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IMAGES IN RHEUMATOLOGY

Fibrodysplasia ossificans progressiva: the stone woman

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A 37-year-old woman presented with a history of hallux shortness since childhood (Figure 1) and the detection of a nodule in the posterior cervical region at the age of 5 years. During adolescence, she experienced the progressive restriction of axial skeleton mobility, with intermittent pain. Several episodes of trauma and infection during her life had resulted in limitations in the movement of her right leg and both upper limbs, a condition that had become permanent. Despite limitations of chest mobility, the patient reported no respiratory symptom. She also presented significant restriction of temporomandibular joint movement, jaw fixation, and reduced cervical mobility, which caused eating difficulty. The final diagnosis was made based on clinical and imaging findings.

Physical examination revealed significant muscle atrophy, with stiffness and limitations of shoulder, elbow, knee, and temporomandibular joint movement. Extension and flexion of the cervical and axial skeleton were also limited, and bilateral shortened hallux was confirmed. Chest radiography (Figure 2) and computed tomography showed extensive areas of heterotopic ossification in the soft tissues of the chest wall and arms, with ligamentous ossification posterior to the cervical and dorsal vertebrae (fusing by an extensive bone bridge), as well as bone bridges between costal arches and in the maxillofacial region (Figures 3 and 4).

Fibrodysplasia ossificans progressiva, or stone man syndrome, is a severely debilitating genetic disorder of the connective tissue characterized by congenital malformations of the hallux and spontaneous, progressive, and irreversible heterotopic ossification at extraskeletal sites. The disease usually presents in the first decade of life as painful inflammations, either spontaneous or in response to trauma (soft-tissue injury, intramuscular injections, viral infection, muscular stretching, and falls), which later ossify with heterotopic bone, leading to severe movement restriction and other disabilities. Attempts to remove the heterotopic bone usually lead to episodes of explosive new bone formation. Immobility is cumulative, and most patients are wheelchair bound by the end of the second decade of life. In addition to progressive immobility, life-threatening complications of this condition include severe weight loss following ankylosis of the jaw and thoracic insufficiency syndrome with restrictive lung disease, pulmonary hyper-

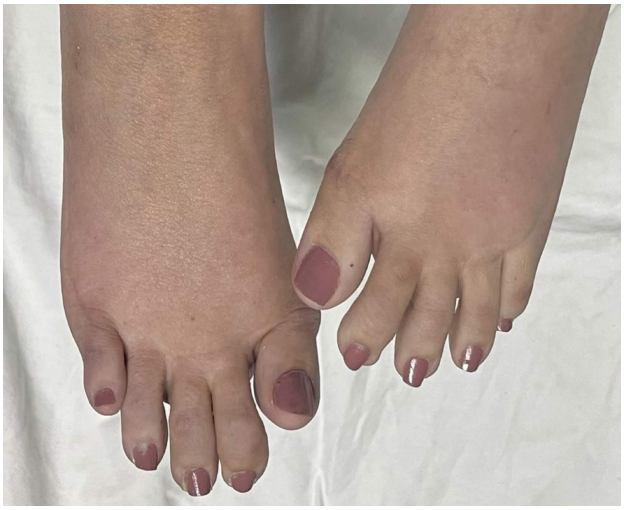

Figure 1. Photograph of the patient's feet showing typical bilateral short hallux.

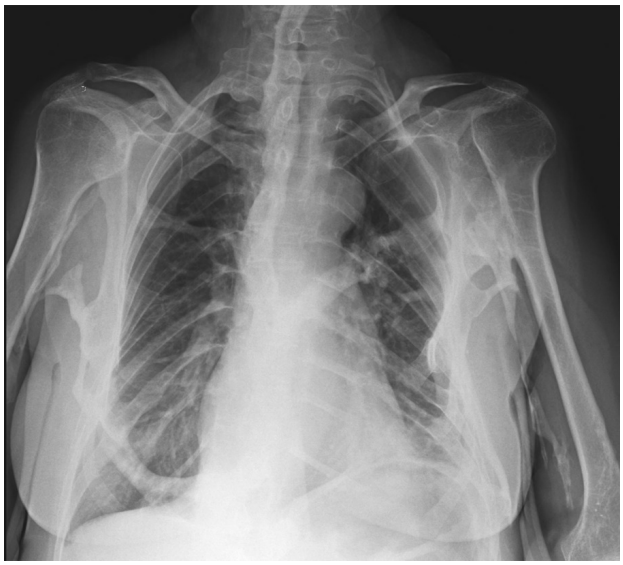

Figure 2. Chest radiograph showing extensive heterotopic ossifications of the chest wall with osseous bridges overlying the muscles and bridging multiple ribs, the upper limbs, and the shoulders.

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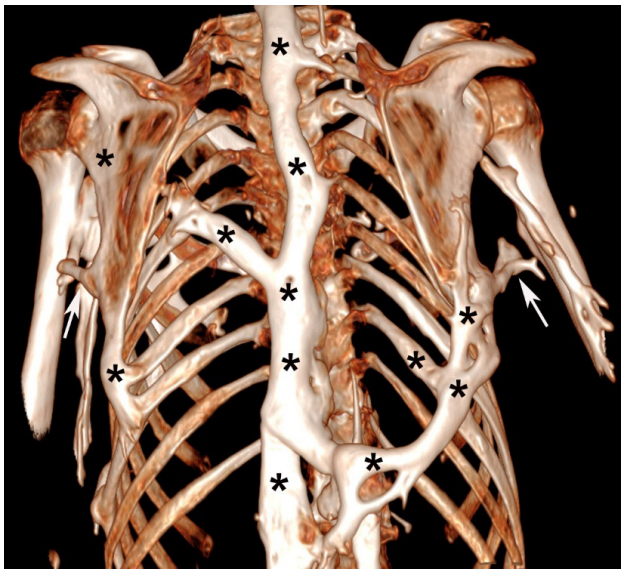

Figure 3. Posterior view of a 3D volume-rendered image of the chest demonstrates a large branching mass of heterotopic ossification of the soft tissues, muscles, and ligaments (asterisks) that fuses with the scapulae and ribs on both sides. The mass also fuses with both humerus (white arrows).

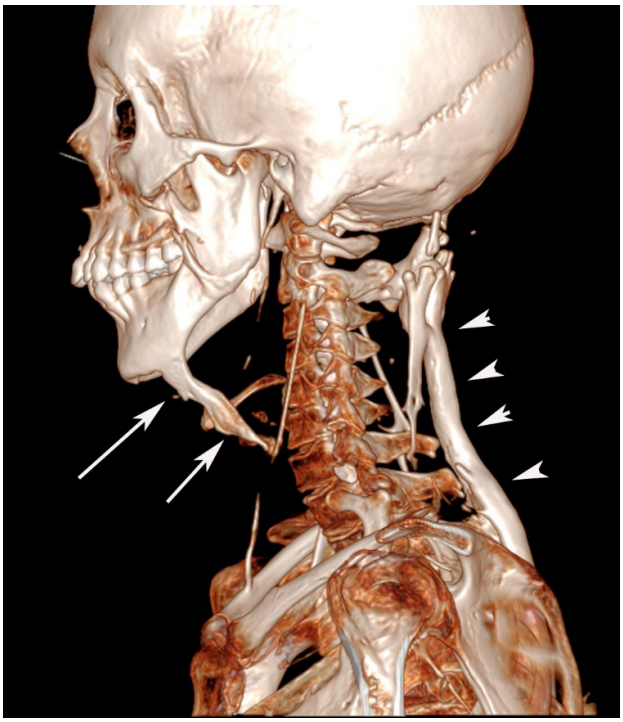

Figure 4. Three-dimensional CT image of the cervical spine shows heterotopic bone formations descending from the occiput (arrowheads) and jaw (arrows), corresponding to ossification confluent with muscles and ligaments.

tension, pneumonia, and right-sided congestive heart failure, eventually causing death. The median lifespan of patients with fibrodysplasia ossificans progressiva is approximately 40 years, and these patients commonly die of pulmonary or cardiac complications. At present, there is no definitive treatment[1-4].

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