

CASE REPORT

UNUSUAL PRESENTATIONS IN MYOSITIS OSSIFICANS PROGRESSIVA A CASE REPORT

S. A. KHAN, M. ZAHID, N. ASIF, N. GOGI

Myositis ossificans progressiva is a rare connective tissue disorder. We present here a case of myositis ossificans progressiva with some unusual presentations and associated congenital skeletal anomalies that are reported very infrequently in the literature. The case report highlights the importance of early diagnosis in a case of rapidly progressive myositis ossificans progressiva.

Keywords : myositis ossificans progressiva ; fibrodysplasia ossificans progressiva ; Munchmeyer's disease ; exostosis ; clinodactyly.

Mots-clés : myosite ossifiante progressive ; fibrodysplasie ossifiante progressive ; Maladie de Munchmeyer ; exostose ; clinodactylie.

INTRODUCTION

Myositis ossificans progressiva, otherwise called fibrodysplasia ossificans progressiva or Munchmeyer's disease, is a rare connective tissue disorder. The condition consists of widespread, progressive ectopic ossification of the striated muscles, ligaments, fascia and the subcutaneous tissues leading to crippling deformities and death. Widespread skeletal anomalies are associated with this disease.

We present here a case of a young boy with myositis ossificans progressiva, presenting with rapidly progressive ossified lesions along with characteristic as well as unusual congenital anomalies.

CASE REPORT

A 15-year-old boy presented in the outpatient clinic of our hospital with the complaints of recur-

rent, painless swellings around the neck since the age of one year along with restriction of all neck movements. According to the parents, similar swellings also appeared along the lower back, shoulders and elbow joints. The swellings gradually regressed in size followed by restriction of movements at these sites. No medical attention was sought for the condition till a couple of months back when similar swellings restricted the movements of the right knee joint as well. There was a history of impaired hearing for the last three years. There was no history of injury. The child was the youngest of three children, with no similar parental or sibling history. The child walked with a lot of difficulty and had an awkward supported gait. According to the parents the walking distance had gradually reduced and the child was able to carry out his daily routine only with assistance.

On examination, the patient was thin built with a short webbed neck and a low hairline (fig. 1). Examination of the spine revealed a balanced dorsolumbar scoliosis and no spinal movements were possible. Both shoulder joints were fixed in neutral position (fig. 1) and there was a fixed flexion deformity at both the hip and elbow joints. The right knee joint was fixed in 10° of flexion. The movements of the left knee, wrists, small hand joints and

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Fig. 1. — Clinical photograph of the child, showing the fixed deformities of both the elbows and shoulders.

the ankles were within normal limits. The child had a bilateral hallux valgus with short great toes. There was bilateral cleinodactyly of the fifth finger.

Examination of both the arms revealed painless, bony hard swellings around the arms. Similar bony swellings were also palpable around the upper end of tibiae and the lateral aspect of the thigh. Chest expansion was restricted (< 3cm.). Other systems were normal on clinical examination.

Investigations

Radiographic evaluation of the child revealed extensive bilateral ectopic ossification of the arm and thigh muscles along with bony bars extending from the arms to the chest on both sides (fig. 2). There was fusion of the posterior elements of the fifth and sixth cervical vertebrae along with ossification in the neck muscles and with a sternocostal

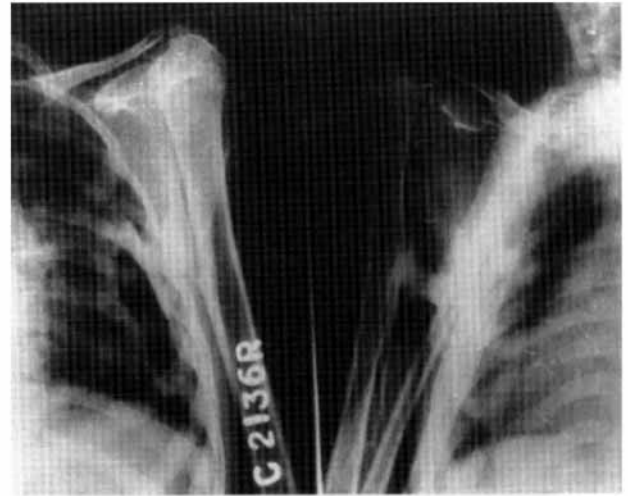


Fig. 2. — Xray (AP views) of both arms showing a bony bar extending from the shaft of the humerus to the lateral chest wall.



Fig. 3. — Xrays of both feet (AP views) showing a monophalangeal great toe with hallux valgus.

bony bar. Pelvic radiographs revealed bilateral shortening of the neck of the femur. The xrays of the lumbosacral spine showed a balanced scoliotic curve along with spina bifida at the first sacral vertebra. Radiographic evaluation of both the tibiae showed bilateral exostosis at the proximal end of each tibia. Xray of the feet showed monophalangeal great toes with hallux valgus (fig. 3). Xray of the hands revealed a short first metacarpal with cleinodactyly of the fifth finger (fig. 4). The routine



Fig. 4. — Xray of the right hand (AP view) showing the short first metacarpal with cleinodactyly of the fifth finger.

blood investigations were normal. There was a mild increase in serum alkaline phosphatase. Serum calcium and phosphorus were within normal limits. Serum uric acid was normal.

DISCUSSION

Myositis ossificans progressiva is an autosomal dominant connective tissue disorder, which is also known to occur sporadically. Guy Patin first described it in 1692. The appearance and disappearance of painless recurrent swellings around the neck, back, shoulders and jaw characterize the disease (2). Ossification usually starts in the upper paravertebral muscles and spreads from axial to appendicular, cranial to caudal and proximal to distal sites. The swellings are by and large painless with no features of acute inflammation, although red inflammatory ectopic masses of calcification are also reported (1). The presentation of the dis-

ease may vary from an asymptomatic phenotypic variant to disabling ectopic ossification causing crippling deformities and premature death. The ectopic ossification leads to various deformities including scoliosis, torticollis and limb deformities. The disease may have an insidious or rapid onset but the course is usually progressive, leading to premature death in the majority of the cases (2). Heterozygous individuals have slower progression and survival into adulthood is common (1).

The condition presents with widespread congenital anomalies including hallux valgus, monophalangic great toe, cleinodactyly, short first metacarpal and reduction defects of the limbs. Impaired hearing, which was seen in our case, is an extremely rare presentation (7). The rarity of impaired hearing in other reports could be due to the fact that most cases present at an early age, when diagnosing the hearing impairment may be difficult. The presence of tibial metaphyseal exostosis and spina bifida of the first sacral vertebra in our case is an extremely rare finding and we could not find any similar occurrence in the literature. The rarity of mental retardation is well known in these cases (2). Biochemical status of the patient is also very typical with normal serum calcium and phosphate and normal or elevated serum alkaline phosphatase (4).

The presence of characteristic congenital anomalies and a progressive ossification pattern clinically differentiates the disease from conditions like tumoral calcinosis, dermatomyositis, and Christian-Weber syndrome (relapsing nodular non-suppurative panniculitis).

The etiology and treatment of myositis ossificans progressiva has by and large been a difficult unsolved mystery (6). The currently presumed etiology of the condition is a causal mutation in the gene responsible for the synthesis of bone morphogenetic protein- 2A (BMP-2A), the gene is also responsible for limb patterning and ossification (5). Known exacerbating factors like trauma, surgery and massage are to be avoided. A number of drugs for the treatment of this condition have been tried. Previously used etidronate is now found to have deleterious effects on the metaphysis (3). Recently the use of isotretinoin (13-cis-retinoic acid) in a dose of 1-2 mg/kg per day has been found to be

useful in some patients (8). Gentle guarded physiotherapy is helpful in preventing joint deformities and thus improving the quality of life.

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SAMENVATTING

S. A. KHAN, M. ZAHID, N. ASIF, N. GOGI. Myositis ossificans progressiva met ongewone aspecten. Gevalstudie.

Myositis ossificans progressiva is een zeldzame bindweefsel-aandoening. Bij het voorliggend geval noteerde men ongewone bijzonderheden naast congenitale afwijkingen zoals er zelden vermeld worden in de literatuur. Dit geval onderstreept het belang van een vroegtijdige diagnose, zeker wanneer de evolutie van het snelle type is.

RÉSUMÉ

S. A. KHAN, M. ZAHID, N. ASIF, N. GOGI. Myosite ossifiante progressive avec des traits inhabituels : présentation d'un cas.

La myosite ossifiante progressive est une affection rare des tissus conjonctifs. Les auteurs présentent un cas de myosite ossifiante progressive qui présentait quelques traits inhabituels et des anomalies congénitales associées, très rarement rapportées dans la littérature. Ce cas illustre l'importance d'un diagnostic précoce, dans le cas d'une myosite ossifiante progressive rapidement évolutive.