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CLINICAL IMAGE

Fibrodysplasia ossificans progressiva: when a double skeleton is present

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Presentation

A 57-year-old female patient was referred to the Rheumatology Department due to progressive stiffness and limited mobility of the cervical and lumbar spine, followed by the shoulder, elbow, and hip joints. These complaints started in childhood and led to total dependence on daily routines. She had no relevant medical or family history. On physical examination (Fig. 1A, B), she presented deformity and anchylosis with severe rigidity in several joints and bilateral hallux shortening. The radiographs (Fig. 1C–K) showed generalized soft-tissue heterotopic ossification (HO). Laboratory investigations were normal. Due to clinical and radiographic findings, fibrodysplasia ossificans progressiva (FOP) was suspected and confirmed by the identification of a heterozygous c.617G > A p.(Arg206His) mutation in the *ACVR1* gene. The patient was treated symptomatically with a nonsteroidal anti-inflammatory drug and advised to avoid trauma. Respiratory function tests and an echocardiogram were requested to assess cardiorespiratory involvement. She also initiated a rehabilitation program.

Discussion

FOP is a rare (1:2,000,000) and disabling genetic disease characterized by progressive HO of soft connective tissues and congenital hallux malformations, caused by gain-of-function mutations in the *ACVR1* gene [1]. Hallux malformations (hallux valgus, malformed first metatarsal, and/or monophalangism) are present at birth and may alert to the early clinical diagnosis before radiographic evidence of HO [2]. Other frequent skeletal malformations include short and broad femoral necks, osteochondromas, and cervical spine abnormalities [3]. HO is preceded by episodic soft-tissue swellings (flare-ups) and may occur spontaneously or precipitated by soft-tissue injury, namely invasive medical procedures [3]. Cumulatively, HO forms rigid bridges across the joints, limiting mobility and affecting vital structures due to chest wall involvement. Therefore, most patients require a wheelchair in the second/third decade, and the median age at death is 40 years [4]. Despite no disease-modifying treatment, the management is focused on the prevention of soft-tissue injury, symptomatic relief of flare-ups, supportive measures, functional rehabilitation, complication surveillance, and genetic counselling [4].

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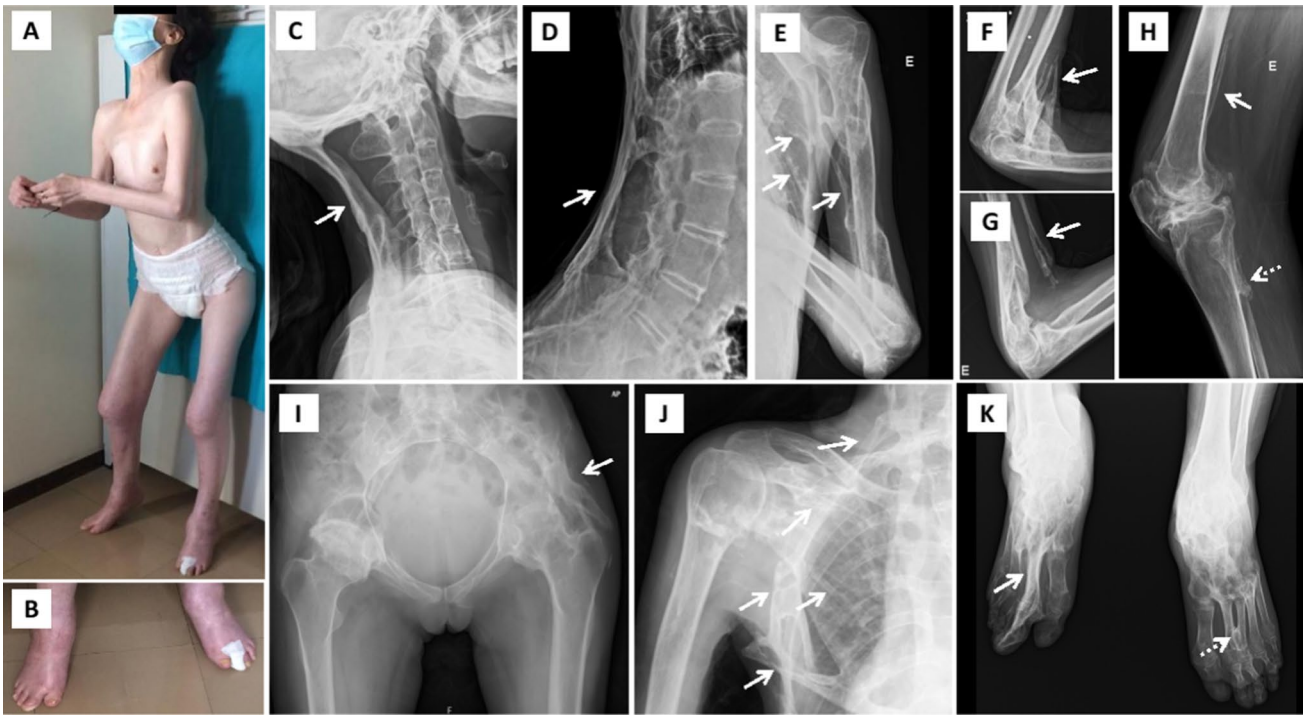

Fig. 1 Physical examination and radiographs of the main affected areas. Rigid posture with scoliotic deformity and significant bilateral shortening of the hallux were observed (**A**, **B**). The radiographs revealed generalized heterotopic ossification (full arrows), which involves the soft tissues of the spine (**C**, **D**), left arm (**E**), elbows (**F**, **G**), hip (**I**), thoracic region (**J**), and feet (**K**). Additional findings included tall and narrow vertebral bodies in the cervical spine with bridging ossification of the posterior elements (**D**), shortened and broad femoral necks (**I**), and exostosis/osteochondroma (dotted arrows) in the proximal region of the left tibia (**H**) and the left 3rd metatarsal (**K**). The foot radiographs showed bilateral hypoplastic hallux (**K**)

Compliance with ethical standards

Consent for publication The patient provided informed consent for the publication of this case study and related clinical images.

Disclosures None.

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