Fibrodysplasia ossificans progressiva – A rare case

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Abstract
Fibrodysplasia Ossificans Progressiva (FOP) also known as Myositis Ossificans Progressiva is a very rare and disabling genetic condition characterized by congenital malformation of the great toes with hallux valgus and progressive heterotopic ossification in specific anatomic pattern.¹ It is a rare condition with worldwide prevalence of about 1 case in 2 million individuals². No ethnic, racial, sex or geographical predisposition is noted. Myositis Ossificans Progressiva term is misnomer and not used now. The case report herein describes this rare condition in a child of one and half year age having the classical clinical presentation and radiological features. The child is under follow-up with measures to prevent fast progression of the condition.

Keywords: Fibrodysplasia, Ossificans, Progressiva, Myositis.

Blood investigations were Hb 10.9 gm/dL, serum protein 7.4gm%, albumin 4.8gm%, and globulin.
2.6gm%. All the parameters were within normal range except serum alkaline phosphatase raised to 702 IU/dL (normal being up to 302IU). X-ray of neck showed fluffy calcification of soft tissues on both the sides of neck. X-ray of shoulders showed dense calcification on inner aspects of arm extending from axilla to lower arm without involvement of humerus bone (Fig. 3). X-ray of spine showed dense calcification of paravertebral tissues in lumbar spine along with dense calcific cord extending obliquely from lumbar spine towards the rib cage (Fig. 4).

**Fig. 3:** Shows irregular ossification of soft tissues on medial aspect of both arms, more on right than left with mild scoliosis

**Fig. 4:** Shows cord like thickening extending bilaterally from L3 vertebrae towards lower ribs and irregular ossification of soft tissues in paravertebral areas

Due to absence of any effective treatment for the condition, the parents were explained the nature of disease and its likely course. A trial of bisphosphonate in the dose of 35 mg/week was started along with vitamin c to retard the process of soft tissue calcification. Avoidance of trauma in the form of massage, falls and intra-muscular injections was also explained. The child continues to be under follow up. Her pain is less, though activities remain limited.

**Discussion**

Fibrodysplasia Ossificans Progressiva (FOP) occurs due to spontaneous genetic mutation and is autosomal dominant in inheritance pattern. Progressive postnatal heterotopic ossification in FOP starts in the first decade of life spontaneously or follows minor trauma. It presents as painful swellings in soft connective tissue, including ligaments, tendons, skeletal muscles and ligaments. Its pathology involves immune system as suggested by presence of macrophages, lymphocytes and mast cells in early lesions, flare-ups following viral fever, and beneficial response in early flare-ups to corticosteroids. The genetic cause is identified as a mutation in the specific domain of activin receptor in all persons suffering with classic Fibrodysplasia ossificans progressive. Individuals with this condition appear normal at birth except for malformations of the great toes, which are seen in all the patients. Episodic painful swellings in soft tissue develop in preteen age. These swellings soon mature to bone. Minor trauma of intramuscular injections, muscle fatigue, bruises, or trauma due to fall may start a flare-up leading to progressive heterotopic ossification. Gradually most of the patients lose joint mobility and ambulation and become confined to wheelchair by third decade of life. Heterotopic ossification in FOP involves typically first in the dorsal, axial, cranial and proximal regions of the body and later in the ventral, appendicular, caudal and distal regions. Diaphragm, tongue, extraocular muscles and smooth muscles including heart are spared. Stiffness of neck may precede appearance of ossification in neck muscles. Anomalies in cervical vertebrae including fusion have been seen in many cases. Other congenital abnormalities include short malformed thumbs, clinodactyly, broad femur neck and osteochondroma on tibia. Severe weight loss due to ankylosed jaws, pneumonia and right heart failure may occur due to rigid fixed chest wall.

Progressive Osseous Heteroplasia (POH) characterized by cutaneous ossification besides soft tissue involvement, Non- Hereditary Heterotopic Ossification (NHHO) occurring following trauma to the localized site only, fibromatosis and lymphedema are common differential diagnosis. The correct diagnosis of FOP can be made clinically, even before radiographic evidence of heterotopic ossification is seen, if soft tissue lesions are associated with symmetrical malformations of the great toes. Biopsy in such cases can lead to rapid progression of the condition. Routine blood investigations are usually normal, though ESR and alkaline phosphatase values may be increased. Definite genetic testing for this condition is available helping in suspected cases. Hearing impairment may occur in 50% cases starting in childhood and progressing gradually. There is no definite treatment available for this condition, but a 4 days course of steroids started within 24 hours of a flare-up, may help reduce inflammation and edema seen in the early stages of the disease.
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Knowledge of Fibrodysplasia Ossificans Progressiva and its prognosis is important for an orthopedic surgeon. Though a very rare condition, it is autosomal dominant in inheritance. Physiotherapy, psychotherapy, genetic counseling, prevention and treatment of chest infections should be included in treatment besides pharmaceutical measures probably slowing down the progress of disease.

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References


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