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Case Report

Multifocal Gorham-Stout disease in a thirteen-year-old boy with horseshoe kidney: A case report ☆

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ABSTRACT

Gorham-Stout disease is a rare condition characterized by the massive osteolysis replaced with vascular or lymphatic proliferation and fibrous tissue. We report the case of a 13-year-old boy complaining of occasional lower back pain. Spinal X-ray showed scoliosis and pelvic asymmetry, CT and MRI revealed multiple osteolysis replaced by soft tissue without osteogenesis in lumbar vertebrae, sacrum, iliac bone, ischium and acetabulum and horseshoe kidney. Laboratory and clinical findings excluded all other potential causes of osteolysis and the patient was diagnosed as Gorham-Stout disease, although no lymphangioma or hemangioma were found in the specimen of right iliac bone. The report shows an unusual, multifocal Gorham-Stout disease in a 13-year-old boy with horseshoe kidney, suggesting that the typical imaging findings and raising awareness of the disease can facilitate timely diagnosis for the disease.

Introduction

Gorham-Stout disease (GSD), also referred to as phantom bone disease, massive osteolysis and vanishing bone disease, is a rare, nonmalignant bone disorder characterized by spontaneous, sometimes progressive, idiopathic osteolysis with replacement by vascular, lymphatic and fibrous tissue proliferation and the lack of new bone formation [1]. Since GSD was first defined in 1955, approximately 400 cases of GSD were reported in the literature up till to 2023 [2,3]. GSD may start at any age and develop in any part of the skeletal system in the human body, such as maxillofacial skeleton, spine and pelvis, its clinical manifestations vary and depend on the bone that has been involved, including pain, swelling, functional impairment or a pathological fracture in the affected part [1,3]. Usually, the GSD is not accompanied by any systemic symptoms and the function of the involved part frequently remains good at the early stage [1], fatal complications, such as neurological defects, chylothorax and respiratory failure, may develop in the patients affecting the spine at the late stage, resulting in high morbidity and mortality rate [4,5]. Although the prognosis of the patient with GSD is unpredictable, timely treatment by pharmacological methods can control the disease

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progression [6]. However, diagnosing GSD is challenging and requires ruling out other potential causes according to laboratory, histological and radiographic findings, and delayed medical treatments ultimately lead to a poor prognosis [2]. Therefore, it is important for physicians to raise awareness of this disease and to diagnose timely in the early stages of the GSD.

According to the classification proposed by Hardegger F, et al, GSD belongs to a unicentric osteolysis with a single osteolytic focus affecting one bone or contiguous bones around one focus, not respecting joint boundaries [7]. However, the multifocal, noncontiguous osteolysis was found in GSD patients [8], which is extremely rare. Here, we present the case report of a 13-year-old boy with multicentric Gorham-Stout disease and horseshoe kidney, diagnosed by laboratory and radiographic findings.

Case presentation

A 13-year-old boy complained of occasional lower back pain. His past medical history was unremarkable, without any known trauma. No hereditary history was identified. Physical examination revealed scoliosis.

Laboratory tests showed slight elevations in serum creatine kinase 292 U/L (reference range, 26 to 174 U/L) and high-sensitivity C-reactive protein 1.8 mg/L (medium risk's reference < 1 mg/L). Haemogram, blood sugar, serum proteins, blood urea nitrogen, serum creatinine, serum cystatin C, estimated glomerular filtration rate (eGFR), blood uric acid, serum alanine aminotransferase, serum aspartate aminotransferase, serum calcium, alkaline phosphatase, prostate acid phosphatase, erythrocyte sedimentation rate and rheumatoid arthritis factor were all within normal limits. The detection of antibodies against hepatitis B virus, hepatitis C virus, treponema pallidum, human immunodeficiency virus was negative. No abnormalities were found in the routine urinalysis and Bence-Jones protein in urine was negative.

The spinal and pelvic X-ray revealed scoliosis, pelvic asymmetry, and the right ischium deformation (Figs. 1A and B).

The spinal computed tomography (CT) demonstrated multiple patchy low attenuation on the body and appendages of lumbar and sacral vertebrae (Figs. 2A and B). The pelvis CT images revealed osteolysis without new bone formation or periosteal reaction in right ilium, acetabulum and ischium (Figs. 2 C-F).

Magnetic resonance imaging (MRI) showed that the osteolytic lesions in L4, L5 and sacrum were replaced with soft tissue, showing hyperintensity on T1-weighted images (T1WI) and T2-weighted images (T2WI), and hypointensity on T2-weighted fat saturation (T2-FS) images (Figs. 3A-C). The contrast-enhanced MRI demonstrated obvious enhancement in the osteolytic lesions. The synovial membrane in sacroiliac joints was thickened and enhanced homogeneously. In addition, the soft tissue in the right ilium and sacrum enhanced homogeneously (Figs. 3D and E). Histopathology of his iliac tissue revealed a small amount of bone tissue, cartilage tissue, inflammatory cells and the connective tissue (Supplementary Fig. 1). Fusion of lower kidneys indicates horseshoe kidney (Fig. 3F).

Discussion

GSD involving the spine is an extremely rare skeletal disorder characterized by idiopathic massive osteolysis [9]. In 1985, Hardegger et al. proposed a commonly accepted classification with 5 types of idiopathic massive osteolysis and GSD belonged to type IV idiopathic massive osteolysis (Gorham's massive osteolysis) [7]. Among them, GSD is differentiated from other types of idiopathic massive osteolysis by its lack of a hereditary pattern and nephropathy. No family history of genetic or bone disease was identified in our case. In spite of a horseshoe kidney found in our patient who is at higher risk of developing end-stage kidney disease [10], he had no renal dysfunction, evidenced by normal levels of blood urea

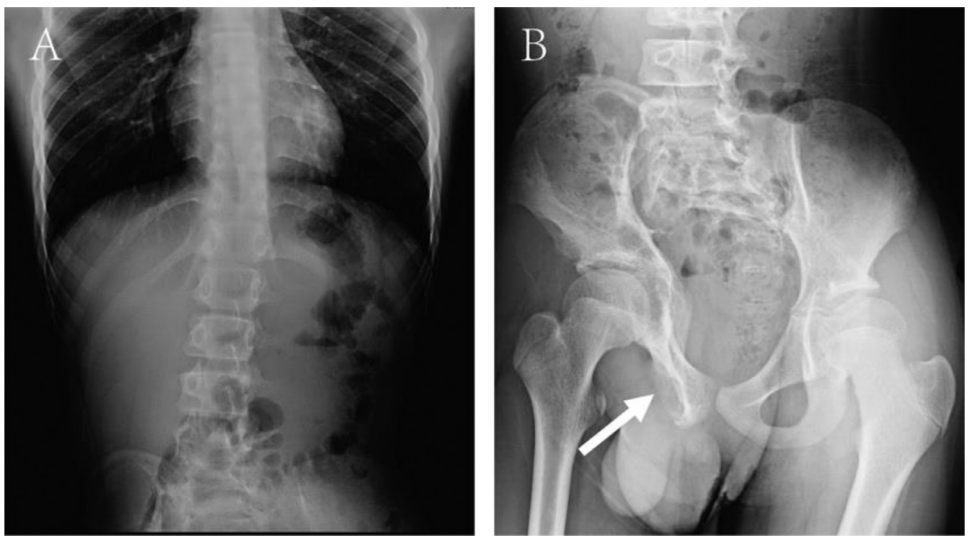

Fig. 1 – X-ray of the spine and pelvis. (A) scoliosis; (B) asymmetric pelvis and the right ischium deformation (white arrow).

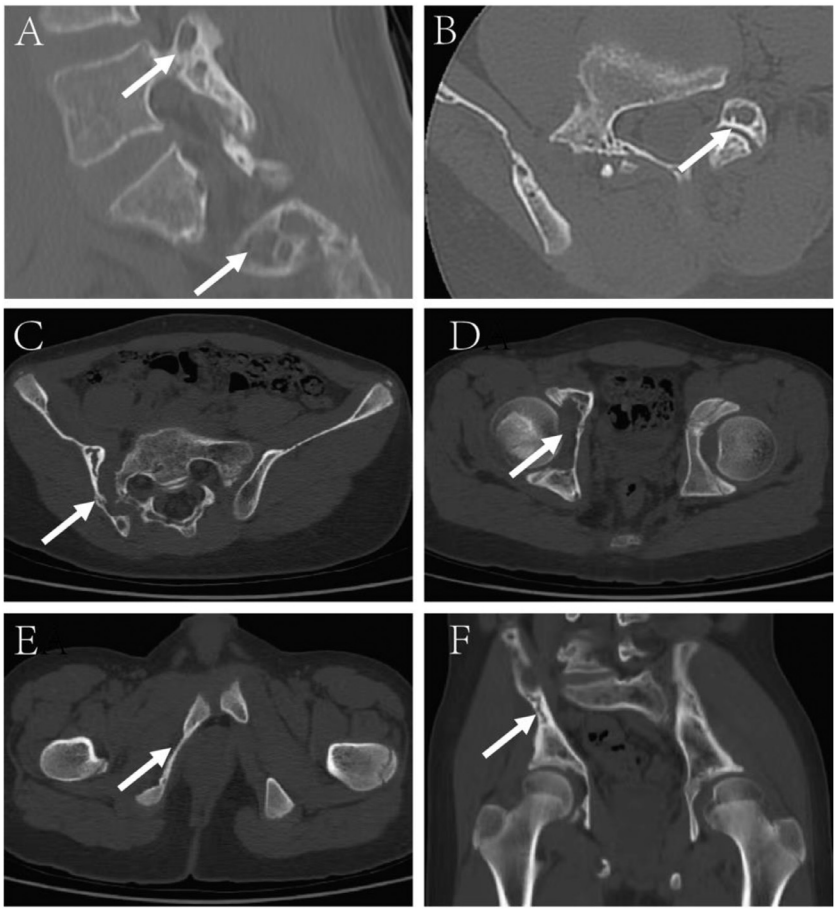

Fig. 2 – CT of the spine and pelvis. (A) Multiple patchy low attenuation of the lumbar and sacral vertebrae, (B) the appendage (white arrows); (C) White arrows indicate the thinning right iliac bone, (D) acetabulum and (E) ischium; (F) Cystic low attenuation of the right iliac bone (white arrow).

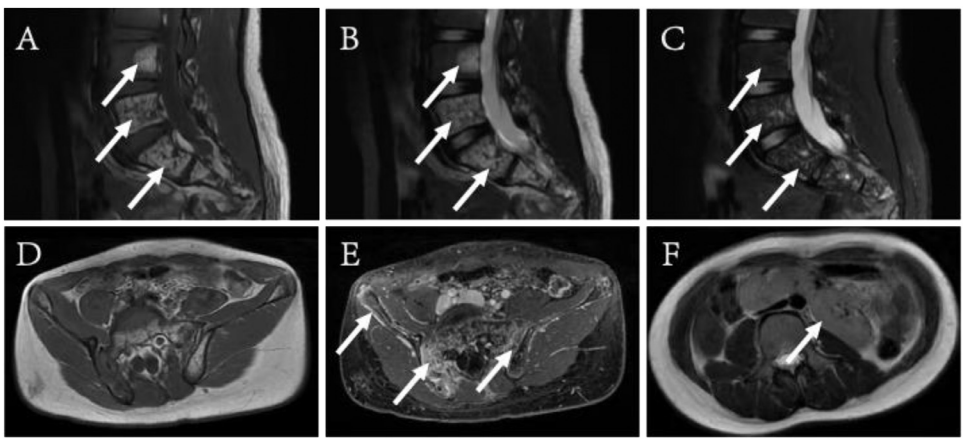

Fig. 3 – MRI of the spine and pelvis. (A) Hyperintensity on sagittal T1-weighted image (T1WI), (B) hyperintensity on sagittal T2-weighted image (T2WI) and (C) hypointensity on sagittal T2-weighted fat saturation (T2-FS) showed the lesions in L4, L5 and sacrum (white arrows); (D) Axial T1-weighted image (T1WI) and (E)the contrast-enhanced MRI demonstrated the osteolytic lesions and the soft tissue in the right ilium and sacrum enhanced obviously and the synovial membrane in sacroiliac joints was thickened and enhanced homogeneously (white arrows); (F) Horseshoe kidney (white arrow).

nitrogen, serum creatinine, serum cystatin C, eGFR and normal urinalysis. Particularly, the spinal CT images of the patient showed multiple patchy osteolysis in the lumbar and sacral vertebrae, the pelvis CT images revealed osteolysis without new bone formation or periosteal reaction in right ilium, acetabulum and ischium and MRI demonstrated that the osteolytic lesions in L4, L5 and sacrum were replaced with soft tissue, all of which are consistent with classic radiological imaging pattern of GSD [3]. Thus, he was highly likely to suffer from GSD even with the negative biopsy findings. As the etiology and precise pathogenesis are unclear, diagnosis of GSD is challenging. Up till now, the diagnosis of GSD is made by the combination of detailed clinical evaluation, radiologic, histopathologic findings and the exclusion of all other potential causes of osteolysis [2,3,11]. In 1983, Heffez L, at al. described the following 8 diagnostic criteria for GSD: (1) a positive biopsy for angiomatous tissue, (2) absence of tumor and cellular atypical features, (3) signs of local progressive osseous resorption, (4) absence of dystrophic calcification and osteoblastic response, (5) nonexpansive and nonulcerative lesion, (6) no visceral involvement, (7) osteolytic radiographic pattern and (8) negative genetic, infectious, immunological, endocrinological, metabolic, neoplastic and rheumatological etiologies, such as osteomyelitis, multiple myeloma, metastatic lytic lesions from the primary tumor and rheumatoid arthritis [12]. In the present case, the patient bad no above-mentioned diseases. There was no primary site of the tumor and infection, and laboratory findings showed no obvious abnormality except for a slight increase in serum creatine kinase and high-sensitivity C-reactive protein. Normal rheumatologic laboratory findings excluded the possibility of rheumatoid arthritis, and negative Bence-Jones protein in urine and pathologic findings ruled out multiple myeloma. Therefore, he was diagnosed as GSD by exclusion of other potential etiologies of osteolysis. However, the histopathological findings did not reveal the existence of lymphangioma or/and hemangioma in the osteolytic lesions. Increasing evidence demonstrated that the pathological features of GSD remained controversial and the proliferation of vascular or lymphatic tissue was not found in the osteolytic lesion of GSD, especially at the early stage of GSD [13]. It was reported that radiological examinations and clinical findings identified the existence of lymphhemangiomatosis at sites around, near or far from the osteolytic lesions [14]. In the present case, On T1WI, osteolytic lesions exhibited hyperintensity with obvious enhancement and the synovial membrane in sacroiliac joints was thickened and enhanced homogeneously, which may indicate the presence of blood vessels or lymphoid tissue. On the other hand, it was demonstrated that the patient with a horseshoe kidney was complicated with renal hemangioma [15]. Our patient suffered from a horseshoe kidney, whether renal hemangioma occurred should be identified.

In addition, edema was found in the right iliac muscle and inflammation was present in the soft tissue of the hips. This maybe explains a slight elevation in serum creatine kinase and high-sensitivity C-reactive protein.

In summary, multifocal, noncontiguous osteolysis without new bone formation was found in an adolescent with horseshoe kidney, clinical and radiographic findings ruled out all other potential causes of osteolysis. Thus, he was diagnosed as GSD, although the lymphangioma or hemangioma tissue was not be found in the osteolytic lesion. Unfortunately, he had developed spinal deformities before GSD was diagnosed. There is a delay in diagnosis of GSD due to the lack of pathologic evidence in many cases, it is very important for physicians to note that raising awareness of this disease can facilitate timely diagnosis and appropriate therapy in the early stage of the disease. CT and MRI as well as regular follow-up play an important role in the early diagnosis of the disease.

Author contributions

Liaoyuan Wang: conceptualization, collecting data, writing original draft. **Xiaokai Mo:** preparing the pictures, resources, review. **Hui Shen:** resources, review. **Shuixing Zhang:** conceptualization, review.

Patient consent

The authors certify that they have obtained the patient consent, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understand that his name will not be published.

Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.radcr.2024.07.170.

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