

TALE OF A MELTING FINGER: RARE CAUSE FOR PROGRESSIVE SHORTENING OF A DIGITGopi Surendra¹, Sudha Kiran Das², Sandeep Ramna³, Kamal Kumar Sen⁴, Vikram B. Pati⁵¹Resident, Department of Radio-diagnosis & Imaging, JSS Medical College & Hospital.²Assistant Professor, Department of Radio-diagnosis & Imaging, JSS Medical College & Hospital.³Resident, Department of Radio-diagnosis & Imaging, JSS Medical College & Hospital.⁴Professor, Department of Radio-diagnosis & Imaging, JSS Medical College & Hospital.⁵Assistant Professor, Department of Radio-diagnosis & Imaging, JSS Medical College & Hospital.**ABSTRACT**

Vanishing bone disease aka Gorham- Stout disease is a rare entity of unknown aetiology, with no definite gender, race or sex predilection. It is characterised by destruction of osseous matrix and proliferation of vascular structures resulting in destruction and resorption of bone. It is usually monostotic, but can be polyostotic. Symptomatology is varied and depends on the site of involvement. We present a case and a short review of aetiopathogenesis, the radiological evolution of this rare condition and the therapeutic options available till date.

KEYWORDS

Digit, Gorham disease, Radiology.

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INTRODUCTION: Gorham's disease consists of progressive idiopathic osteolysis of a bone or bones adjacent and surrounding an angiomatous focus, without respecting joint borders. It can affect any part of the skeleton; most commonly affected are cranium, shoulder and pelvis. The onset of the disease is insidious, the course of the disease is progressive and the clinical presentation is varied. The disease is benign in aetiology as well as its course; however, complications comprising pericardial and pleural effusions bring about fatality. Radiological examinations, especially x-rays, computed tomography (CT) and magnetic resonance imaging (MRI), in association with bone biopsies are essential for making the diagnosis.

CASE REPORT: A 35-year-old farmer presented to us with gradual onset of swelling with painless foreshortening of his right middle finger for over the last two years. At the onset of disease, he noticed minimal localised soft tissue swelling and erythema. Subsequently, there was progressive collapse of the digit. No preceding history of trauma. No history of pustular eruptions or discharge was noted. Physical examination revealed localised thickening of the soft tissues, which was not tender, preservation of the skin creases corresponding to the interphalangeal joints and an impalpable middle phalanx was noted. Blood investigations revealed no metabolic abnormality, serum calcium and phosphorous levels were normal, parathyroid levels also were normal. Erythrocyte sedimentation rate was normal and the tuberculin test was negative. Sequential plain

radiographs were evaluated (Figure 1a, 1b and 1c; 2013, 2014 and 2015 respectively), and patient was evaluated with contrast enhanced MRI (Figure 2, in 2015). As the disease was isolated to the digit with no other areas of skeletal involvement, a differential diagnosis of Tuberculous dactylitis or an angiomatous lesion involving the digit as in vanishing bone disease was given based on the clinico-radiological evaluation. Following reparative surgical exploration, histopathology confirmed the diagnosis of Gorham-Stout disease.

DISCUSSION: Jackson first reported a case of massive osteolysis of the humerus in a 12-year-old boy.^[1] In 1955, Gorham and Stout further characterised the main pathologic features of this rare disease in about 24 cases as nonmalignant intraosseous proliferation of hemangiomas or lymphangiomas tissue that caused massive osteolysis^[2,3] Varied nomenclature has been given to this disease including vanishing bone disease, phantom bone, massive osteolysis, hemangiomas, lymphangiomas, and Gorham-Stout disease [GSD], but it is best known as Gorham's disease.^[4]

It is characterised by benign vascular proliferation of bone with progressive bone loss (osteolysis) associated with the overgrowth (proliferation) of lymphatic vessels. It is most commonly seen in children and young adults in either sex. Till date, about 300 cases have been reported in the literature. No definite race, sex predilection (1.6:1; male: female ratio) or geographic distribution is attributable for GSD.

GSD may affect any bone in the body, but there is a predilection for bones that develop by intramembranous ossification, with the shoulder girdle and mandible being the most common bones affected. The lesion is typically nonexpansile and nonulcerative and is usually monocentric, but locally aggressive, with resorption of the affected bone. The vascular lesion may spread into soft tissue and

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Corresponding Author:

Dr. Sudha Kiran Das,

*Department of Radio-diagnosis & Imaging,
JSS Medical College & Hospital, JSS University, M. G. Road,
Mysore-570004, Karnataka, India.*

E-mail: sudha7492@gmail.com

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contiguous bones. A few case reports have documented a polyostotic occurrence of this disease as well. Symptoms at presentation are dependent upon the location(s) of the disease; the most common symptom is localised pain.

Aetiology of GSD is still elusive. Histopathologically, the process is characterised by replacement of normal bone by an aggressively expanding but non-neoplastic vascular tissue, similar to a haemangioma or lymphangioma. Proliferating neovascular tissue causes massive bone loss. In the early stage of the lesion, the bone undergoes resorption, and is replaced by hypervascular fibrous connective tissue and angiomatous tissue [Figure 3]. Involved bones show a non-malignant proliferation of thin-walled vessels; the proliferative vessels may be capillary, sinusoidal or cavernous. In late stages, there is progressive dissolution of the bone leading to massive osteolysis, with the osseous tissue being replaced by fibrous tissue.^[5,6]

Radiologically, the initial feature of Gorham disease may reveal radiolucent foci in the intramedullary or subcortical regions that resemble osteoporosis. Subsequently, progressive dissolution and disappearance of a portion of the bone may occur. The osteolysis may extend to the contiguous bone and cross the intervening joint [Figure 1]. The osseous destruction may persist for a period of years and may eventually stabilise. CT is useful in the delineation of the soft-tissue extension, and it enables biopsy guidance. Three-dimensional CT reconstructions have been valuable to the orthopaedic surgeon planning a reconstruction attempt. Magnetic resonance imaging (MRI) shows complete resorption of bone and replacement with infiltrative soft tissue that is of low signal intensity on T1-weighted imaging and high signal intensity on T2, with intense enhancement on contrast imaging [Figure 2]. Tc-99 m bone scintigraphy may demonstrate increased uptake of the radiopharmaceutical agents on the initial images and, subsequently, an area of decreased uptake corresponding to the diminished bone region. Lymphangiography has been used to assess the thoracic duct in patients with chylothorax. The lymphatic vessels and nodes have a normal appearance, although altered lymphatic flow can lead to obstruction and oedema. Angiography depicts absence of neovascularity in the involved area.^[7,8]

Differential diagnosis includes causes of osteolysis such as infection, cancer (primary or metastatic), inflammatory or endocrine disorders. Other diseases which can mimic GSD are generalised lymphatic anomaly; however, this lacks the progressive osteolysis seen in GSD. Multicentric carpotarsal osteolysis with or without nephropathy, autosomal recessive carpotarsal osteolysis, hereditary sensory and autonomic neuropathy type 2, Farber lipogranulomatosis, Torg-Winchester syndrome, idiopathic phalangeal acro-osteolysis are also a few differentials to be considered.

Essential osteolysis is characterised by resorption of carpal bones, tarsal bones, or both, with progressive renal failure. The renal failure, multifocality, and absence of vascular proliferation within the involved bones distinguish essential osteolysis from Gorham syndrome. Hereditary osteolysis occurs in childhood, lacks vascular proliferation,

involves primarily the hands and feet, and tends to be multicentric. Osteolysis may also result from systemic diseases such as rheumatoid arthritis, syphilis, and hyperparathyroidism; however, these disorders are histologically and clinically distinct from Gorham syndrome.^[9,10] Finally, myeloma and lymphoma should be considered, as they are common diseases that can cause a moth-eaten appearance. Patients with these diseases generally have associated clinical and radiologic findings that aid in making the correct diagnosis.

The goal of therapy is to stabilise progressive disease and provide surgical stabilisation of affected regions. Anti-osteoclastic drugs like bisphosphonates and/or interferon alpha 2b & Sirolimus used along the medical lines of management. The surgical options include resection of the lesion and filling with autologous or heterologous graft material or reconstruction of the joint through arthroplastic resources. Radiotherapy may be used in combination with these therapies, but is generally reserved for refractory or rapidly progressive disease.^[10] Prognosis depends on the extent and location of affected areas and is varied from mild disease that is stable over many years as opposed to fatal occurrences with involvement of craniofacial and/or thoracic areas.

CONCLUSION: The diagnosis of Gorham's disease is one of exclusion and is based mainly on clinico-radiological findings, evolution and compatible histological findings. The aim of this case report is to emphasise the Gorham's disease as a rare differential diagnosis for progressive shortening of a digit and to illustrate the evolution of radiological appearances over a period of time with particular reference to MRI.



Figure 1: Serial Radiographs: (1a) At the onset of disease. (1b) A year later (1c) Two years later: Progressive resorption of the middle phalanx with associated soft tissue thickening. No periosteitis/ phleboliths noted



Figure 2: Pre and post Contrast MRI correlated with photograph of the hand and correlative plain radiograph: MRI show intensely enhancing soft tissue mass associated with non-visualization of middle phalanx of right middle finger and tapering of the proximal phalanx (licked candy appearance)

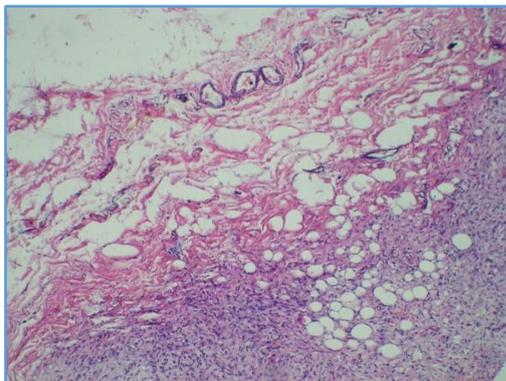


Figure 3: Photomicrograph of the bone specimen shows fibromyxoid tissue with highly vascularised collagenous tissue and the small capillary-like vessels

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