FIBRODYSPLASIA OSSIFICANS PROGRESSIVE (FOP): CASE REPORT

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BACKGROUND

Fibrodysplasia ossificans progressive (FOP) is a rare and disabling genetic condition characterized by congenital deformity of the hallux and progressive heterotopic ossifications in the connective tissue. It has variable expression, reaching about 1.3 per million inhabitants, without distinction between races, genders and ethnicities. The gene that causes FOP is the type 1 activin A receptor, a signaling component of bone morphogenetic protein, which normally acts by inhibiting osteoblastogenesis. It is usually diagnosed clinically and radiologically in childhood, and laboratory findings are uncommon. Death is usually caused by pulmonary complications due to chest wall involvement. Treatment for FOP is still preventive and supportive.

CASE REPORT

RSB, 7 years old, brown, male, evolving for 1 year with multiple nodules in the dorsal region, initially of elastic consistency, mobile and painful, with subsequent stiffening of the affected parts. No relevant personal or family history. On physical examination, evidenced multiple, irregular, hardened nodules, adhered to deep planes, painless on palpation and without inflammatory signs. Functional limitation stood out, with reduced mobility of the axial skeleton and foci of ectopic ossifications in the paravertebral musculature (Figure 1). The hallux was significantly shorter than the other toes and in valgus (Figure 2). Laboratory tests showed no changes. Radiographs documented calcifications in soft parts of the neck (Figure 3) and dorsal region.

Figure 1. Congenital deformity of the hallux (hallux valgus).

Figure 2. Multiple nodules of stony consistency in the dorsal region.

CONCLUSION

Patients with FOP appear normal at birth, except for a bilateral malformation of the hallux, typically short and laterally displaced. The radiographic features of FOP include joint malformations, particularly of the hallux and soft tissue ossification. Fibrodysplasia ossificans progressive is a clinical diagnosis confirmed by molecular genetics. It should be suspected in children with hallux valgus, progressive soft tissue swelling, and nodules in the head or back. Biopsy of lesions can precipitate an outbreak and should be avoided, as well as other invasive procedures and trauma in general. There is no treatment to prevent or reverse the ossification associated with FOP. Management of FOP is supportive and focuses on preventing outbreaks, educating and counseling the patient and family, and improving quality of life.

KEYWORDS

Fibrodysplasia ossificans progressive, heterotopic ossifications, myositis ossificans.