

## Bone Manifestations of Gaucher Disease: A CT-Based Case Report of H-Shaped Vertebrae and Aseptic Osteonecrosis

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## Abstract

## Case Report

Gaucher disease (GD) is a lysosomal storage disorder that frequently results in severe skeletal morbidity [1,2]. While marrow infiltration is the primary event, the secondary structural complications—specifically vertebral collapse and osteonecrosis—are best characterized by computed tomography (CT) [1]. We report the case of a 25-year-old male with long-standing GD presenting with classic "H-shaped" vertebrae and bilateral femoral head involvement. This report highlights the CT semiology of Gaucher-related bone disease, focusing on the biconcave vertebral deformity resulting from central endplate ischemia and the progression of aseptic osteonecrosis.

**Keywords:** Gaucher disease; H-shaped vertebrae; Biconcave vertebrae; Osteonecrosis; CT imaging; Skeletal manifestations.

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### INTRODUCTION

Gaucher disease (GD), the most common lysosomal storage disorder, results from a deficiency in glucocerebrosidase, leading to the accumulation of "Gaucher cells" in the bone marrow [1,3]. Skeletal involvement is a frequent and often the most debilitating aspect of the disease, reported as highly prevalent in large cohorts and reviews [2,3]. The spectrum of bone manifestations ranges from modeling deformities (Erlenmeyer flask deformity) to irreversible structural failures such as avascular necrosis (AVN) and vertebral collapse [2,3]. In the spine, the "H-shaped" vertebra is a highly characteristic finding and is well described in imaging literature [1,6]. This case report illustrates the classic CT features of advanced skeletal GD in a young adult.

### CASE PRESENTATION

A 25-year-old male, followed for Type 1 Gaucher disease since 2007, was referred for a thoraco-abdomino-pelvic CT evaluation. The patient's surgical history was significant for a splenectomy in 2016 and a total hip arthroplasty (THA) on the left side in 2022 due to advanced osteonecrosis.

#### CT Imaging Findings

Sagittal reconstructions of the thoracolumbar spine demonstrate a generalized biconcave appearance of

the vertebral bodies. There is a characteristic central depression of both the superior and inferior endplates, while the peripheral heights of the vertebrae are relatively preserved, creating the classic "H-shaped" or "wafer" vertebral morphology (Figure 1). These findings are indicative of chronic endplate ischemia and micro-infarctions described in GD and other small-vessel occlusive marrow disorders [2,5].



Figure 1: Sagittal CT reformat of the spine showing multiple H-shaped vertebrae with central endplate depression

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**Figure 2: Coronal CT reformat of the pelvis showing subchondral collapse and sclerosis of the right femoral head (arrow). Left total hip arthroplasty is present**

Coronal reformats show advanced aseptic osteonecrosis of the right femoral head, characterized by subchondral sclerosis, cystic changes, and significant articular surface collapse (Figure 2). The left side shows the previously implanted total hip prosthesis. There is evidence of diffuse osteopenia and cortical thinning, consistent with long-standing medullary expansion by Gaucher cells [2,7].

## DISCUSSION

The skeletal manifestations of Gaucher disease are the result of a complex interplay between marrow infiltration, increased intraosseous pressure, and vascular compromise [2,5]. Marrow replacement by Gaucher cells leads to venous stasis and elevated intraosseous pressure, which can cause focal ischemia and infarction.

The "H-shaped" vertebra (also referred to as biconcave vertebra or Reynold's-type deformity) is a hallmark imaging feature in disorders that preferentially affect central endplate perfusion (e.g., Gaucher disease and sickle cell disease) [6,4]. In GD, accumulation of Gaucher cells in the marrow leads to venous stasis and retrograde pressure affecting nutrient arteries supplying the central vertebral endplate, resulting in central endplate necrosis and the characteristic biconcave depression on sagittal CT images [1,2]. Unlike osteoporotic wedge fractures, the peripheral vertebral margins are often preserved due to collateral periosteal blood supply [1,2].

Aseptic osteonecrosis (AVN) is one of the most severe complications of GD and frequently involves the femoral head [2,3,5]. Pathophysiology includes mechanical compression of intraosseous capillaries by Gaucher cells and subsequent bone infarction. CT is particularly useful for evaluating chronic structural changes such as subchondral collapse, sclerosis, and the crescent sign that guide orthopedic management, though MRI remains the most sensitive modality for early marrow changes and "bone crises" [1,5].

Early hematologic optimization and timely orthopedic intervention (including arthroplasty when collapse is advanced) are guided by the structural map provided by CT and by clinical correlation with MRI when early disease is suspected [1,3,5]. Prior splenectomy and delayed initiation of specific therapy have been associated with higher rates of osteonecrosis and fracture in cohort studies [3,4].

## CONCLUSION

GD is recognized as the most common of the lysosomal storage diseases. Continued research and advances in understanding this disease are fundamental to improving early diagnosis, developing more effective treatments, and providing patients with a better quality of life.

Radiologists must recognize the "H-shaped" vertebral deformity and the CT patterns of osteonecrosis as important signature of Gaucher disease, to provide early diagnosis whenever possible.

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