Left clavicular fracture

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ANSWER: Pyknodysostosis

Pyknodysostosis, also known as Maroteaux-Lamy disease and Toulouse-Lautrec syndrome, is a rare autosomal recessive genetic sclerosing bone dysplasia with an estimated incidence of 1.7 per million births with no gender predilection [1]. The disorder is caused by a mutation of a lysosomal cysteine proteinase, cathepsin K, that is required for normal degradation of type I collagen, the major constituent (95 %) of organic bone matrix [1, 2]. The resultant osteoclast dysfunction impairs bone remodeling with resultant dense but fragile bones. Although a cathepsin K gene mutation analysis can provide confirmation, the diagnosis is usually based on the characteristic clinical and radiographic findings [3].

Clinically, patients demonstrate short-stature measuring <150 cm (4 feet 11 inches) and dysmorphic facial features including a beaked nose and micrognathia. Underlying bone

fragility is clinically manifest as multiple fractures after only minor trauma [4]. It is believed that the famous French post-impressionist painter, Henri de Toulouse-Lautrec had this disorder and suffered from bilateral femoral fractures at a young age from falls [5]. Typical locations for pathologic fracture include the jaw, clavicles, and lower extremities. Because of impaired vascularity in the dense bones, there is a propensity to develop jaw osteomyelitis after dental extraction or injury that can be refractory to treatment [6]. Affected individuals are usually diagnosed at a young age because of the characteristic appearance and bone fragility, but in some instances, as in this case, the diagnosis is made later in life. Cognitive function and life expectancy are normal [1].

Radiographic findings include generalized osteosclerosis with preservation of the medullary canal of long bones. For our patient, the osteosclerosis was not radiographically obvious but the DEXA scan detected increased bone density. There is foreshortening of the distal phalanges with irregular terminal tufts simulating acro-osteolysis, a finding considered a pathognomonic feature [4]. The calvarium demonstrates open fontanelles, widened sutures and, often times, wormian bones. There is also hypoplasia of the paranasal sinuses and an obtuse mandibular angle. Less commonly there is a thoracic deformity including kyphosis and pectus excavatum and distal clavicular hypoplasia. Segmentation abnormalities and spondylolisthesis in the cervical or lumbar spine can also be seen [7]. Long bone fractures are in the midshaft and are typically transverse rather than oblique, as with other pathological bone fractures.

The radiological differential diagnosis includes osteopetrosis, which also presents with brittle sclerotic bones; however, with osteopetrosis, sutures of the skull are closed and there is typically obliteration of the medullary canal of the long bones with resultant anemia. Cleidocranial dysplasia may have some similar imaging features including open

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fontanelles and wormian bones; nevertheless, there should not be increased bone density and the clavicles are typically hypoplastic. The appearance of the hands of our patient could raise consideration of other causes of acro-osteolysis; however, the other imaging features are characteristic of pyknodysostosis.

Management of patients with pyknodysostosis is primarily supportive. Fractures are treated conservatively or with internal fixation if necessary. Special consideration should be given to patients undergoing general anesthesia due to anticipated difficult intubation and cervical spine anomalies [8]. In conclusion, pyknodysostosis should be considered in the differential diagnosis of patients with elevated bone mineral density and fragility fractures. While unusual, the disorder may go unrecognized until adulthood and be first diagnosed by radiological findings.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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