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