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Femoral shaft fracture complicating Gaucher disease in adult

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Abstract
Gaucher's disease is a rare disease of progressive evolution characterized by a lysosomal storage disorder. It is rarely responsible for femoral fracture. Patients with Gaucher disease may present with musculoskeletal complications, including osteopenia, medullary bone infarcts, and avascular necrosis. We report a case of femur fracture due to gaucher disease in a 28-year-old woman who underwent two operations for pathological femoral fractures.

Keywords: Gaucher disease, erlenmeyer deformity, splenectomy, Centromedullary nailing

Introduction
Gaucher disease (GD) is a rare disease. It is the most common lysosomal disease characterized by an accumulation of glucocerebroside in the cells of the reticuloendothelial system due to a lack of glucocerebrosidase. Its prevalence is estimated at 1/300,000 to 1/100,000. There are three types of Gaucher disease: type 1 is the most common form and presents with hepatosplenomegaly, anemia, thrombocytopenia and bone pain or fractures without neurological involvement. Type 2 accounts for less than 1% of cases and is the most severe with a neurological problem, so most of these patients die before the age of 2 years [1]. Type 3 is rare, starts in childhood and leads to moderate neurological symptoms. GD is more common in Jews [2]. However, type III is commonly encountered in Asian and Arab countries [3]. In the present case, the patient presented with type 1 Gaucher disease associated with a pathologic fracture of the left femoral bone.

Case presentation
A 28-year-old woman presented to the emergency department with thigh pain and functional impotence of the left lower extremity after falling from her height one hour earlier. In her history, she had a normal physical development. At the age of 12, a diagnosis of GD type I was made, confirmed by a medullary and hepatic histological study following digestive manifestations (abdominal pain, weakness and asymmetric enlargement of the abdomen). At the age of 14, she underwent splenectomy and was then treated symptomatically and never had specific treatment with enzyme therapy. Traumatologically, she had limitations of movement and a history of seven spontaneous fractures of the upper and lower limb bones after minor trauma with a history of centromedullary nail osteosynthesis.
On admission, clinical examination revealed pain of the entire thigh with limitations of movement. Standard radiological examination revealed a fracture of the left femoral bone without displacement, associated with an Erlenmeyer flask deformity. These deformities are clear in the bilateral humerus and tibia but with normal appearance of the vertebral bone and pelvis. On the blood count, she had a hemoglobin of 10.3 g/dl, white blood cells of 32.9 G/dl, and platelets of 231,103/mm3. There was no thrombocytopenia.
In the operating room, the procedure was performed with care to avoid new fractures due to bone fragility by centromedullary nail osteosynthesis. Then, she was treated symptomatically with blood transfusion and never received specific treatment with enzyme therapy, as this type of treatment is not available in our country. And after monitoring the patient's condition for several months, she was able to walk again with a satisfactory range of motion.

Discussion

Gaucher disease is one of the lysosomal storage diseases and is rarely observed. It is an autosomal recessive disease due to a series of mutations in the GBA (Glucosylceramide Beta) gene, located on chromosome 1g21.

This chromosome causes a lack of the enzyme B-glucocerebroside. This results in an excessive accumulation of lipids in the musculoskeletal and reticuloendothelial system [4].

In 80% of patients with GD, deformation of the long bones develops. This explains the severe involvement of the musculoskeletal system [5]. Children may present with symptoms such as growth retardation, osteopenia, lytic lesions, osteonecrosis, medullary cortical bone infarction, pathological fracture [6]. Erlenmeyer flask deformity, which is a significant radiological finding, begins in the prepubertal period [7], as it is a well-known remodeling of the metaphyseal segment of the tubular bone. This sign was observed in our patient at the humerus, tibia and femur. Chronic secondary infarction related to ischemia is the possible mechanism and is unalterable. Damage to the femur and humerus and vertebral bodies due to osteonecrosis induces joint collapse [8].

Organ enlargement, especially splenomegaly and hepatomegaly, is a common feature of this disease. Splenomegaly can cause severe anemia, leukopenia, thrombocytopenia and splenic infarction. The purpose of splenectomy is only to correct the hematological problems, it is used as an alternative. But it has no effect on the progression of the disease [9]. In a series of 35 cases reported by Ida *et al.* [10], it was reported that the incidence of severe bone disease was significantly higher in patients who underwent splenectomy. Patients with Gaucher develop hematomas called "Gaucheromas" in the adjacent muscles. It is important for the surgeon to remember this to avoid these complications. Lebel *et al.* [11] recommend the administration of erythropoietin preoperatively in patients with GD. In our patient, the postoperative course was unremarkable.

Enzyme replacement therapy is an essential treatment in Gaucher disease. Hematological abnormalities and organomegaly may disappear after 1 year of treatment. Bone damage disappears slowly, up to 4 years [12]. In our case, the patient might never have developed skeletal deformity if she had received enzyme replacement therapy at the time of diagnosis. However, as this treatment was not available in our country at that time, she developed these deformities.

Femoral shaft fracture complicating gaucher disease in adult

Case Report

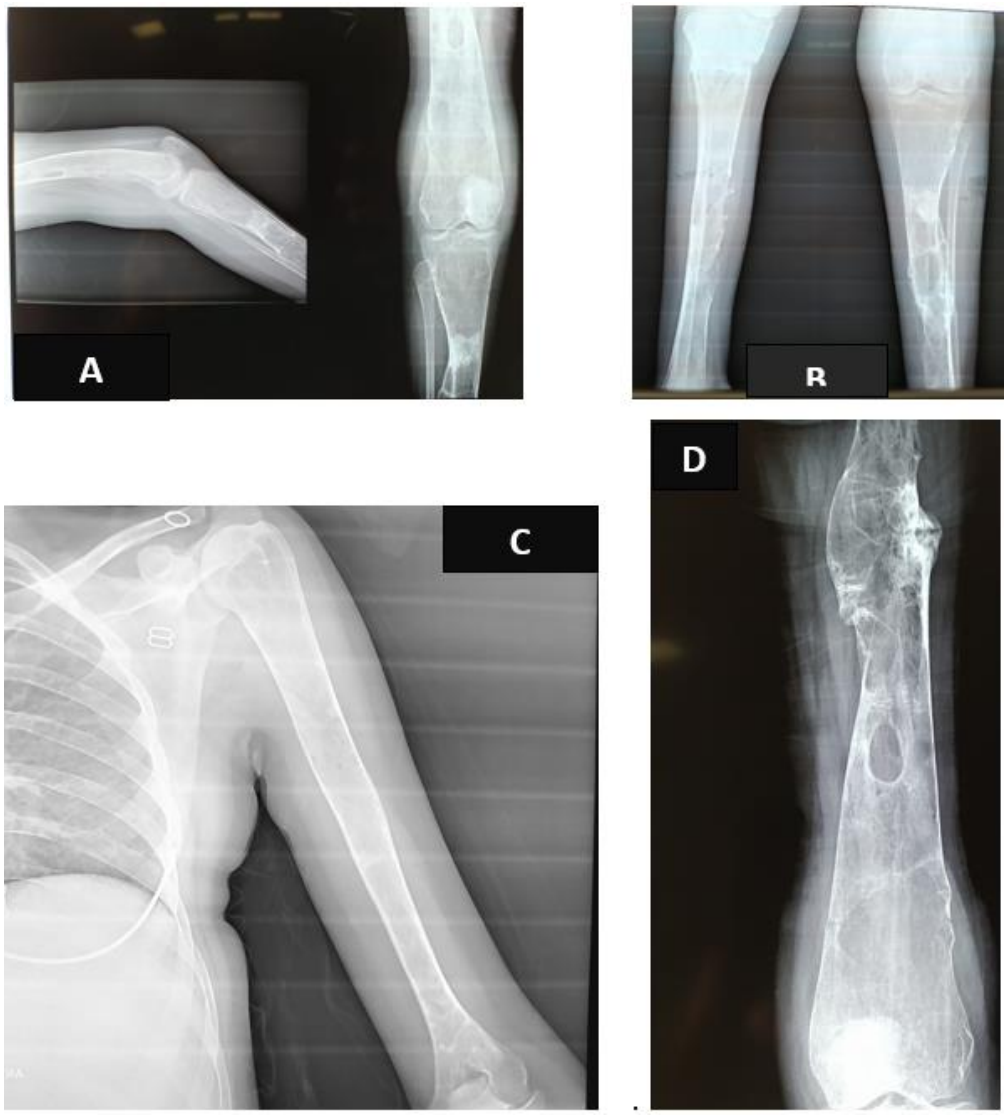

Fig 1 (A, B, C, D): The Erlenmeyer flask deformity at level of tibia-femoral and humerus bones

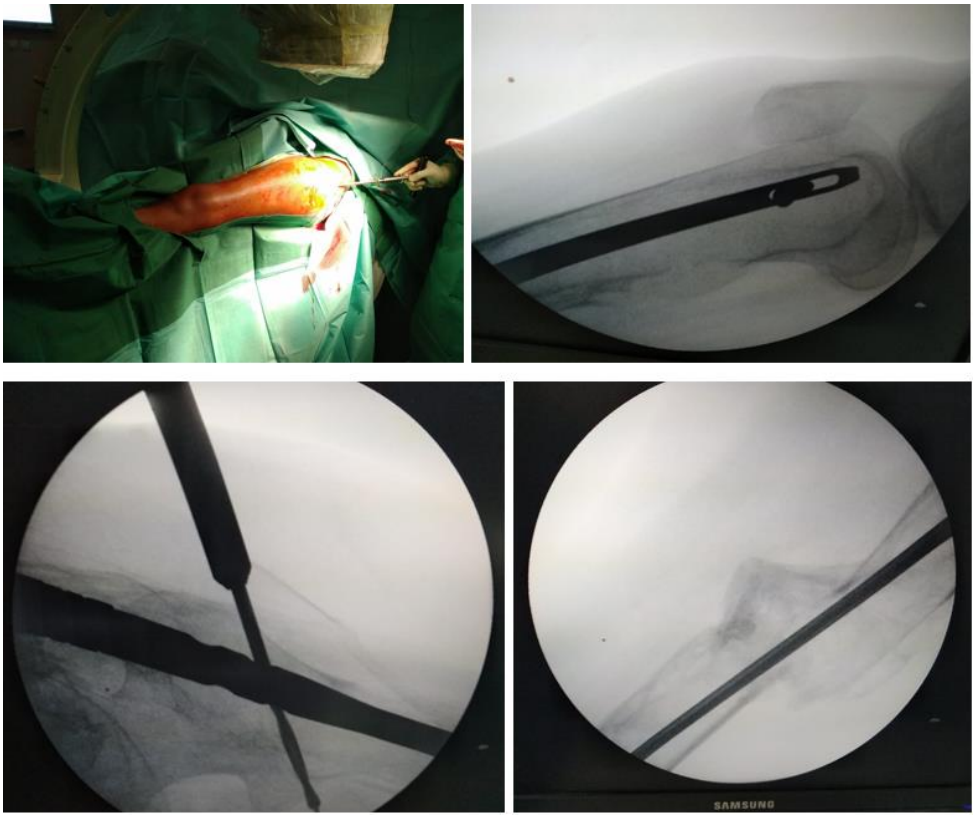

Fig 2: Installation of the patient and during operation pictures

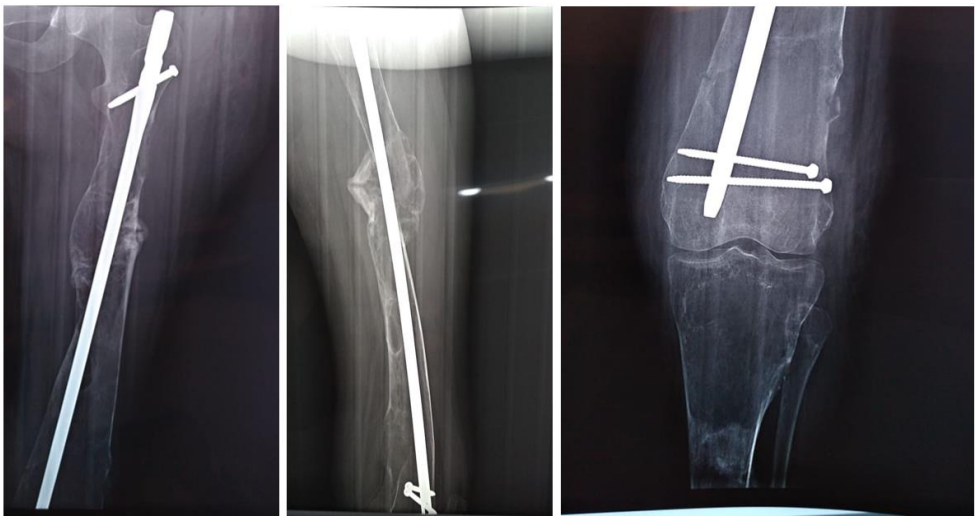

Fig 3: Post operative pictures with centromedullary nailing osteosynthesis.

Conclusion

Among the lysosomal diseases, Gaucher disease is the most common form. It can manifest itself by hematological complications or hepatosplenomegaly. It can lead to bone damage that can affect daily physical activities or even cause pathological fracture. In the present case report, the patient was followed for several years and had joint deformities and pathological fractures with centromedullary nail osteosynthesis on two occasions. The purpose of this case is to educate young orthopedic physicians about the diagnosis and treatment of GD because of its rarity.

Conflict of Interest

Not available

Financial Support

Not available

References

1. Lutsky KF, Tejwani NC. Orthopaedic manifestations of Gaucher disease. Bull NYU Hosp JT Dis 2007;65:37-42.
2. Mehta A. Epidemiology and history of Gaucher's disease. Eur J Intern Med, Nov 2006;(17 Suppl):S2-5.
3. Zimran A. How I tread Gaucher disease. Blood. 2011;118(6):1463-71.
4. Ozcan HN, Kara M, Kara O, *et al*. Severe skeletal involvement in a patient with Gaucher's disease. J Orthop Sci. 2009;14(4):465-468.
5. Rosenbloom BE, Weinreb NJ. Gaucher disease: A comprehensive review. Crit Rev Oncog. 2013;18(3):16-75.
6. Simpson WL, Hermann G, Balwani M. Imaging of Gaucher Disease. World J Radiol. 2014 Sep

28;6(9):657-68.
7. Masi L, Brandi ML. Gaucher disease. The role of the specialist on metabolic bone diseases. Clin Cases Miner Bone Metab. 2015 May-Aug;12(2):165.
8. Stowens DW, Teitelbaum SL, Kaha AJ *et al*. Skeletal complications of Gaucher disease. Medicine (Baltimore). 1985;64(5):310-22.
9. Lachiewicz PF, Gucher's disease. Orthop Clin North Am. 1984;15(4):765-74.
10. Ida H, Rennert OM, Kato S, Ueda T, Uishi K, Maekawa K, *et al*. Severe skeletal complications in Japanaes patients with type 1 Gaucher disease. J Inherit Metab Dis. 1999;22(1):63-73.
11. Lebel E, Ioscovich A, Itschaki M, *et al*. Hip artrhroplasty in patients with gaucher disease. Blood Cells Mol Dis. 2011;46(1):60-65.
12. Guggenbuhl P, Grosbois B, Chates G. Gaucher diease. Joint Bone Spine. 2008;75(2):116-124.

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