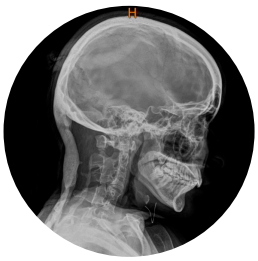

Fibrodysplasia ossificans progressive. Report of a rare case

Fibrodisplasia osificante progresiva. Presentación de un caso poco frecuente

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Summary

Fibrodysplasia ossificans progressive is a rare disorder that affects the connective tissue, characterized by progressive heterotopic ossification of soft tissues that can spread to joints. We present the case of a 16-year-old female patient with 13-year long history of development of multiple masses in the nuchal, dorsal and lumbar regions associated with progressive stiffness in the neck and trunk, with prior clinical history of congenital malformation of the right hallux (Hallux valgus). There are various radiological findings in this pathology, including the deformation of shortened hallux fingers with or without absence of a phalanx, ectopic calcifications within the soft tissues, wide and shortened femoral necks, heterotopic bone formations. In this case, heterotopic ossification was observed mainly with formation in the soft tissues and ligaments. It is very important to avoid or minimize any factor that may trigger or aggravate the development of plaques, such as trauma, injections or biopsy, hence the great utility of diagnostic images in this rare disease.

Resumen

La fibrodisplasia osificante progresiva es un trastorno poco común que afecta el tejido conjuntivo. Se caracteriza por la osificación heterotópica y progresiva de los tejidos blandos y se puede extender a las articulaciones. Se presenta el caso de una paciente femenina de 16 años con cuadro clínico de 13 años de evolución caracterizado por aparición de múltiples masas en la región nucal, dorsal y lumbar asociadas a rigidez progresiva en cuello y tronco. La paciente tenía antecedente de malformación congénita del *hallux* derecho (*hallux valgus*). Son múltiples los hallazgos radiológicos en esta patología, incluyendo la deformación de los *hallux* acortados con o sin ausencia de una falange, calcificaciones ectópicas en los tejidos blandos, cuellos femorales anchos y acortados, formaciones óseas heterotópicas. En el caso que se documenta se observa principalmente osificación heterotópica con formación en los tejidos blandos y ligamentos. Es muy importante evitar o minimizar cualquier factor que pueda desencadenar o agravar el desarrollo de las placas, como los traumatismos, inyecciones o toma de biopsia, de ahí la gran utilidad de las imágenes diagnósticas en este tipo de casos poco frecuentes.

Introduction

Fibrodysplasia ossificans progressiva (FOP) is a rare disorder affecting connective tissue. It is characterized by progressive heterotopic ossification of soft tissues that may extend to the joints. The prevalence of this disease is 1 in 2 million people, with no known sex, race, ethnicity or other risk factors (1). Most cases are sporadic, but there are reports of autosomal dominant germline transmission. Genetically, alterations of the *ACVR1 / ALK2* gene on chromosome 2q24 have been described (1-3). Pathophysiologically, overexposure of bone-forming protein has been documented, inducing heterotopic bone formation. Clinically, it is characterized by congenital malformations and the development of mature bone plates in muscle and other connective tissue-rich structures. Newborns have a normal appearance, except for the presence of congenital *hallux valgus*. There are other manifestations, such as shortening of phalanges, metatarsals and metacarpals, synostosis, clinodactyly, neck stiffness and hearing loss (1-4).

Clinical case

A 16-year-old female patient with a clinical picture of 13 years of evolution characterized by the appearance of multiple masses in the nuchal, dorsal and lumbar region associated with progressive stiffness in the neck and trunk. History of congenital malformation of the right *hallux* (*hallux valgus*). At the time of consultation, the patient reported severe limitation of movement in the neck, trunk and extremities, associated with chronic pain; also increase in the size of the masses, for which complementary imaging studies were performed.

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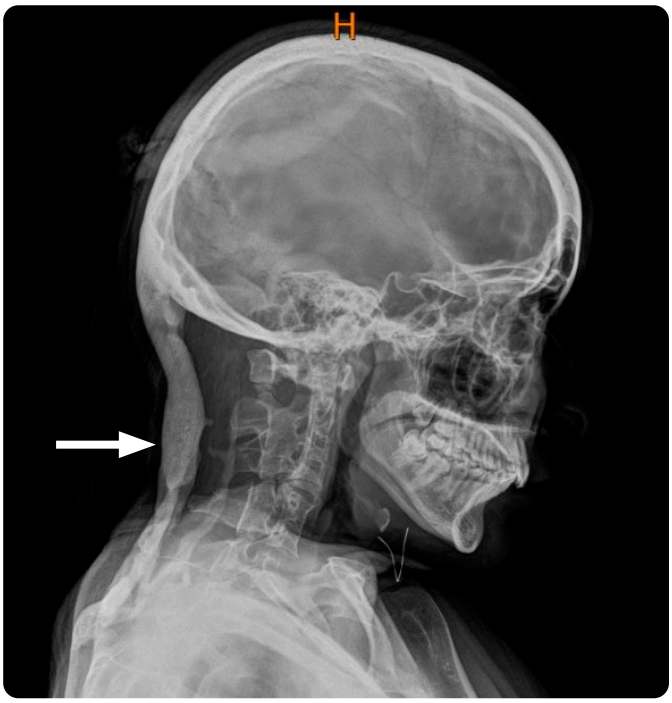

Figure 1. Simple skull X-ray, lateral projection. There is a large thick hyperdense plaque causing an anchorage of the skull and occipital bone (arrow).

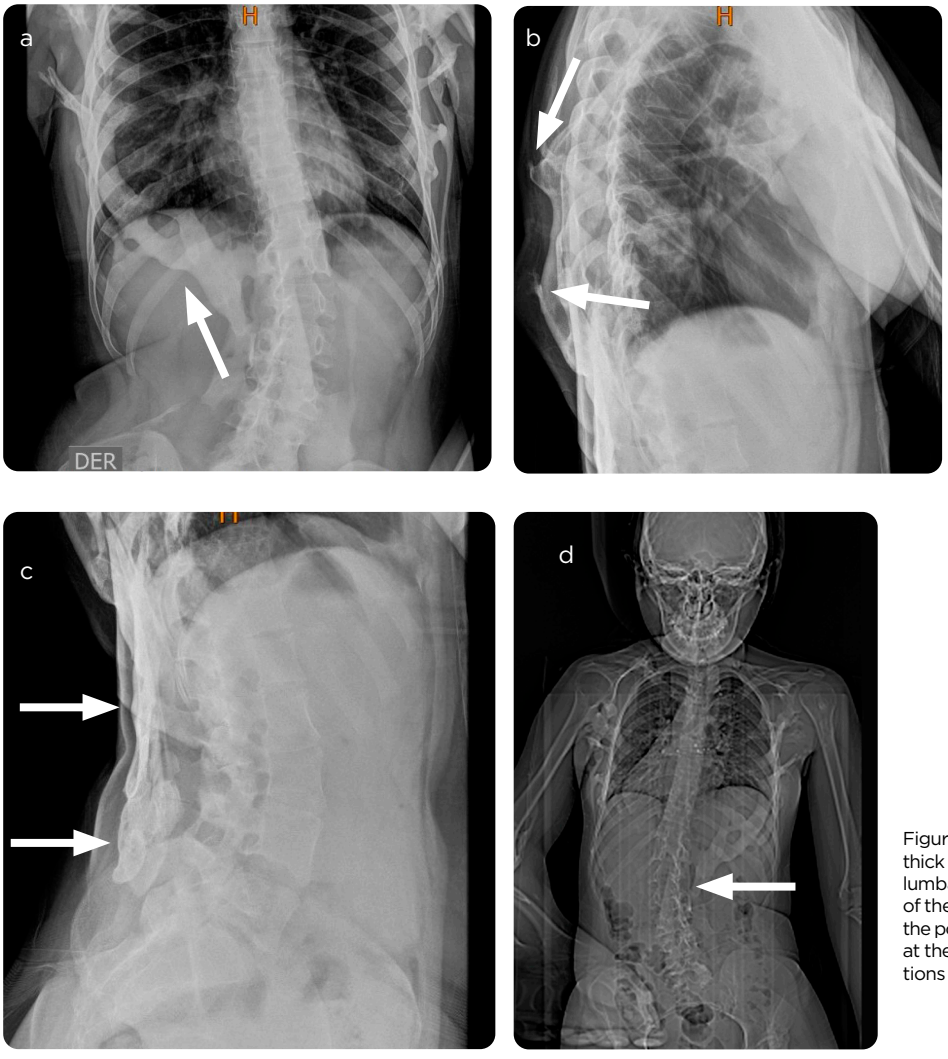

Figure 2. Plain spine X-ray. a) AP projection: prominent thick calcification in the soft tissues of the right thoracolumbar region (arrow). b) Lateral projection at the level of the thoracic spine: ligamentary calcifications next to the posterior dorsal arches (arrow). c) Lateral projection at the level of the thoracic spine: ligamentary calcifications next to the posterior dorsal arches (arrow).

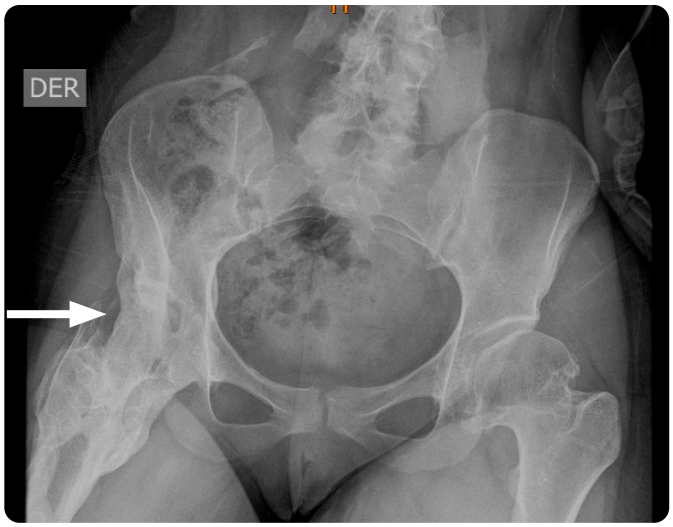

Figure 3. X-ray of the pelvis, anteroposterior projection. Extensive bony plate joining the iliac bone and the right femur (arrow), causing severe immobilization of the right coxofemoral joint, with almost complete joint fusion.

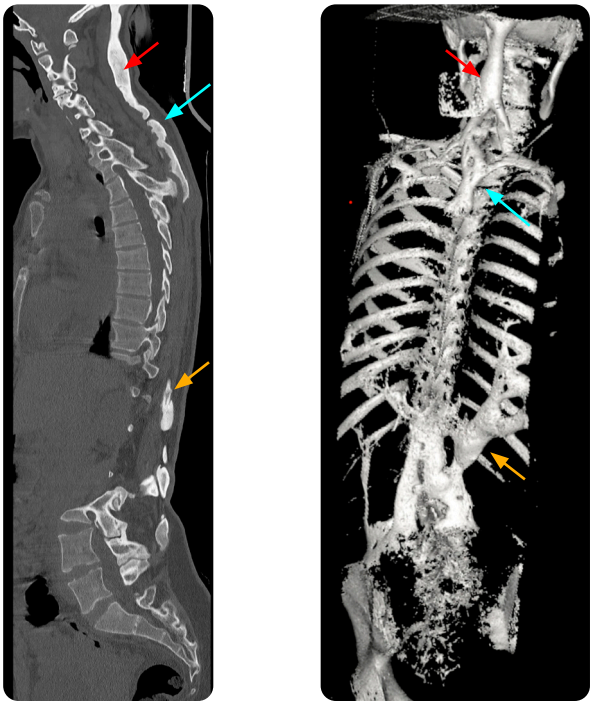

Figure 4. Simple CT of the complete spine. a) Sagittal. b) 3D reconstruction. Multiple prominent heterotopic ossification plaques are observed in the soft tissues adjacent to the cervical posterior arches (red arrow), in the connective tissue covering the dorsal spine and lumbar arches (blue arrow), and in the soft tissues of the right thoracolumbar region (yellow arrow).

Discussion

Progressive fibrodysplasia, also known as myositis ossificans, is a rare and disabling disorder. About 3600 cases have been described in the world (5). In Colombia, three cases have been documented from 2011 to the present (6-8). The classic clinical features are malformations of the hallux, which may be shortened with or without absence of a phalanx and progressive heterotopic endochondritis. Other clinical manifestations in the first decade of life include the appearance of extensive areas of episodic and painful soft tissue swelling. Over time, connective tissue, aponeuroses, fascia, ligaments, tendons and skeletal muscle develop into heterotopic bone or large ossification plates. These replace the tissues through a process of endochondral ossification that progressively causes immobilization of the patient. Another common finding is kyphoscoliosis as a consequence of the formation of asymmetric plates of heterotopic bone and ossification of paravertebral muscles (3).

Once ossification has begun, limitation of movement will depend on the region affected; neck stiffness is an early finding in most patients. This usually precedes the onset of calcifications in the temporomandibular joint, spinal deformities, cardiopulmonary restriction and mixed hearing loss due to ossification of the auditory system (5).

Differential diagnoses at an early stage include sarcoma, aggressive fibromatosis, post-traumatic myositis and rheumatoid arthritis; and at a late stage, osteosarcoma, progressive bone heteroplasty, hypervitaminosis D and traumatic myositis ossificans (1).

Timely identification and early diagnosis are essential and can be made by the use of computed tomography (CT) and magnetic resonance imaging (MRI). Early detection of inflammation of fascial and muscle planes before the development of heterotopic ossification is key; these findings can be confirmed by DNA-specific molecular testing of the *ACVR1* gene (2).

In the present case we observed mainly heterotopic ossification affecting soft tissues and ligaments, associated with the classic clinical manifestations and a history of right hallux valgus. With these findings, the diagnosis of fibrodysplasia ossificans progressiva was proposed, and in a new interrogation, the patient stated that she had molecular genetic studies confirming this pathology.

Early and timely diagnosis of this entity can improve the prognosis of patients, help to better control the symptoms and avoid major complications. In case the diagnosis is not clear or cannot be adequately suspected, procedures can be performed that end up being deleterious for the patient, such as biopsy and resection of the lesions, which are contraindicated due to the risk of an aggressive bone reaction in the soft tissues, which worsens the clinical picture.

Conclusion

There is currently no effective treatment for FOP. The goal is symptomatic relief, supportive measures and functional recovery, as well as genetic counseling (4). It is very important to avoid or minimize any factor that may trigger or aggravate the development of plaques, such as trauma, injections or biopsy; these procedures are contraindicated in these cases. Hence the importance of the radiology team for a correct and early diagnosis, in order to minimize invasive maneuvers that alter the integrity of the connective tissues.

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