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18F-FDG PET/CT and Gaucher Disease: A Suggestive Metabolic Signature

Anouar Jaouad[1*], Amine Maach[1], Hicham Eddou[1]

[1]Department of Clinical Hematology, Moulay Ismail Military Hospital, Meknes, Morocco

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***Corresponding author:** Anouar Jaouad
Department of Clinical Hematology, Moulay Ismail Military Hospital, Meknes, Morocco

Abstract **Case Report**

18F-FDG positron emission tomography (FDG-PET) is a metabolic imaging technique used in many pathologies, including Gaucher disease (GD), a rare lysosomal disorder caused by an enzyme deficiency that leads to the accumulation of glucocerebroside in macrophages. This article presents the case of a 55-year-old man with symptoms including bone pain, anemia, and an enlarged spleen. The diagnosis of type 1 Gaucher disease was confirmed by biological and genetic analysis. PET-CT (Positron Emission Tomography-Computed Tomography) showed intense uptake in the long bones associated with signs of osteonecrosis, as well as splenic and hepatic involvement. The discussion highlights the bone manifestations of GD (osteopenia, osteonecrosis, fractures) and the limitations of conventional imaging techniques (MRI, scintigraphy, CT). FDG-PET proves to be an essential tool in disease assessment, helping to identify areas of active infiltration and guide management.

Keywords: PET-CT, Gaucher Disease, Lysosomal Storage Disorder.

INTRODUCTION

18F-fluorodeoxyglucose positron emission tomography (FDG-PET) is a metabolic imaging technique that measures glucose consumption by cancerous or inflammatory cells [1]. Its use dates back to the 1990s and has since gained importance, particularly in oncology [2]. Gaucher disease (GD) is a rare hematologic lysosomal disorder of autosomal recessive inheritance that affects glucocerebrosidase, an enzyme essential for the degradation of sphingolipids [3]. Lysosomes are cellular organelles responsible for degrading intracellular molecules and organelles, as well as extracellular molecules internalized by the cell, through their hydrolytic enzyme content (autophagy and heterophagy) [4]. The purpose of this article is to illustrate the characteristic PET-CT findings of Gaucher disease.

CASE REPORT

A 55-year-old male retired military personnel presented with chronic bone pain in the femurs, tibias, and humerus, described as dull and worsened by exertion. He also reported persistent fatigue, progressive abdominal enlargement with a sensation of fullness, and episodes of nasal bleeding and pallor. His past medical history was unremarkable. Laboratory tests revealed anemia (hemoglobin 9.5 g/dL), thrombocytopenia (platelets 85,000/mm³), and moderately elevated liver enzymes. Bone marrow aspiration revealed Gaucher cells, a beta-glucocerebrosidase activity assay confirmed reduced enzymatic activity, and genetic analysis identified a mutation in the GBA1 gene, confirming the diagnosis of type 1 Gaucher disease.

An 18F-FDG PET scan combined with computed tomography (PET-CT) was performed to assess disease extent.

The PET-CT showed intense uptake in the femurs, tibias, and humerus, corresponding to areas of active Gaucher cell infiltration. In the femurs, uptake was particularly pronounced in the proximal regions, with signs of osteonecrosis and bone infarction. Tibias exhibited diffuse uptake, especially in the metaphyses, suggesting inflammatory damage or active infiltration. The humeri showed moderate uptake in the diaphyses without evidence of fracture or destructive lesions. Additionally, the spleen appeared massively enlarged (22 cm long axis) with heterogeneous uptake, reflecting diffuse infiltration. The liver exhibited moderate hepatomegaly with mild uptake, suggesting less severe involvement. No abnormal uptake was observed in the lungs or other organs. These results confirm the

multisystem involvement of Gaucher disease, with widespread bone (femur, tibia, humerus), spleen, and liver involvement. PET-CT thus plays a key role in diagnosing and managing this rare disease by identifying active lesions and guiding treatment.

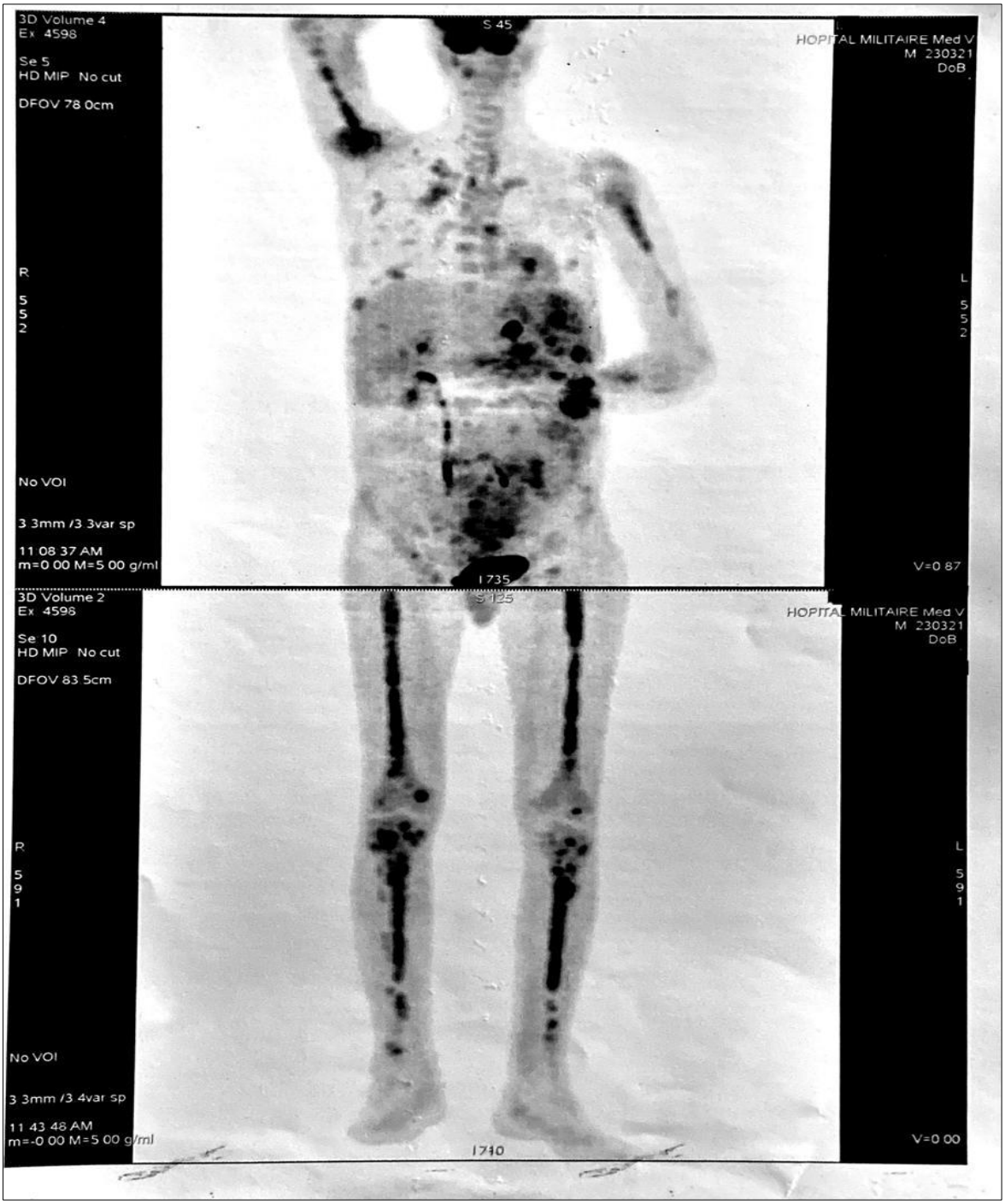

Figure 1: PET-CT scan showing diffuse bone, spleen, and liver hyperfixation in a patient with Gaucher disease.

DISCUSSION

Gaucher disease is a rare genetic lysosomal disorder caused by a deficiency of the enzyme beta-glucocerebrosidase, resulting in the accumulation of glucocerebroside in macrophage lysosomes. The skeletal manifestations of this disease are among the most disabling.

Multiple bone abnormalities: The Erlenmeyer flask deformity, characterized by enlargement of the distal femoral metaphysis, is a commonly observed bone abnormality. Osteopenia and osteoporosis, leading to generalized bone density reduction, are detectable by dual-energy X-ray absorptiometry (DXA). Osteosclerosis and lytic lesions manifest as bone density abnormalities visible on radiography. Avascular

necrosis, resulting from blood supply interruption, leads to progressive bone tissue destruction and is usually detected by MRI. Bone marrow infiltration, visible on MRI as T1 and T2 hypointensity, results from bone marrow fat depletion. Finally, pathologic fractures, promoted by Gaucher cell infiltration, weaken bones and increase the risk of orthopedic complications [5, 6].

Imaging limitations in Gaucher disease vary depending on the technique used:

- MRI is the preferred method for assessing bone marrow involvement in Gaucher disease. However, conventional MRI lacks quantitative capabilities, limiting its ability to accurately measure disease burden. Nevertheless, advanced techniques such as quantitative Dixon MRI can provide a more precise evaluation [7].
- Scintigraphy with 99mTc-sestamibi can identify bone marrow infiltration by Gaucher cells but lacks specificity and may be influenced by other hematologic disorders [8].
- Ultrasound and computed tomography (CT) can detect focal lesions in the liver and spleen but cannot reliably differentiate them from other pathologies such as hepatocellular carcinoma [9].
- Conventional radiography is limited to detecting advanced skeletal manifestations and is not useful for early-stage disease assessment [8].

The role of positron emission tomography (PET) in Gaucher disease primarily relates to the assessment of tissue distribution and the pharmacokinetics of enzyme replacement therapy (ERT) [10]. While PET is not routinely used for this purpose, ongoing research explores its potential in evaluating treatment response. Additionally, PET has been used to study the neurobiological aspects of Gaucher disease, particularly in patients with GBA mutations, a known risk factor for Parkinson's disease. Studies have utilized this technique to assess dopamine synthesis and regional cerebral blood flow in patients with Gaucher-related parkinsonism, providing valuable insights into the pathophysiology of these linked disorders [11, 12].

In summary, PET is a valuable tool for assessing bone and visceral involvement in Gaucher disease, enabling precise visualization of metabolic abnormalities in affected tissues.

CONCLUSION

18F-fluorodeoxyglucose (FDG) positron emission tomography (PET) combined with computed tomography (PET-CT) is an essential functional imaging tool in hematology, particularly for evaluating and monitoring complex diseases such as Gaucher disease. This technique enables visualization of metabolic abnormalities, identification of active lesions, and guidance of therapeutic decisions. In Gaucher disease, a rare lysosomal pathology characterized by glucosylceramide accumulation in macrophages, PET-CT reveals bone lesions (uptake in femurs, tibias, humeri), visceral lesions (splenomegaly, hepatomegaly), and excludes other pathologies. Although expensive and requiring expertise, PET-CT improves patient management by providing an accurate, non-invasive assessment. It remains a valuable tool for diagnosis, follow-up, and treatment optimization in this rare disease.

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