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Fibrodysplasia Ossificans Progressiva in A 13-Year-Old Girl: A Case Report

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Abstract **Case Report**

Fibrodysplasia ossificans progressiva (FOP) is a rare and disabling genetic disorder characterized by progressive heterotopic ossification of soft tissues and congenital skeletal malformations, particularly of the great toes. We report the case of a 13-year-old girl with no significant past medical history, presenting with progressive, painless stiffness of the spine and limbs, more pronounced at the elbows. The condition evolved through inflammatory flare-ups in an atraumatic and afebrile context, with preserved general condition. Clinical examination revealed cervical spine straightening, bilateral elbow tendon retractions, and bilateral hallux valgus deformity. Radiographic evaluation demonstrated soft tissue calcifications in the right thigh, bilateral monophalangic great toes with hallux valgus, bilateral periarticular heterotopic ossifications at the distal humerus, and extensive spinal heterotopic ossifications with posterior arch fusion from C3 to C6 on CT scan. Early recognition of the characteristic clinical and radiologic features of FOP is crucial to avoid unnecessary invasive procedures that may exacerbate disease progression.

Keywords: Fibrodysplasia ossificans progressiva, Heterotopic ossification, ACVR1 gene, Hallux valgus, Monophalangism, Case report.

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INTRODUCTION

Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare autosomal dominant disorder caused by mutations in the ACVR1 gene, leading to abnormal bone formation in muscles, tendons, fascia, and ligaments [1]. The disease typically begins in childhood and progresses through episodic inflammatory flare-ups, resulting in cumulative heterotopic ossification and progressive disability [2].

Congenital malformations of the great toes, especially monophalangism and hallux valgus, are considered hallmark features and are often present at birth [3]. Imaging plays a central role in confirming the diagnosis and assessing the extent of skeletal involvement.

CASE PRESENTATION

A 13-year-old girl, with no prior medical history, presented with progressive, painless stiffness of the spine and upper limbs, more marked at the elbows. The symptoms evolved through recurrent inflammatory flare-ups in an atraumatic, afebrile setting, with preserved general health.

Clinical examination revealed straightening of the cervical spine, along with bilateral elbow tendon contractures and a bilateral hallux valgus deformity.

Plain radiographs of both knees revealed soft tissue calcifications in the right thigh, suggestive of heterotopic ossification (figure1).

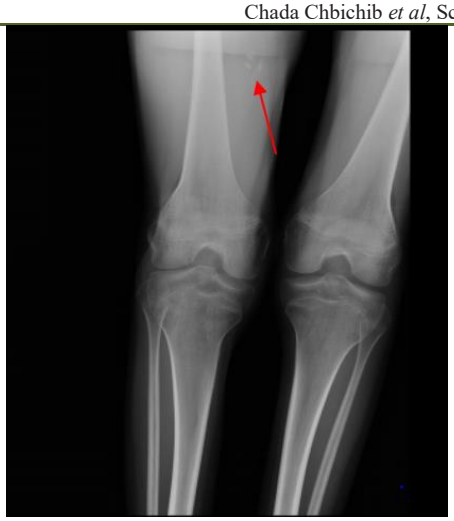

Figure 1. Bilateral knee radiographs

Anteroposterior view showing irregular calcifications within the soft tissues of the right thigh (arrows), consistent with heterotopic ossification without underlying bone destruction.

Radiographs of both feet demonstrated bilateral hallux valgus deformity associated with monophalangic great toes (figure 2).

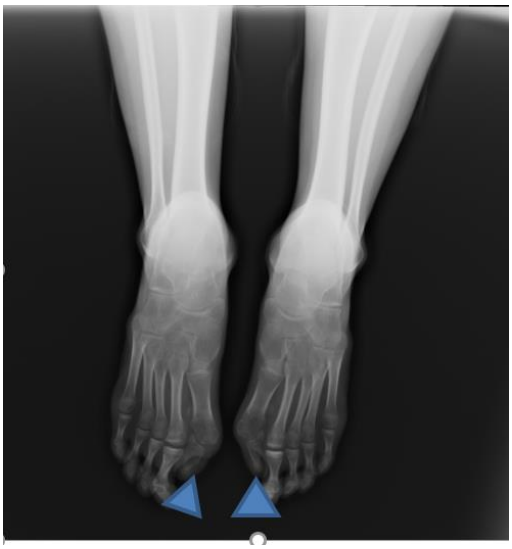

Figure 2. Bilateral foot radiographs

Anteroposterior views demonstrating bilateral hallux valgus deformity with monophalangic great toes (arrowheads), a characteristic congenital abnormality of FOP.

Radiographs of both elbows showed ossification of the anterior compartments of the arms with bilateral periarticular heterotopic ossifications at the distal humerus (figure3).

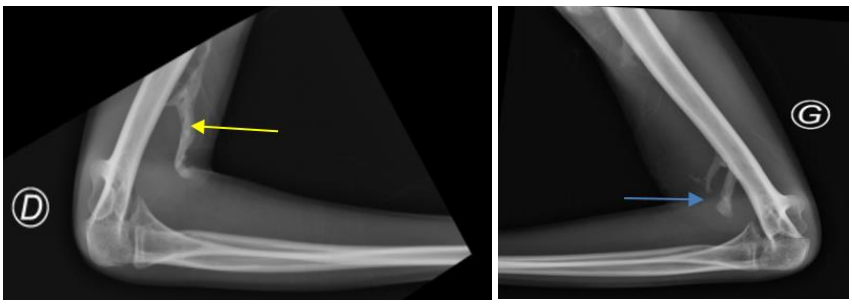

Figure 3. Lateral elbow radiographs

Lateral views of both elbows demonstrating heterotopic ossifications within the anterior soft tissue compartments. The image displayed on the right corresponds to the left elbow, while the image displayed on the left corresponds to the right elbow. Periarticular ossifications are noted at the distal humerus bilaterally (arrows), contributing to progressive joint stiffness.

CT scan of the entire spine demonstrated dense heterotopic ossifications with trabecular centers and peripheral corticalization, forming skeletal bridges along muscular planes. Multilevel posterior arch fusion from C3 to C6 was observed (figure 4).

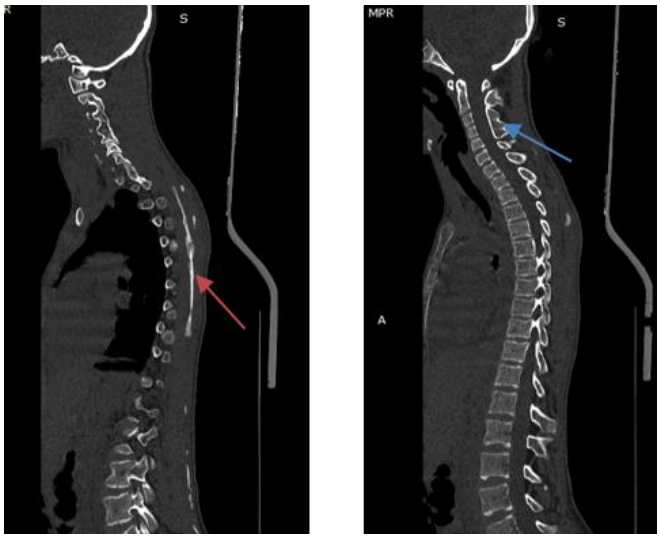

Figure 4. Spinal CT scan (sagittal reconstruction)

Dense, well-corticalized heterotopic ossifications forming osseous bridges along paraspinal muscles (arrows). Multilevel fusion of the posterior arches from C3 to C6 explains spinal rigidity.

DISCUSSION

Fibrodysplasia ossificans progressiva is characterized by progressive heterotopic endochondral ossification triggered by minor trauma, intramuscular injections, or inflammatory events. The underlying mutation in the **ACVR1** gene leads to abnormal activation of BMP signaling pathways and inappropriate osteogenic differentiation in soft tissues [1,2].

From a radiological standpoint, imaging findings are highly characteristic and often sufficient for diagnosis when combined with clinical features.

The most specific early radiologic feature is bilateral monophalangism of the great toes associated with hallux valgus deformity. This anomaly is present in nearly all patients and represents a pathognomonic sign [3]. In our patient, foot radiographs clearly demonstrated these abnormalities, strongly supporting the diagnosis.

Heterotopic ossification in FOP follows a predictable pattern of evolution, beginning with an early inflammatory stage characterized by soft tissue swelling, best detected on MRI, followed by an intermediate stage marked by amorphous calcifications, and ultimately progressing to a mature stage with well-organized ossifications demonstrating cortical and trabecular bone structure.

In our case, radiographs and CT demonstrated mature ossifications with peripheral corticalization and internal trabecular architecture. The lesions were oriented along muscular planes, a typical distribution pattern in FOP.

Spinal involvement significantly contributes to disability. CT imaging clearly demonstrated osseous bridges and posterior arch fusion from C3 to C6. Progressive ankylosis explains the cervical straightening and limited mobility observed clinically.

CT is particularly valuable in evaluating the extent of mature ossification and differentiating FOP from other causes of spinal ankylosis such as ankylosing spondylitis.

Radiologic differential diagnoses include myositis ossificans, progressive osseous heteroplasia, ankylosing spondylitis, and calcified soft tissue sarcomas.

However, the combination of congenital great toe abnormalities and progressive heterotopic ossifications strongly favors FOP [3].

Biopsy or surgical intervention can trigger explosive ossification and worsen the disease [2,4]. Therefore, radiologic recognition is critical to establish a non-invasive diagnosis and prevent harmful procedures.

CONCLUSION

Fibrodysplasia ossificans progressiva is a rare but devastating disorder. The presence of congenital great toe malformations combined with progressive heterotopic ossifications on imaging establishes the diagnosis. Radiologic evaluation plays a central role in early recognition, assessment of disease extent, and prevention of unnecessary invasive interventions.

REFERENCES

1. Pignolo RJ, Shore EM, Kaplan FS. Fibrodysplasia ossificans progressiva: clinical and genetic aspects. Nat Rev Rheumatol. 2011 ;7(12):701–9.
2. Kaplan FS, Shore EM, Connor JM. The FOP metamorphogene encodes a novel type I receptor that dysregulates BMP signaling. Clin Orthop Relat Res. 2009 ;467(12):3087–95.
3. Connor JM, Evans DA. Fibrodysplasia ossificans progressiva. The clinical features and natural history of 34 patients. J Bone Joint Surg Br. 1982 ;64(1):76–83.
4. Kaplan FS, Glaser DL, Shore EM, *et al.,* The medical management of fibrodysplasia ossificans progressiva: current treatment considerations. Clin Proc Intl Clin Council FOP. 2019.