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## CASE REPORT

# Primary Idiopathic Osteolysis Syndrome: Case Report And Review Of Literature

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### ABSTRACT

Primary idiopathic osteolysis (massive osteolysis, essential osteolysis, progressive atrophy of bone, spontaneous absorption of bone, phantom bone) is an extremely rare occurrence. This disorder is characterised by spontaneous or post traumatic bone resorption of uncertain etiology with unpredictable prognosis, and any effective therapy is largely unknown [1]. We report the clinico-radiological features of a 10-year-old male patient with this condition, and present a short review of the available literature.

**Key words:** Osteolysis, Bone disorders

### Introduction

Primary idiopathic osteolysis was first described nearly 150 years back by Jackson in 1838. On literature review, it is evident that various eponyms have been used to describe this mysterious disorder of the musculoskeletal system. In 1955, Gorham and Stout provided a more comprehensive report on this subject [1],[2],[3]. Based on their experience and case reports available in literature, they found that most cases of osteolysis syndromes are associated with abnormal vascular proliferation in the vicinity of resorbed bone, the alteration of acid-base balance in local milieu being responsible for the pathology. The nomenclature these authors assigned to such osteolysis secondary to vascular proliferations was “Vanishing Bone Disease”. This disease is also known as Gorham’s disease [2],[3]. No specific cause could be ascertained for a minority of cases that have been henceforth classified under the umbrella of “Primary Idiopathic Osteolysis Syndromes”. The purpose of this article is to report this rare entity and to review the etiopathology, clinical presentation, radiographic

findings, differential diagnoses, and treatment options for patients with primary idiopathic osteolysis syndromes [3].

### Case Report

A 10-year-old male patient presented to us with progressively deforming joints of hands and elbow for the last three years.

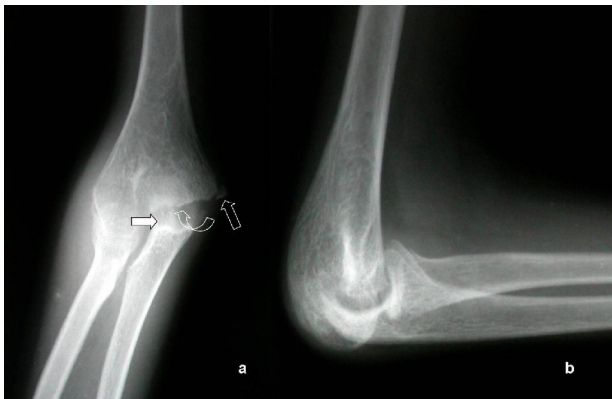


**Table/Fig. 1.** Plain radiograph of both hands shows tapered / pointed ends of terminal phalanges (block arrows). The epiphyses of phalanges, metacarpals (hollow arrows) and distal ends of radius and ulna (curved arrows) and carpal bones (straight arrows) appear irregular and reduced in size.

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He was a school going child demonstrating normal scholastic and developmental milestones. There was a history of insidious onset of pain, limitation of motion, and progressive weakness in bilateral wrist and elbow joints with no associated soft tissue swelling. There was no history of significant trauma.

The routine laboratory blood tests including full blood count and erythrocyte sedimentation rate were normal. Neither rheumatoid factor nor antinuclear factor was detected. The serum alkaline phosphatase level was minimally elevated with a value of 134 IU/l. The pedigree analysis was non-contributory. A skeletal survey was done. The survey indicated that radiolucent intramedullary and subcortical foci with erosions of the periarticular cortex in bilateral metacarpophalangeal and interphalangeal joints [Table/Fig. 1], at the lower end of humerus and the upper ends of radius and ulna [Table/Fig 2] and in the vertebral end plates [Table/Fig 3]. All epiphyses appeared reduced in size and irregular with irregularity and remodelling noted at contiguous metaphyses. Specky periosteal reaction was observed at certain sites.



**Table/Fig. 2.** Plain radiograph of right elbow showing resorption and supero-medial subluxation of trochlear epiphysis (hollow arrow). Adjacent metaphysis (curved arrow) and articular surfaces of radius and ulna (block arrow) are irregular.

Tapering or 'pointing' of other bones including carpals with atrophy of overlying soft tissues was associated. On the basis of this typical pattern of regional osseous destruction on the radiographs and the exclusion of other differential diagnostic considerations, the patient was diagnosed as a case of primary idiopathic osteolysis. The patient was placed on anti-osteoclastic medication that is ,

bisphosphonates, with calcium supplements and was doing well till the last out patient follow up.

### Discussion

Destruction of bone (osteolysis) or cartilage (chondrolysis) may be an associated feature of numerous neoplastic, infectious, metabolic, traumatic, vascular, congenital, or vascular disorders. Severe and progressive cartilaginous and/or bone dissolution at multiple sites can lead to considerable deformity in some of these disorders like Rheumatoid arthritis, Juvenile Idiopathic arthritis, Psoriatic arthritis, Systemic Lupus erythematosus, Multicentric Reticulohistiocytosis, .



**Table/Fig. 3.** Plain radiograph of dorsal spine showing irregularity of the end plates representing changes at the superior and inferior vertebral epiphyses (curved arrows).

In addition, osteolysis may occur after frost bite, animal bite, irradiation, occupational injury, thermal injury, and electrocution [3]. A group of heterogeneous conditions remain in which the cause is not known. These conditions are termed Primary Idiopathic Osteolysis Syndromes. A long list of conditions is included under this rubric, the chief being Acro-osteolysis of Hajdu and Cheney, Massive Osteolysis of Gorham, Idiopathic Multicentric Osteolysis, Carpo-tarsal Osteolysis. Other rarer conditions include neurogenic osteolysis, Acro-osteolysis of Joseph, Acro-osteolysis of Shinz, Farber's Disease, Winchester Syndrome, and Osteolysis with Dendritic

synovitis[3],[5],[6]. All the above disorders can be classified or differentiated on the basis of age of onset, presence or absence and type of genetic transmission, major location of osteolysis, and associated clinical features[1]. We labelled our patient to be suffering from Carpo-tarsal osteolysis due to the predominant progressive involvement of small bones of the hand and feet [4],[6].

Tyler and Rosenbaum [6] proposed the terminology. Till then, it was known by multiple names, such as Essential Osteolysis, Idiopathic Osteolysis, Essential Acro-osteolysis, Familial Osteolysis, Hereditary osteolysis, Carpal and Tarsal agenesis, Familial Dysostosis Carpi and bilateral carpal necrosis [3],[6]. The basic pathophysiological event in these idiopathic conditions is increased osteoclastic activity with increased vascularity but no associated inflammation. The destroyed areas are replaced by fibrous tissue. The joints are primarily normal. Increased urinary hydroxyproline and serum phosphate is a constant feature [1]. These metabolites however were at top normal limits. Therefore, they were not considered significant enough for a diagnosis.

Idiopathic Multicentric Osteolysis, which is also known as Carpotalar Osteolysis, can be further divided into two entities, Multicentric osteolysis with Nephropathy and Hereditary Multicentric Osteolysis. A third miscellaneous group is reserved for conditions that do not have features of either. The former is a sporadic (occasionally dominant) condition having onset in infancy and childhood. It mainly affects the carpals, tarsals, and elbows. Associated features are osteoporosis, deformities, hypertension, and renal failure and could be fatal in a few cases. The latter is mostly Autosomal Dominant (occasionally AR or Sporadic). Its onset is between one to five years of age, with the carpals, tarsal, elbow, and digits being chiefly involved. Progressive deformities are an important feature [6]. The present case can be easily classified as group II because no renal insufficiency has been established.

The key imaging feature in both the conditions is progressive dissolution and disappearance of carpals and tarsals. Tapering of adjacent bones is observed mainly in Multicentric osteolysis with Nephropathy [3]-[7].

The main differential diagnostic considerations are Leprosy, Juvenile Chronic Arthritis, Diabetes Mellitus, Winchester's Syndrome (characterized by the presence of corneal opacities), and Farber

disease (characterized by the presence of s/c nodules)[3].

The clinical presentation is variable and depends on the site of involvement. It often takes many months or years to diagnose correctly the offending lesion. A high index of clinical suspicion is required to arrive at an early and accurate diagnosis. It is progressive in most patients. In some cases however, the disease process is self-limiting. The clinical course is generally protracted but rarely fatal, with eventual stabilization of the affected bone being the most common sequelae. Chylous pericardial and pleural effusions may occur due to mediastinal extension of the disease process from the involved vertebra, scapula, rib, or sternum and can be life threatening. High morbidity and mortality is observed in patients with spinal and/or visceral involvement. No complication was encountered in the present case [3]-[7].

### Conclusion

The diagnosis of primary idiopathic osteolysis syndrome involves exclusion of other commoner conditions that can affect a particular patient. In our case, we excluded other possibilities, such as Juvenile Idiopathic arthritis and Reticulohistiocytosis by lab and clinical data.

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