ORIGINAL RESEARCH PAPER

Radiodiagnosis

FIBRODYSPLASIA OSSIFICANS PROGRESSIVA: A RARE ENTITY

KEY WORDS: Fibrodysplasia ossificans progressiva, Computed tomography, Heterotopic ossification

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ABSTRACT

Myositis ossificans progressiva is a rare connective tissue disease characterized by widespread, progressive, heterotopic ossification of soft tissues, congenital skeletal anomalies and progressive ectopic bone formation. We report 2 cases which presented with complains of difficulty in walking, stiffness and pain in both hip joints since 15 years. CT reveals extensive bridging extra skeletal areas of sclerosis and ossification involving the soft tissue around the hip and lower extremities. Though rare, diagnosis of fibrodysplasia ossificans progressiva should be considered whenever characteristic radiographic features of multifocal heterotopic bone formation are seen. Early diagnosis is necessary to prevent its debilitating progression.

INTRODUCTION

Fibrodysplasia Ossificans progressiva (FOP) is a disorder of the mesodermal tissue characterized by initial inflammation and subsequently proliferation of fibrous tissue and formation of ectopic bone tissue. The ectopic bone tissue formed is located in soft parts mainly in the connective tissue of striated musculature [1]. It is an autosomal dominant genetic disease with complete penetrance and variable expression [2]. Plain radiograph is the initial modality of choice for providing characteristic findings however CT is gold standard for detecting subtle areas of heterotopic ossification.

The disease might flare up sites of trauma or injury[2].

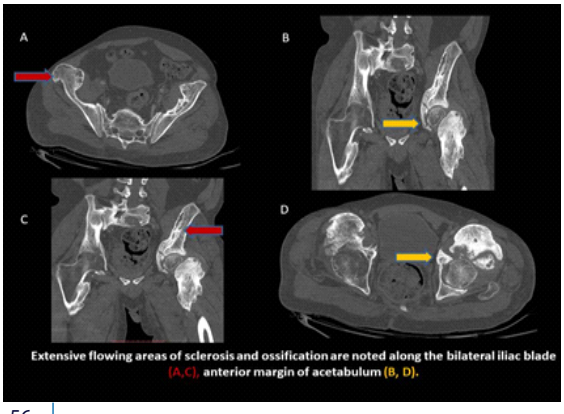
Case report:

Case 1.

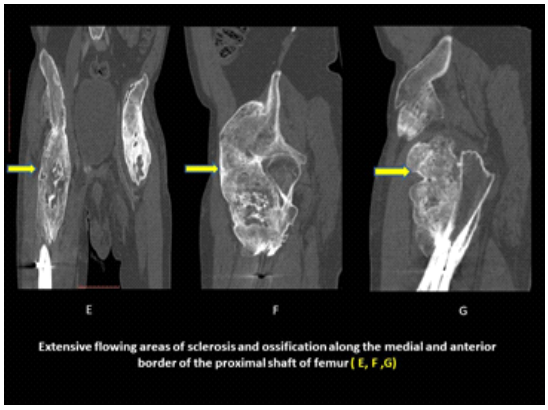
A 35 year old male presented with complains of difficulty in walking, stiffness and pain in both hip joints since 15 years.

Patient had history of repeated trivial trauma.
Coronal and axial reformatted CT images reveals extensive areas of sclerosis and heterotopic ossification involving bilateral iliac blade and acetabulum as shown in the images (Figure 1 and Figure 2)

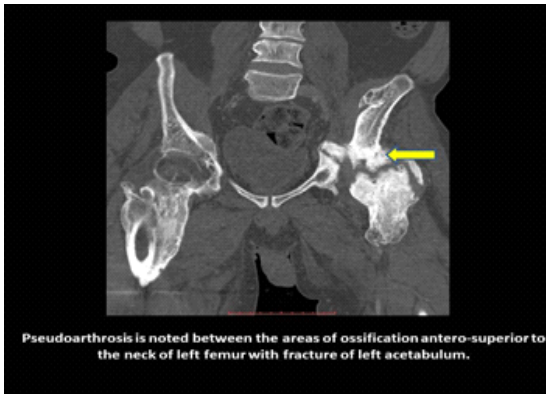
Coronal CT images reveals pseudoarthrosis between the areas of ossification antero-superior to the neck of left femur with fracture of left acetabulum (Figure 3)



Extensive flowing areas of sclerosis and ossification are noted along the bilateral iliac blade (A,C), anterior margin of acetabulum (B, D).



Extensive flowing areas of sclerosis and ossification along the medial and anterior border of the proximal shaft of femur (E, F ,G)


Pseudoarthrosis is noted between the areas of ossification antero-superior to the neck of left femur with fracture of left acetabulum.

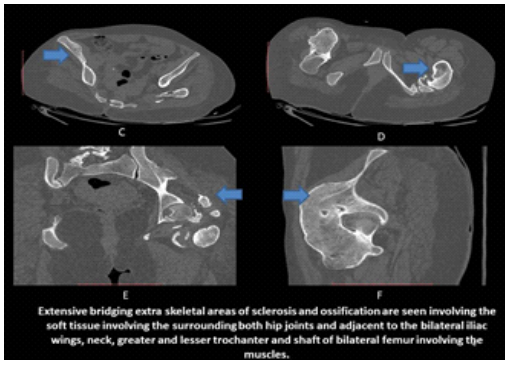
Case 2:

A 25 year old female presented with fixed coxa vara deformity and difficulty in mobility since birth.

X-ray and CT reveals areas of extra-skeletal sclerosis and ossification involving the soft tissue surrounding greater and lesser trochanter and proximal shaft of femur.

Areas of heterotopic ossification are also seen involving the bilateral iliac blades and acetabulum (Figure 4 and Figure 5)

Extensive bridging extra skeletal areas of sclerosis and ossification are seen involving the soft tissue surrounding greater and lesser trochanter and shaft of right femur (A& B).



Extensive bridging extra skeletal areas of sclerosis and ossification are seen involving the soft tissue involving the surrounding both hip joints and adjacent to the bilateral iliac wings, neck, greater and lesser trochanter and shaft of bilateral femur involving the muscles.

DISCUSSION:

Myositis ossificans progressiva is a rare connective tissue disease characterized progressive, heterotopic ossification of soft tissues, which is usually complicated by restriction of movements at the affected sites [1].

It is estimated that the incidence of the disease is 1 per 2 million people and the prevalence is 2500 cases worldwide [3].

The etiology of ectopic bone formation is unknown however overproduction of bone morphogenetic protein-4 in lesion cells and lymphocytic cells are reported as one of the contributing factors [7].

It is clinically characterized by two main features, anomalies of the great toes and thumbs and progressive ectopic ossification of soft tissues with average onset age of the about 5 years [7], however patient presented late in our case.

Restricted movements, stiffness and walking difficulty are well demonstrated in some cases. There is also evidence of short (macrodactylia with hypoplasia or synostosis of the phalanges) and deformed great toes (Valgus deformity) noted [7].

The disease usually starts in the paravertebral musculature and progresses into scapula, proximal portions of arms, jaw, pelvic area and skull [2].

It progresses in a cranio-caudal dorsal-ventral and proximo-distal manner [3].

It causes bridging between extremities and torso, ribs and between thorax and pelvis with severe restriction of motion [7]. As in case of our patient, there is bridging seen in pelvis and extremities.

Association of conductive hearing loss due to fusion of the ear ossicles is reported in some cases [2].However it is not appreciated in our study.

Affected patients usually become dependent and confined to wheel-chair or bed at the second decade of their life as a result of ankylosing of all major joints of both axial and appendicular skeleton [2], this is appreciated in our case.

Though it is a clinical diagnosis, presence of congenital anomalies of the great toes, progressive heterotopic ossification seen in X-ray and CT and classical pattern of disease progression help in formulating a definitive diagnosis [6].

Biopsies are not recommended for the diagnosis because they might worsen the ossification at the site [6].

X-ray is the initial modality of choice to look for areas of extraskeleton ossification.

However Computed Tomography (CT) provides valuable information about the extent of the disease in the preosseous stages, which may manifest as swelling and edema of the muscular fascia planes and muscular bundles [8]. In our case extraosseous muscle and soft tissue involvement is well appreciated on CT.

There is no definitive treatment for FOP[2, 5]. However prevention of trauma or injuries by all means, helps prevent flare-ups.

Intramuscular vaccines should not be given to the patients as this may exacerbate the progression [2]. Many medications have been implemented however no significant evidence of their effectiveness is noted[2]. Corticosteroids, non-steroidal anti-inflammatory drugs, leukotrienes and Cox-2 inhibitors have been tried[2, 5]. Some studies suggested the use of high doses of bisphosphonate (Etidronate) which showed good results due to its potential effect on inhibiting mineralization of the newly formed cartilage [2].

Differential diagnosis:

1 Parosteal osteosarcoma
2 Soft tissue sarcomas like
- Malignant fibrous histiocytoma.
- Synovial sarcoma.

CONCLUSION:

Myositis ossificans progressiva is a rare congenital disease of progressive ectopic ossification of soft tissues. Physicians should be able to diagnose this disease in its early stages in order to prevent its disabling progression. It usually presents in a classical pattern and it has characteristic radiological findings in plain films and CT scan. Although drugs can be used to decrease some symptoms, the best approach is still the early diagnosis and prevention of trauma that can provide a better quality of life.

Learning points:

1. Myositis ossificans progressiva though a rare congenital disease, however early diagnosis is necessary to prevent its debilitating progression.
2. Computed tomography plays a critical role in its diagnosis and follow up detecting trivial fractures.
3. Best treatment is prevention of trauma with judicious use of bisphosphonates and corticosteroids in acute flare-ups.

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