Osteogenesis imperfecta type V

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Osteogenesis imperfecta type V was first described in 2000. It is a distinct clinical entity with unique clinical, radiological, and histological features. Clinically, it is only moderately deforming. Patients have normal sclera and teeth. Radiological diagnostic criteria include a triad of calcification of the radioulnar interosseous membrane, presence of hypertrophic callus at fractures or post-operative sites, and radiodense metaphyseal band adjacent to growth plates [1]. Histologically, it is distinguished by a mesh-like pattern of lamellation under polarized light microscopy for iliac bone samples.

Ossification of the interosseous membrane of the forearm is a constant feature, although it may vary in its extent. The presence of ossification can severely limit movements of the forearm, and is associated with secondary dislocation of the radial head [1, 2]. The frequency of radial head dislocation/subluxation is significantly higher in type V osteogenesis imperfecta (86%) than in the other types (0% to 29%) [3]. The presence of interosseous membrane ossification in a pediatric patient, with or without radial head dislocation, should prompt radiologists to consider OI type V as a diagnosis.

The formation of hypertrophic callus, if present, is the most conspicuous clinical symptom in OI type V [1]. It is reported that while not all OI type V patients have hypertrophic callus formation, all patients with hypertrophic callus formation are OI type V in the proper clinical context [1, 4]. Lesions form during the growth years at sites of rapid periosteal apposition. The long bones are most often affected, particularly in the lower extremities [4]. Hypertrophic callus can be precipitated by fracture or surgery, or arise spontaneously. It can become very large or even mimic osteosarcoma [2, 5]. In unclear cases, MRI and CT can be helpful in distinguishing hypertrophic callus from sarcoma [6, 7]. Evolution of the lesions is variable, ranging from complete resolution to significant persisting morbidity.

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