

Myositis Ossificans Progressiva

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Myositis ossificans progressiva (MOP) is an extremely rare disease with an estimated incidence of about 0.1 per million(1). First described by Paten in 1692(2), there are about 500 cases reported in world literature and less than five in Indian literature(3,4). The characteristic clinical findings make the diagnosis simple on clinical examination and radiology alone(3,4).

Case Reports

Case 1: A two year eight month old female child presented with swellings over both scapulae and back of four months duration without a history of trauma. The swellings were not tender when noticed and there were no signs of inflammation. The swellings subsided over weeks leaving behind hard masses and marked restriction of movements of the underlying spine. There was no history of easy bruising in the past or

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a family history of such illness. A complete hematological workup had been done in another hospital for the same complaints.

On examination (*Fig. 1*) scalp hair was sparse and the first toes of both feet were very short with absent phalanges and nails. There were no other obvious congenital malformations. There were bony hard subcutaneous swellings overlying both scapulae and on either side of the dorsolumbar spine. There was severe restriction of all movements of the spine.

Radiographs showed broad and deformed first metatarsals with absent phalanges. Subperiosteal growth of bone was seen on the medial aspects of the proximal tibiae. Soft tissue ossification was visible over the medial aspects of the scapulae and

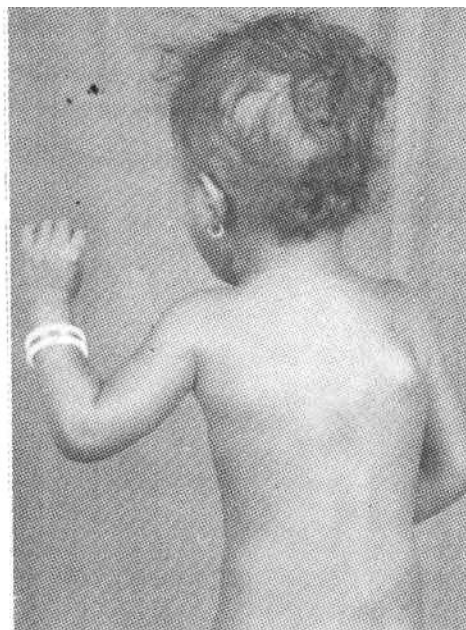


Fig. 1. Clinical photograph showing sparse scalp hair and subcutaneous masses on trunk.

paraspinally in the dorsolumbar region (Fig. 2). Routine hematological and biochemical investigations were normal. The audiogram was normal.

Case 2: A nine-year-old female patient came with a three year history of progressive impairment of movement of the entire spine, both shoulders and hips. The major complaint was an inability to open the mouth due to immobility of the temporomandibular (TM) joint, which had worsened after surgery of TM joint for the same complaints. On examination, she had laterally deviated short first toes of both feet. There was marked restriction of movements of the hip, spine and TM joints with multiple hard subcutaneous swellings especially on the trunk. Radiographs showed ossification of muscles and subcutaneous tissues.

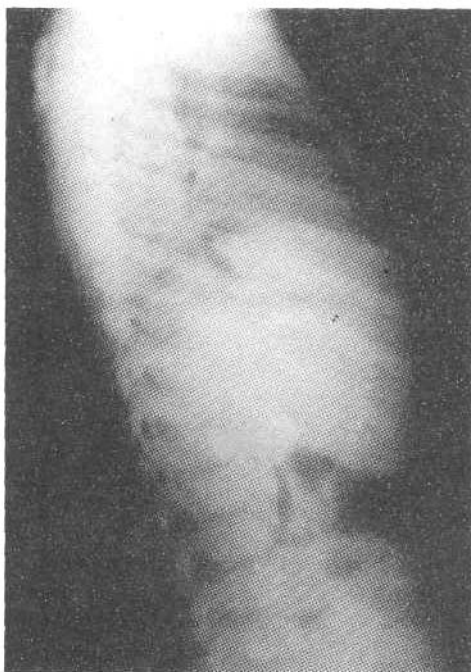


Fig. 2. Radiograph showing soft tissue ossification of the paraspinal region of dorsal spine.

Discussion

Spontaneous ossification associated with specific deformities of big toes is diagnostic of MOP(2). Although in myositis ossificans circumscripta and traumatica ossification is seen following none to mild or significant trauma, respectively(5), the lesions then observed are single, non-recurring on excision and do not involve other portions of the body. MOP, an autosomal dominant disorder is pathologically characterized by progressive replacement of muscles, tendons, ligaments, fascia and aponeurosis by bone leading to progressive stiffness of the adjacent joints and particularly the chest wall leading to pneumonia and death. The presence of type I collagen in these tissues probably determines its eventual ossification. Congenital abnormalities usually associated with short and/or stiff great toes include short thumbs, fifth finger clinodactyly, short broad femoral neck, exostoses of proximal tibiae and abnormal cervical vertebrae with small bodies, large pedicles and large spinous processes. Other associated problems include deafness in 25% of cases(2).

The mode of presentation in our patients was typical. The disorder usually manifests between birth and 10 years with a mean of three years. Our patients presented at three and nine years of age, respectively. Spine and shoulder stiffness develops by 10 years and restricted hip movements develop by 20 years. These patients are usually confined to bed by the age of 30 years. Both our patients had decreased mobility of the spine and shoulder on presentation while case 2 had already developed decreased mobility of the hips and TM joint. Sites of primary involvement include the neck (50%), dorsal paraspinal region (30%) (as seen in both our cases), head (10%), or limbs (10%).

Seventy per cent develop temporomandibular (TM) joint ankylosis (as in *Case 2*) which used to lead to death by starvation(2). Other common sites of ossification include joint capsules, ligaments and plantar fascia. However, connective tissue of facial and extraocular muscles, intestines, tongue, larynx and skin are not affected. Trauma is the most important exacerbating factor and biopsies should, therefore, be avoided(2). Episodes of ossification are, however, usually spontaneous. Operations in -uninvolved sites are uneventful. Surgical removal of the extra new bone is not helpful as reformation of the myositic bone occurs within a short time. Physiotherapy, change in type of work and genetic counselling should be advised. Numerous drugs have been tried for the primary disease but without success.

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Joubert Syndrome

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Joubert Syndrome is an autosomal recessive anomaly characterized by agenesis of the cerebellar vermis. The symptoms comprise of episodic panting tachypnea,

rhythmic protrusion of the tongue, and jerky eye movements in the neonatal period with subsequent ataxia, dysequilibrium, and psychomotor retardation. There are no reports of this condition in the Indian literature. We present a neonate with Joubert Syndrome to alert pediatricians to resort to cranial imaging in neonates with unexplained panting tachypnea.

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