FIBRODYSPLASIA OSSIFICANS PROGRESSIVA OR MUNCHMEYER'S DISEASE : A CASE REPORT

ABSTRACT :

Fibrodysplasia ossificans progressive (FOP) is a rare variety of myositis ossificans characterized by congenital malformations of the great toes and progressive heterotopic ossification that can induce a disabling second skeleton.
This heterotopic ossification can occur at any location, but typically affects regions in close proximity to the axial skeleton in the early/mild stages, before progressing to the appendicular skeleton. This can lead to restriction of movement as a result of ossification impacting joint mobility. Problems with swallowing and speaking can occur with ossification affecting the jaw, head, and neck.
The number of authentic cases of this disease in the literature is small.
We report the case of a patient with late-onset FOP and discuss the clinical, radiological and , evolutionary aspects of this disease.

Keywords : Munchmeyer's disease, myositis ossificans, heterotopic ossification, hallux valgus

INTRODUCTION :

Fibrodysplasia ossificans progressiva (FOP) or Münchmeyer's disease is a rare genetic disorder described by Patin in 1692, then by Munchmeyer in 1869. It is often sporadic and rarely familial, with autosomal dominant transmission, due to a mutation in the ACVR1 (Activin Receptor IA) gene, which codes for a morphogenetic protein, characterised by episodes of inflammatory flare-ups leading to the development of heterotopic ossifications in connective and muscular tissues.
These flare-ups occur at varying ages, are progressive and cumulative, leading to generalised ankylosis, respiratory and thromboembolic complications and early death in adulthood.

CASE REPORT :

The patient was 37 years old and had no previous pathological history. She presented with ankylosis of both upper and lower limbs causing disabling functional impotence since the age of 14 following trauma to the paravertebral muscles. The evolution was marked by the appearance of other ossified nodosities affecting various muscles, the last of which were the masticatory muscles, resulting in limited opening of the mouth and a deterioration in general condition due to eating disorders.

The clinical examination revealed stiffness of the entire spine, the heterotopic ossification on the back **(Figure 1)**, tendon retractions in all four limbs with a semi-flexed posture and limited mouth opening. She also had a genu valgum, a hallux valgus deformity of the big toes, which were short and roughly triangular in shape **(Figure 2).**

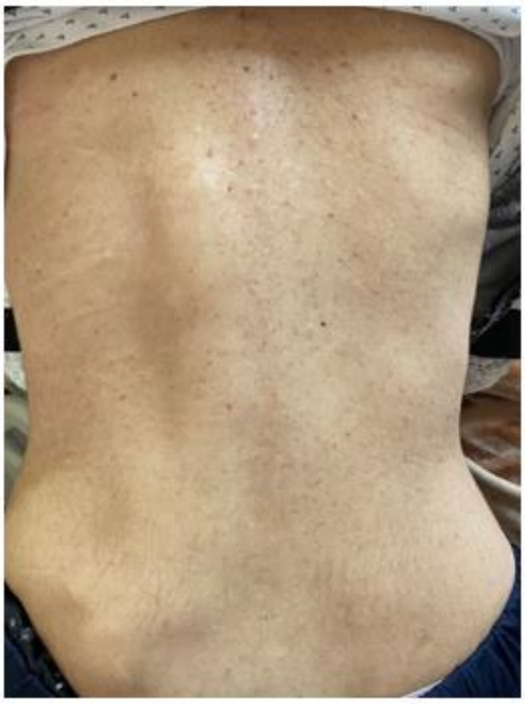


Figure 1 : Soft-tissue indurations representing early heterotopic ossification on the back of a patient with FOP

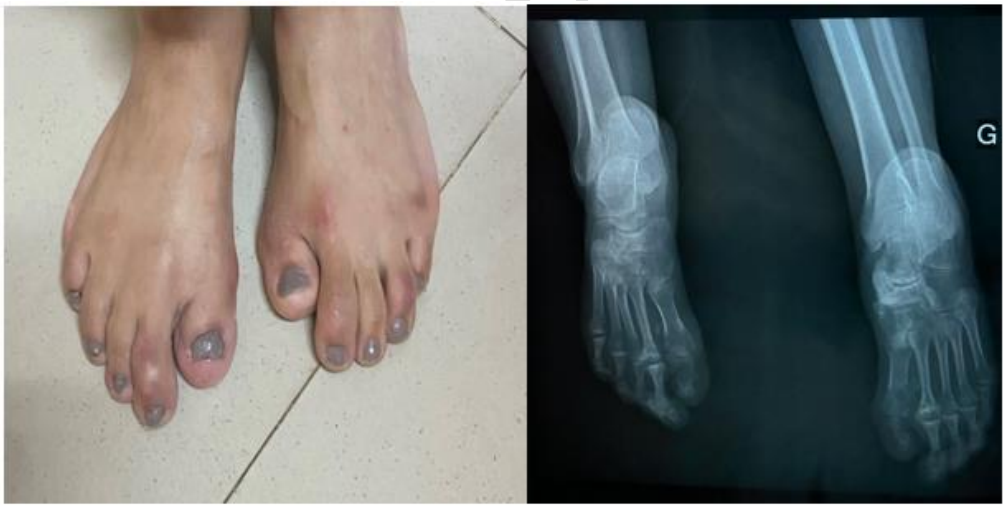


Figure 2 : Microdactyly of the great toe bilaterally and a radiograph of the foot showing microdactyly of the hallux.

Inflammatory and phosphocalcic tests were strictly normal. Bone imaging revealed heterotopic ossifications of the soft tissues in the pericervical, pre-scapular, pericostal, right peri-humeral, periarticular especially the right hip and right perifemoral areas **(figure 3) (Figure 4).**

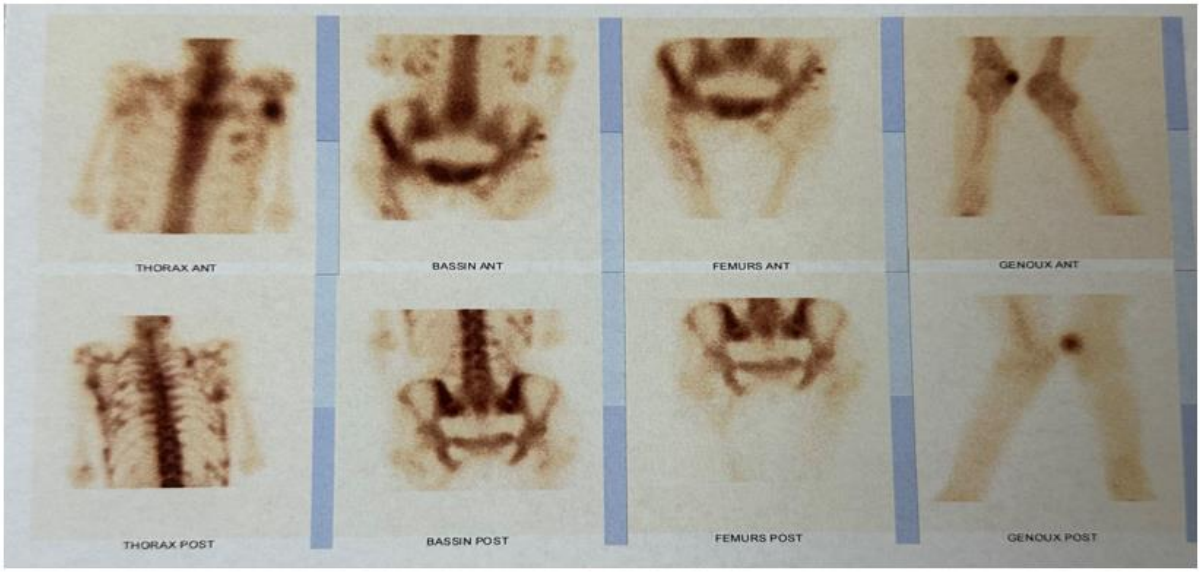
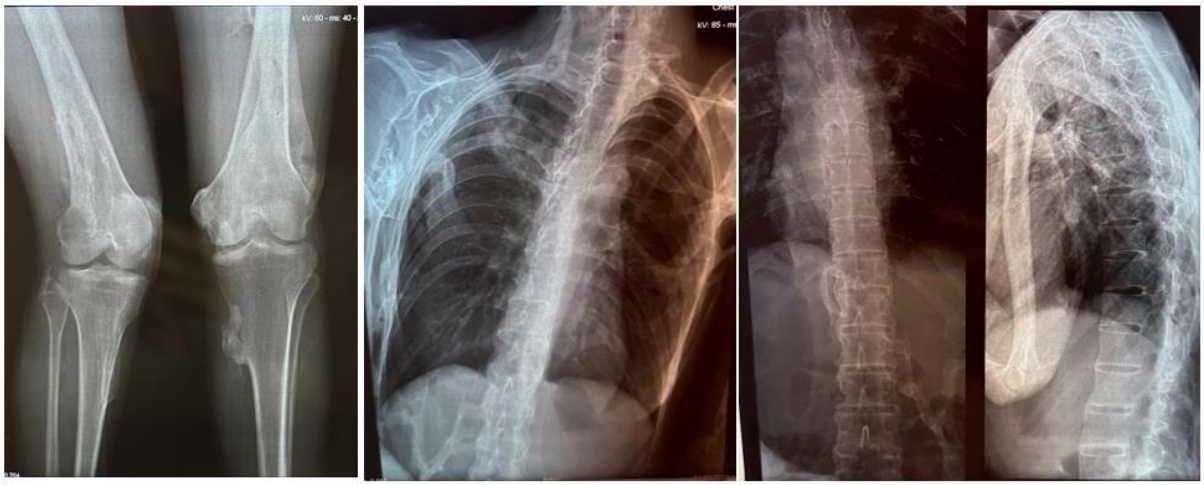


Figure 3 : Bone scintigraphy showed several foci of hyperfixation



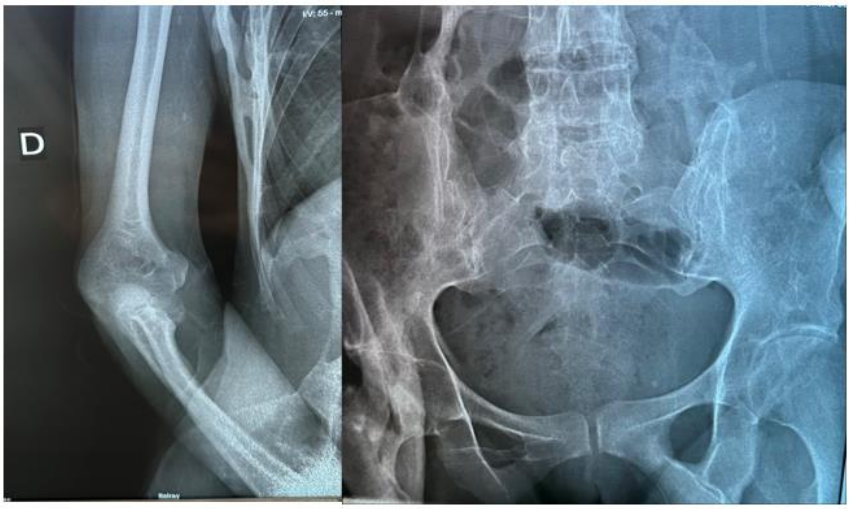


Figure 4 : Radiographs show bizarre and widespread heterotopic ossification of the soft tissues

The appearance of these ossifications and their association with hallux valgus led to the diagnosis of progressive myositis ossificans. No biopsy or surgery to free these heterotopic ossifications was indicated, as this would encourage their spread.

DISCUSSION:

Fibrodysplasia ossificans progressiva (FOP) is a rare autosomal-dominant disorder characterized by progressive extra bone formation in soft tissues (heterotopic ossification), such as skeletal muscle, tendons and ligaments.

The incidence of FOP is estimated at 1 in a population of 2 million, regardless of race, geographic area, age of person with the disease, or gender.

The gene responsible for FOP, ACVR1, was identified in 2006 **[1].** However, the molecular mechanisms underlying heterotopic ossification remained unclear, and there are currently no approved drugs that ameliorate heterotopic ossification in patients

The etiology of Munchmeyer's disease remains obscure. Trauma may be a precipitating factor, as was reported in our case. However, in view of the fact that 90% of the cases is associated with congenital anomalies, most frequent being microdactyly of the great toe, it has been suggested by many authors that the disease is either congenital or inherited.

FOP diagnosis is clinical, and it is usually made on the basis of the presence of three major criteria **[2, 3]:** congenital malformation of the great toes, progressive heterotopic endochondral ossification, and progression of the disease in well-defined anatomical and temporal patterns

Progression is usually craniocaudal, proximodistal, axoappendicular, with knee and ankle involvement
are often delayed **[4].**

Conventional radiology is the key to diagnosis, showing suggestive images such as ectopic corticalised calcification of the affected muscles and, at an advanced stage, bone bridges between different parts of the skeleton with a true ectopic skeleton. It can also show typical congenital bone malformations.

The discovery of AVRC1 genetic mutations confirms the diagnosis.

A biopsy is contraindicated due to risk of catastrophic explosive new bone formation**[5].**

No specific preventive or curative treatment has yet been validated.

Most patients with POF die of cardio respiratory failure linked to the chronic reduction thoracic expansion **[6].** Although the average age of death due to cardio respiratory failure is 40 years, the overall average life expectancy of patients is 57 years **[6].**

CONCLUSION:

Fibrodysplasia ossificans progressiva (FOP) is a rare disease with less than a thousand confirmed cases. It is a severely disabling genetic condition that affects soft tissues and is characterized by progressive extraskeletal heterotopic ossification and great toe deformities.

A malformation of the big toes (hallux valgus) is present from birth: it is typical and can be a warning sign.

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CONSENT :

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL :

Ethical approval was exempted by the Ethical Committee at Ibn Roch university hospital for reporting this case.