Case Report

Myositis ossificans progressiva

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Myositis Ossificans Progressiva (MOP) is a very rare disease caused by heterotopic ossification of muscles and connective tissue. At present the preferred name is Fibrodysplasia Ossificans Progressiva (FOP). This disease is so rare that full spectrum of clinical features is yet to be established. This case has thoracic deformity, which is not found on careful review of literature. It is being reported to sensitize the physicians, so that the diagnosis may not be delayed. This case report also shows the rare association of thoracic deformity with MOP.

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Key words **: Thoracic deformity, fibrodysplasia ossificans progressiva.**

Myositis ossificans Progressiva is a rare autosomal dominant disorder of connective tissue characterized by congenital malformations and progressive ectopic calcification of striated muscles and connective tissue[1]. The point prevalence of MOP is approximately 1 per 2 million of the population worldwide with no racial, ethnic, sexual or geographic predisposition[1]. Only a few cases have been reported from India and around 700 cases have been reported till now in the literature[2,3]. It is such a rare disease that many of its features may remain unrecognized and unknown[4]. The case which is reported here has got a thoracic deformity in the form of depression of the left lower part of the anterior chest. MOP is always associated with a number of specific congenital bony deformities[1,3]. On careful review of literature, association of such congenital deformity of thoracic cage with MOP is not found[1-4]. This case is being reported to show the rare association of congenital thoracic deformity with the rare disease of MOP. This case report will also sensitize the physicians about this rare disease.

Case Report

An eleven and a half year old boy born to a non-consanguineous couple, presented with stiffness and painful restriction of movements of neck, trunk and limbs. He also complained of multiple hard swellings on neck, trunk and proximal limbs. The disease process started at the age of 3½ years. He first developed painful swelling of upper part of back including neck. This was associated with fever. Pain subsided after a few weeks. Diffuse induration persisted for several months and multiple hard swellings developed in the affected area over years. Such attacks occurred recurrently involving newer areas of trunk and proximal limbs. Gradually, the patient lost the ease of movements of the spine initially and limbs subsequently. Family history was negative.

Examination **—** On examination, the upright posture was characterized by fixed flexion and left lateral bending of the spine. The limbs were fixed at proximal joints (Fig 1). Gait was rather stiff with markedly diminished accessory movements. Movements of the distal small joints including ankles and wrists were, however, normal. There were multiple hard bony swellings on neck, trunk and proximal limbs (Fig 2). Great toes of both sides were significantly small (Fig 1). Thoracic deformity in the form of depression of the anterior wall of the chest on it's left lower part was remarkable (Fig 1).

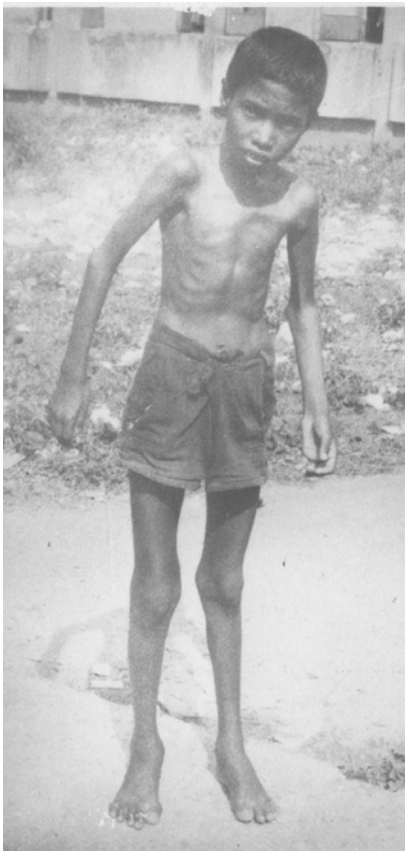

Fig 1 — Showing stiff posture, thoracic deformity and short great toes

Investigation — X-ray examination showed ossification in the soft tissues at multiple sites of cervical, dorsal, lumber regions of the trunk and proximal limbs. Serum biochemistry was normal (Calcium 9.25 mg/dl, Phosphate 5.5 mg/dl, Alkaline phosphate 80 U/L, CPK 45 U/L). Routine Blood Examination did not reveal any abnormality.

Treatment — Analgesics and corticosteroids were given during relapse for symptomatic relief.

Discussion

Fibrodysplasia ossificans progressiva (FOP) is the preferred

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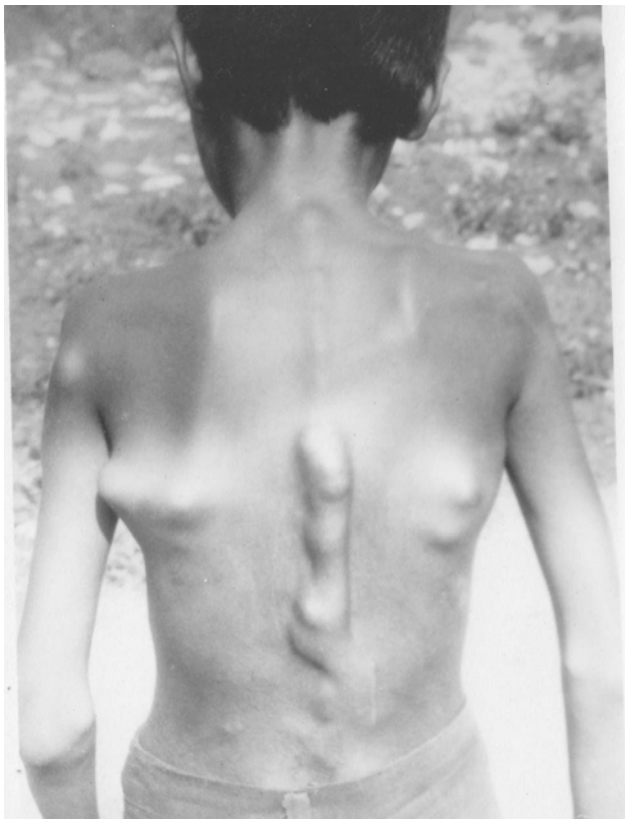

Fig 2 — Showing multiple hard bony swellings

name at present.[5] Spontaneous ossification of muscles and connective tissue associated with specific deformities of the great toes is diagnostic of MOP[1,3,6].

The condition is an autosomal dominant trait and most of the affected persons (95%) represent new mutation for the determinant gene, $ACVR_1$, chromosomal locus 2q23-24[7]. The identification of the mutant gene is possible by genetic linkage studies. Antenatal diagnosis is possible, but prevention of birth of affected babies may not be possible by antenatal screening as 95% of the cases are sporadic.

MOP usually manifests between birth and 10 years of age with a mean age of 3 years. Ectopic ossification is usually preceded by episodes of myositis. The process often starts at the neck and progresses gradually to involve the dorsal and the lumber regions of the trunk followed by proximal limbs. Ultimately, the child is encased in a rigid sheet with fixed posture. Ambulation is classically lost in the twenties and thirties due to ankylosis of hips. Hands, forearms and lower legs are usually spared. Major complications arise from rigidity of the rib cage and ankylosis of the jaw causing restrictive lung disease and nutritional impairment respectively[1,2,4].

People with MOP form skeletal deformities in two ways. Deformities which are produced during embryogenesis as a part of normotopic bone formation are congenital. Heterotopic ossification of muscles and connective tissue causes acquired deformity and develops after birth[1]. The majority of the patients of MOP are born with congenital bony malformations that include short hallux, microdactyly and clinodactyly of fingers, polydactyly, webbing of the toes, exostoses, abnormal shape of the long bones, shortening of the femoral neck, spina bifida, fusion of the cervical vertebrae, abnormal cervical vertebrae with small body, large spinous process, deformity of ears and deafness[1-3]. In our patient, great toes were found to be unusually short. Left side of the lower part of anterior chest wall was remarkably depressed from the birth. This type of congenital bony thoracic deformity is an unknown association of MOP. Acquired thoracic deformity as a part of heterotopic ossification would not be uncommon. In acute inflammatory stage such lesion may be mistaken as osteosarcoma as it is reported earlier[8]. We wanted to emphasize the association of congenital thoracic deformity with MOP and not the association of acquired thoracic deformity[8].

In general, there is no cure for MOP. Further abnormal ossification is prevented by avoiding soft tissue injury and muscle damage. Trauma, intra-muscular injections and surgery are usually discouraged for the same reason. Current guidelines have classified drugs into 3 groups. Class I drugs include corticosteroids and non-steroidal anti-inflammatory agents for symptomatic relief of acute episodes. Class II drugs include leukoteriene-inhibitors, mast-cell stabilizers and aminobisphosphonates. They have theoretical application to MOP and may be useful in selected cases. Thalidomide, VEGF trap and Noggin (pre-clinical) belongs to Class III drugs and at present they are experimental agents[1].

Conflict of Interest : None

Contribution : SKG, SD and SS contributed in evaluation and management of the patient. MG contributed in drafting the article.

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