Fibrodysplasia Ossificans Progressiva

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A 2-year-old boy, born in Guinea Bissau, was referred to a pediatric hospital for evaluation and clinical follow-up of sporadic episodes of generalized pain and localized swelling. He was diagnosed with fibrodysplasia ossificans progressiva (FOP) at the age of 3, with a genetic test confirmation.

During the subsequent 10 years, he had multiple sporadic episodes of painful soft tissue swelling (elbows, hips, and dorsal spine) resulting in progressive stiffness and decreased mobility of the affected joints.

Two series of images document the disease progression over the years (Figs. 1 and 2). Radio-opaque ill-defined neoformations are present in the upper left limb and both hips. Their progression over time conditioned the rigid ankylosis of the affected joints.

Fibrodysplasia ossificans progressiva (FOP) affects 1:2,000,000 newborns. Classic FOP is caused by a recurrent activating mutation in the gene *ACVR1/ALK2*. It has an autosomal dominant transmission pattern, but in most cases occurs spontaneously.[1]

Patients with classical FOP are initially asymptomatic, and congenital malformations of the halluces are a characteristic sign.[1] In the first decade of life, it manifests as acute sporadic episodes of painful soft tissue swelling, some of which develop heterotopic ossification.[2]

Over time, extra articular heterotopic formations condition the child mobility. Its cumulative effects, including costovertebral ankyloses, intercostal musculoaponeurotic ossification, progressive kyphoscoliosis, ultimately leads to the development of thoracic insufficiency syndrome, markedly reducing life expectancy. At the present time, there are no established preventions or treatments for FOP. Fibrodysplasia ossificans progressiva is accelerated by trauma, so these patients must always be managed gently and any invasive procedure, such as surgery, is contraindicated. Physiotherapy, hydrotherapy, and kinesiotherapy may be helpful.[3] Median age of survival is approximately 40 years.

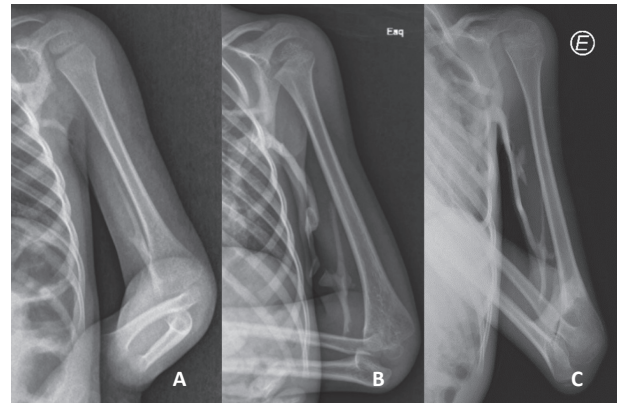

Figure 2. Left upper limb radiographs at 3 (A), 4 (B), and 12 (C) years of age.

Keywords: Child; Myositis Ossificans/complications; Myositis Ossificans/diagnostic imaging; Myositis Ossificans/genetics

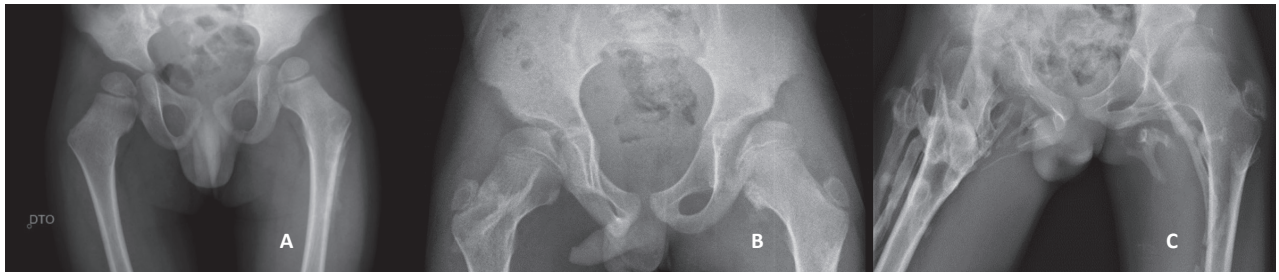

Figure 1. Anteroposterior pelvis view radiographs at 3 (A), 9 (B), and 12 (C) years of age.

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WHAT THIS REPORT ADDS

- Fibrodysplasia ossificans progressiva is a very rare and disabling genetic condition and is the most catastrophic disorder of heterotopic ossification in humans.
- Death often results from complications of thoracic insufficiency syndrome, around the age of 40. At the present time, there are no established preventions or treatments.
- Minor trauma can trigger painful new flare-ups of fibrodysplasia ossificans progressiva leading to progressive heterotopic ossification.
- Fibrodysplasia ossificans progressiva is commonly misdiagnosed. Children often undergo unnecessary and harmful diagnostic biopsies that exacerbate the progression of the condition.
- Definitive genetic testing of fibrodysplasia ossificans progressiva is now available and can confirm its diagnosis prior to the appearance of heterotopic ossification.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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