

## Jaccoud's arthropathy in osteogenesis imperfecta

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Type I osteogenesis imperfecta is a genetic disease associated with mutations in the pro-alpha1 chains of type 1 collagen encoder gene COL1A1. It has an autosomal dominant pattern of inheritance, and leads to reduction in the amount of structurally normal collagen and subsequently, disorders of the extracellular membrane of skin, tendons and bone [1].

We present a 60-year-old woman whose son has type I osteogenesis imperfecta (OI) (heterozygous c.2032>T; p.(Glu678\*), COL1A1 exon 31), that presented to the Rheumatology clinic with progressively worsening swan-neck and z-thumb deformities despite the absence of arthralgia or signs of arthritis. Her medical history included osteoporosis (T-score -3.1 at the femoral neck), a low-impact radial head fracture, and multiple teeth losses. Examination revealed short stature (143 cm), reversible joint deformities without arthritis. She had lumbar scoliosis

and blue sclerae bilaterally. Laboratory results were negative for rheumatoid factor, anti-citrullinated protein, and antinuclear antibodies. Hand ultrasound confirmed the absence of significant synovitis, except for low-grade synovitis in the left fifth proximal interphalangeal joint, radiocarpal joint, and right mid-carpal region. Radiographs showed no bone erosions or severe joint damage (Figure 1). Based on the family history, clinical features, and imaging findings, the patient was diagnosed with type I OI with associated Jaccoud's arthropathy.

Musculoskeletal features of type I OI typically include generalized osteoporosis with associated fractures and short stature [1]. Tendon laxity due to pathological changes in type I collagen has been documented, manifesting as scoliosis, spondylolisthesis, ankle laxity, and flatfoot [2]. Upper limb laxity primarily manifests as polyarthralgia, with deformities only rarely reported [3].



**Figure 1.** Jaccoud's arthropathy - physical examination and radiographic findings. (A) Jaccoud's arthropathy—luxation of the 1st metacarpophalangeal joint (MCPJ) and swan-neck deformities. (B) Hand radiography (anteroposterior view): z-thumb deformity, multiple subluxations of proximal interphalangeal (PIPJ) and distal interphalangeal (DIPJ) joints, exuberant osteoarthritis of DIPJs. (C) Hand radiography (oblique view): luxation of the 1st right MCPJ and subluxation of the left 1st MCPJ, subluxation of multiple PIPJs and swan-neck deformities of DIPJs, in the absence of erosions.

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Jaccoud's arthropathy is a deforming non-erosive arthropathy characterized by ulnar deviation of the 2nd to 5th fingers due to metacarpophalangeal joint subluxation, amenable to correction with physical manipulation. This may represent a late-stage manifestation in some individuals with OI, after chronic benign ligamentous and capsular laxity and occasional low-grade synovitis [4]. In this case, the patient presented with a clinical picture readily misconstrued as rheumatoid arthritis (RA) by the untrained eye. Despite sharing defective collagen synthesis as a pathophysiologic mechanism [5], RA deformities are fixed, often accompanied by overt synovitis and elevated inflammatory markers. Symptomatic treatment with non-steroidal anti-inflammatory drugs, short course of steroids, methotrexate or hydroxychloroquine have been used for management of this condition in patients with RA and systemic lupus erythematosus, with variable success [4]. Physical therapy and orthotic devices may play a role in ameliorating symptoms [4]. In the absence of pain, analgesia wasn't warranted in this patient. This case highlights Jaccoud's arthropathy as a tell-tale sign of a non-degenerative disease in individuals with a genetic metabolic bone disorder.

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## CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

## AUTHOR CONTRIBUTIONS

FRM collected the clinical data, wrote the paper and obtained the patient's informed consent. MLR and ATC collected the imaging data and provided photo documentation during the consultation. VST was responsible for conceptualization and supervision. All co-authors revised and approved the final version of the manuscript.

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