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Munchmeyer's Disease - Fibrodysplasia Ossificans Progressiva: A Rare Case Report

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Abstract **Case Report**

Munchmeyer's disease or Fibrodysplasia ossificans progressiva (FOP) is a rare, disabling connective tissue disorder involving malformation of the big toes and progressive heterotopic ossifications, which evolves in flare-ups and leads to disabling joint stiffness. We report the case of a 20-month-old infant who presented with multiple painless swellings of spontaneous and progressive onset on the back, elbow and lower limbs. These swellings were associated with bilateral hallux valgus. The radiological and CT scan findings were highly suggestive, and the diagnosis was confirmed by genetic study, which identified a mutation in the ACVR1 gene, confirming Fibrodysplasia ossificans progressiva syndrome. Epidemiological, etiopathogenic, diagnostic, evolutionary and therapeutic aspects are discussed through a review of the literature.

Keywords: Munchmeyer's disease, heterotopic ossification, progressiva myositis ossificans.

INTRODUCTION

Fibrodysplasia ossificans progressiva (FOP), also known as MUNCHMEYER disease, is an extremely rare genetic disorder with variable expression, which occurs during the period of growth and insidiously but inevitably brings about immobilization. This condition is characterized by the ossification of muscles, tendons, fasciae and aponeuroses which evolves in flare-ups and leads to disabling joint stiffness. The disease usually begins during the first two decades of life and is gradually progressive, resulting ultimately in the complete immobilization of nearly every joint of the body. Treatment of this disorder has so far proved unsuccessful. The prognosis of the disease is poor.3 The rare incidence, obscure aetiology, unknown definitive treatment plan and poor prognosis make this condition to be presented and help to disclose the mystery behind this disease [1].

CASE REPORT

Y.Z, 20-month-old infant born of non-consanguineous parents, and from a pregnancy carried to term, with good psychomotor development, consulted for hard, painless skin nodules on the back (FIg 1), elbows and two lower limbs. The clinical examination revealed a very limited standing position, irreducible flessum of the right knee (Fig 3), irreducible flessum of the right elbow, multiple indurations on the left leg and opposite the 2 shoulder blades, retraction of the shoulder girdle muscles, and bilateral hallux valgus (Fig 2).

The phosphocalcic and haemograms were normal, and standard X-rays of the upper and lower limbs, (Fig 4 & 5) supplemented by ultrasound of the clinically palpated swellings, revealed bone excrescences on the right costal, left femoral and bilateral scapular bones, strongly suggestive of exostatic disease, complemented by thoraco-abdominal CT scans of both lower limbs, showing hypodense subcutaneous nodules in the right costal gril, with calcifications of the subscapular muscles, axilla and paravertebral muscles, with frank bone exostoses of the greater trochanters and bilateral femorals (Fig 6), suggesting fibrodysplasia ossificans progressiva.

The radiological tests were supported by a bone sintigraphy with tc 99, which showed intense hyperfixation in the affected areas mentioned above. Diagnostic confirmation was obtained by genetic study, which showed the presence of a heterozygous c.617 G > A mutation in exon 6 of the ACVR1 gene.

The child was put on analgesic symptomatic treatment with motor physiotherapy, with regular follow-up in paediatric rheumatology and orthopaedics.

Fig 1: Subcutaneous and bony indurations in the thoracic cavity

Fig 2: Characteristic deformity of the big toe: hallus valgus

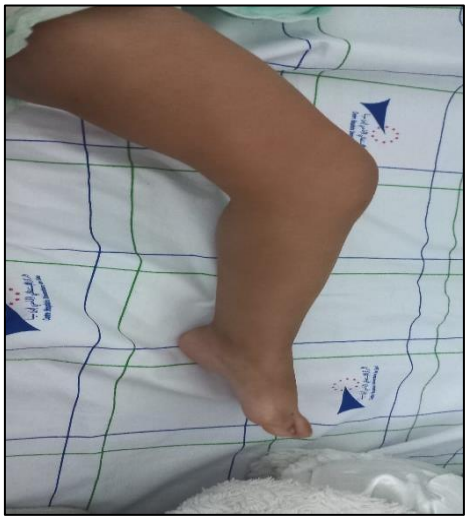

Fig 3: Irreducible flessum of the right knee

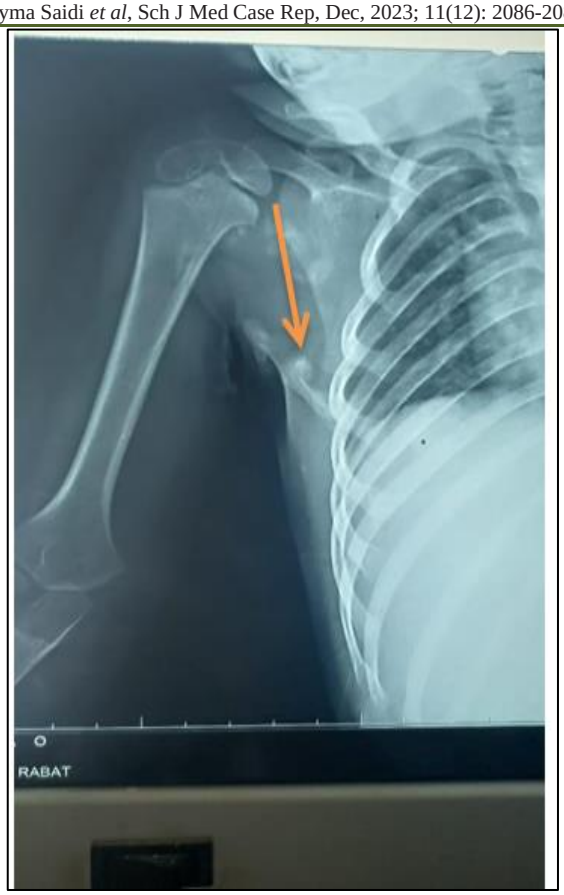

Fig 4: Ossifications opposite muscles Paravertebral

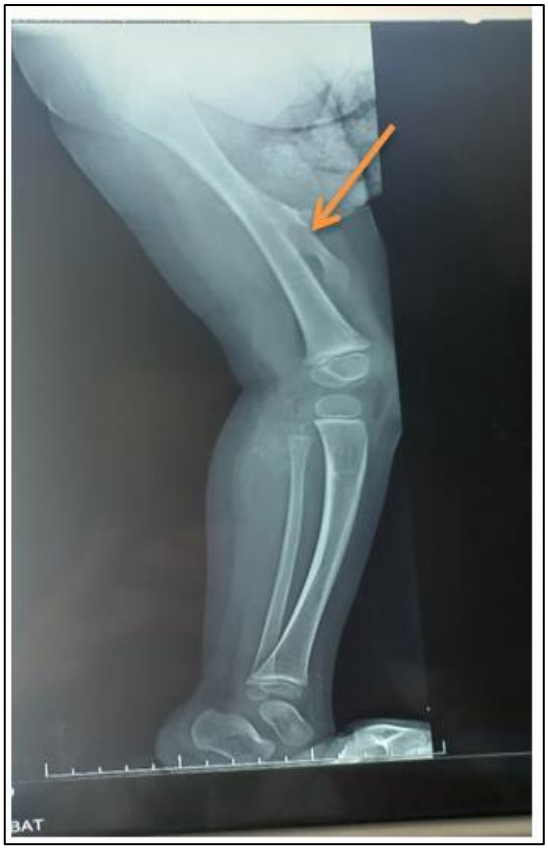

Fig 5: Right femoral bone excrescence

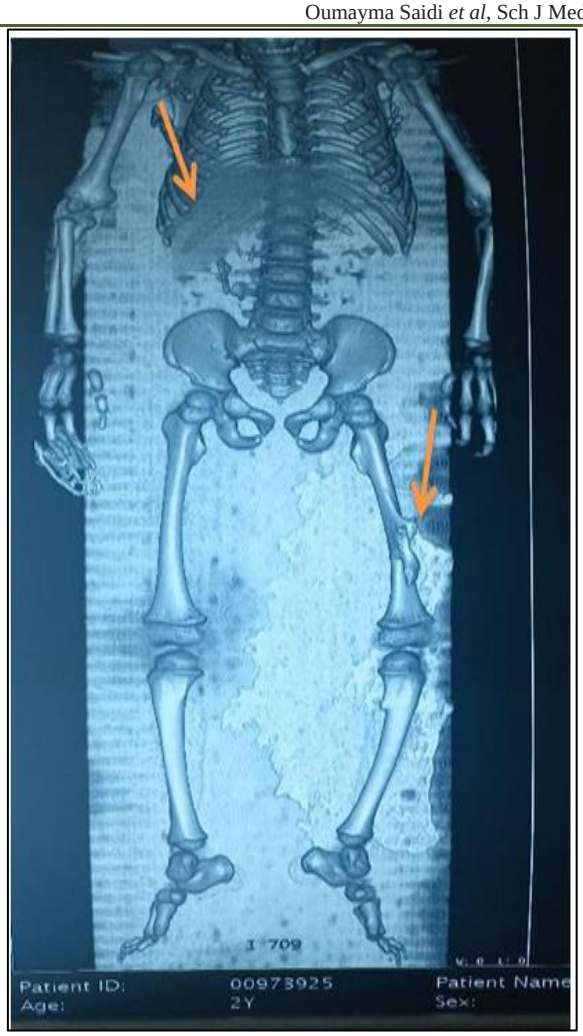

Fig 6: Three-dimensional CT scan showing bone excrescences

DISCUSSION

Fibrodysplasia ossificans progressiva, is an extremely rare genetic disorder, affects an average of 1 in 2 million people [2]. Described mainly in young children, it affects both sexes equally.

Both sexes: 41% of FOPs are detected before the age of 2, 80% before the age of 10, and 95% before the age of 15 [3]. It is caused by a spontaneous mutation of the ACVR1 gene on chromosome 4, which is autosomal dominant and codes for a receptor for BMP4 proteins involved in bone growth and shaping [4].

The symptomatology of FOP combines a fever with the appearance of inflammatory swellings affecting connective tissues and muscles. These will ossify and progressively spread to all regions of the body. This process initially affects the neck and paravertebral muscles, with extension following a proximo-distal and cranio-caudal pattern [5]. Muscles that do not fit are spared. This explains why the ocular muscles, diaphragm, tongue, pharynx, larynx and smooth muscles are respected [5].

Conventional radiology is the key to diagnosis, showing suggestive images such as ectopic cortical calcification of affected muscles at an advanced stage.

CT scans provide a better analysis of ossifications and their extent. MRI and scintigraphy can show lesions that are initially not yet ossified.

To date, no treatment has proven effective. However, several therapeutic approaches are available: Gentle physiotherapy is proposed for pain reliefduring flare-ups. Medication includes corticoids, mast cell inhibitors, cyclooxygenase 2 inhibitors,non-steroidal anti-inflammatories and amino-biphosphonates.

Surgical removal of these ossifications constitutes a new trauma that favours the development of additional heterotopic ossifications [2].

CONCLUSION

The absence of curative treatment to date means that the functional prognosis for carriers of this rare and disabling condition remains bleak. Until a genetic cure is

found, management remains symptomatic and preventive.

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