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Fibrodysplasia Ossificans Progressiva A Case Report

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Abstract:- Fibrodysplasia ossificans progressive (FOP) or Munchmeyer's disease is an autosomal dominant disease affecting the musculoskeletal system. During this rare condition, an inflammatory process is responsible for progressive soft tissue calcification. We report the case of a 15-year-old patient with joint deformities, subcutaneous dorsal and hard lumbosacral nodules and rigidity of the right shoulder and elbow. Clinical examination revealed dysmorphic facial features. Magnetic resonance showed heterotopic ossification. The patient was treated with monthly pamidronate cures and short-term corticosteroid therapy during painful flare-ups.

Keywords:- *Fibrodysplasia ossificans progressiva, Corticosteroid, Bisphosphonates.*

I. INTRODUCTION

Described for the first time by Patin in 1692, then by Munchmeyer in 1869, Fibrodysplasia ossificans progressiva (FOP) is a genetic disease, with a prevalence of 1 in 2,000,000. It is characterized by heterotopic ossification of tendons, facias, fasciae and muscles, becoming more noticeable in the second decade of life. FOP is associated with a high mortality rate caused by respiratory complications.

After an initial trigger, an inflammatory edema will develop in soft tissues, preferentially in the lumbar and neck area, followed by more rapid ossification in the injured area, causing rigidity and/or painful immobilization of the structures concerned. The treatment is essentially symptomatic. Several therapeutic protocols have been used combining analgesics, corticosteroid, acetic acid, and bisphosphonates, but with unsatisfactory responses.

II. CASE REPORT:

A 15 years oldpatient,non-consanguineous,without any pathological history, presented with a progressive functional impotence over 6 years, affecting the upper limbs associated with a thoracic kyphosis. He also reported intermittent asymmetrical bilateral skeletal deformities in the trunk and upper limbs of appearance with no apparent triggers. These deformities progressively increased in size, and were associated with painful attacks which were slightly improved by analgesics.

The clinical examination showed a forward head posture dorsal kyphosis with a significant cervical and lumbar spinal stiffness, and non-inflammatory fixed painful swellings of the paravertebral dorsal and muscles, against the right pectoral muscle (Figure1). In addition, the patient presented with right parasternal hard subcutaneous nodules. These swellings were hard, painless, fixed and non-inflammatory, compatible with confluent diffuse bony outgrowths with a confluent appearance. Examination of the feet revealed bilateral hallux valgus (Figure 2) . Both shoulders were limited. The skeletal examination did not reveal any other abnormalities.

Soft tissue ultrasonography showed a deep heterogeneous non-vascular mass of the scapular region. Cervical and thoracic MRI revealed three fused subcutaneous soft tissue masses with ill-defined margins in the lumbar, right scapulothoracic and left paravertebral region measuring 75x27mm, 120x23mm and 18x15mm respectively, with multiple calcifications and muscular aponeurosisenhancement after contrast material injection. At the cervical level, the imagery showed a swollen and fused aspect of the posterior arches of the vertebrae with loss of cervical physiological lordosis. The electromyogram was normal. The phosphocalcicassessment (calcium, phosphorus and parathormone) was normal. Muscle biopsy showed a non-specific muscular dystrophy.

The patient was initially treated with ibuprofen without any improvement. Faced with this flare-up, treatment with bisphosphonates (Pamidronate 60 mg /day for 03 days per month for 03 months) was instituted, combined with oral corticosteroid therapy (1mg/kg/day of prednisone) for 2 to 5 days during the progressive painful attacks, with a favorable evolution with regression and stabilization of bone lesions and pain, thus improving our patient's quality of life.

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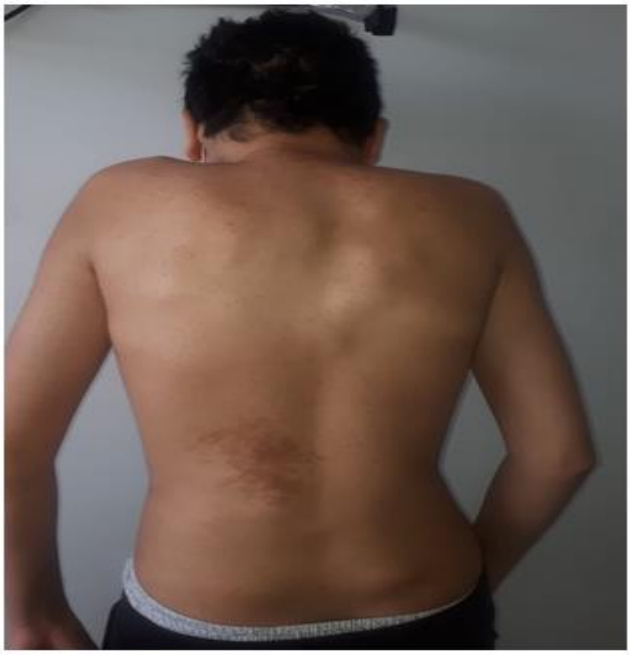

Figure1 : bony swellings over back

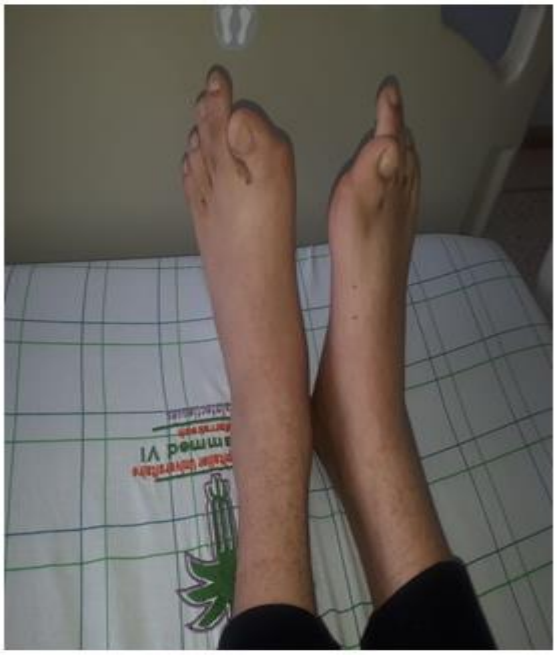

Figure 2 : Bilateral hallux valgus

III. DISCUSSION

FOP is an autosomal dominant genetic disease. It is usually revealed in the second decade of life by deformities and subcutaneous nodules of the posterior cervical and lumbar region. Nevertheless, late forms in adulthood have been reported. [1-2]

The association of facial dysmorphia with hallux valgus and diffuse soft tissue calcifications from the second decade of life should suggest progressive ossifying myositis. Therefore, an early diagnosis based on clinical and radiological arguments can help avoid surgical biopsies which arenot recommended in FOP patients. [3-4-5]

The main differential diagnoses of FOP includedermatomyositis, idiopathic calcinosis, bone tumors, sarcomas, Albright's hereditary osteodystrophy, and progressive osseous heteroplasia. [6-7]

The course is unpredictable, and FOP may progress to regression, stabilization or progression, with alternating periods of worsening and of remission occurring spontaneously or under treatment. These flare-ups of ossification may occur in cases of muscle trauma, surgery, muscle injections or dental care that require local anaesthesia. However, flare-ups can be spontaneous with no triggering factor. Classically the progression of FPO is very slow, but very rapid forms have been described.

During flare-ups, muscular and tendon ossifications lead to a functional handicap, with a progressive loss of autonomy. Thoracic impairment hinders the respiratory amplitude responsible for a restrictive respiratory syndrome. [8]

To date, no treatment has proven effective in FOP. The use of corticosteroids is usually based on its powerful anti-inflammatory effect. Prednisone at 2 mg / kg / day is administered (maximum 100 mg) orally for 3 to 4 days to prevent relapses. NSAIDs can be used as a relay to corticosteroid therapy and often help maintaining the initial remission. [9]

The use of bisphosphonates with their anti-angiogenic and anti-inflammatory effects have been associated in isolated cases with an improvement of the inflammatory process. However, they do not prevent the occurrence of subsequent outbreaks [10-11]. Physical Therapy and chest physiotherapy are needed to reduce the functional and thoracic repercussions of this disease. [12]

IV. CONCLUSION

FOP is a very rare entity. This diagnosis should be suspected in adolescents with osteo-muscular excrescences associated with a hallux valgus. More studies are needed to further develop promising therapies able to modify the evolution of the disease.

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