Case Report

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Gaucher Disease - From Textbook Obscurity to Clinical Reality: A Case Report with Diagnostic and Therapeutic Implications at Moi Teaching and Referral Hospital

Isaac Osore Ogola[*1], **Alice Gichemi**[1,2,3], **Maureen Mumbua Kiio**[1], **Wendy Matendechele**[1], **Gilbert Olbara**[1,2], **Festus Muigai**[1,2]

1. Department of Pediatric Hematology and Oncology, Moi Teaching and Referral Hospital, Eldoret, Kenya.
2. Department of Child Health and Pediatrics, Moi University, Eldoret, Kenya.
3. Oncology Fellowship Program, Moi University/AMPATH, Eldoret, Kenya.

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Running Title Gaucher Disease - From Textbook Obscurity to Clinical Reality

ABSTRACT

Gaucher's disease is a rare lysosomal storage disorder caused by a deficiency of the enzyme β-glucocerebrosidase, leading to progressive accumulation of glucocerebroside within macrophages. Clinical presentations vary widely, often resulting in misdiagnosis, particularly in resource-limited settings where advanced diagnostics are unavailable. A 1-year-old female presented to Moi Teaching and Referral Hospital (MTRH), Kenya, with a 2-month history of progressive, painless abdominal swelling, without fever, vomiting, jaundice, or weight loss. She had been misdiagnosed with sickle cell disease and treated with hydroxyurea, without improvement. Examination revealed pallor, massive splenomegaly (20 cm), and hepatomegaly (4 cm), with no lymphadenopathy or bone deformities. Laboratory tests showed anemia, thrombocytopenia, and elevated LDH. Bone marrow biopsy revealed Gaucher cells, confirming Type 1 GD. The patient was managed supportively with transfusions, nutritional support, and infection prevention, and referred for enzyme assays, enzyme replacement therapy, and follow-up. This case highlights the diagnostic challenges in low-resource settings, where advanced enzyme assays and molecular testing are often unavailable, and treatment options such as enzyme replacement therapy (ERT) remain inaccessible. It emphasizes the need for heightened clinical suspicion, improved diagnostic infrastructure, and advocacy for affordable therapeutic access in sub-Saharan Africa.

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*** Corresponding Author:**
Isaac Osore Ogola
Address: *Department of Pediatric Hematology and Oncology AMPATH - Moi Teaching and Referral Hospital P.O. Box 3-30100, Eldoret, Kenya.*
E-mail: *osoreisaac@gmail.com*

Introduction

Gaucher disease is the most prevalent lysosomal storage disorder, with an estimated global incidence of 1 in 40,000 to 1 in 60,000 live births [1]. The condition arises from pathogenic variants in the *GBA* gene, leading to a deficiency of glucocerebrosidase and subsequent accumulation of glucosylceramide within macrophages, termed Gaucher cells [2]. These cells infiltrate multiple organs, producing clinical features including hepatosplenomegaly, cytopenias, bone involvement, and, in neuronopathic forms, neurological impairment [3,4]. Three clinical subtypes are recognized: type 1 (non-neuronopathic), type 2 (acute neuronopathic), and type 3 (chronic neuronopathic), with type 1 being the most common worldwide [5]. Though widely reported in Ashkenazi Jewish populations, Gaucher disease is underdiagnosed in Africa due to low awareness, lack of diagnostic facilities, and clinical overlap with more common pediatric conditions such as malaria, leukemia, and sickle cell disease [6,7]. Early and accurate diagnosis is crucial, as timely initiation of disease-specific therapies such as enzyme replacement therapy (ERT) or substrate reduction therapy (SRT) can substantially modify disease progression and improve quality of life [8,9]. However, in resource-limited settings, diagnosis is often delayed or missed, and therapeutic options remain inaccessible [10].

We report a case of a one-year-old child presenting with abdominal distension and massive splenomegaly, initially suspected to have a hematologic malignancy but later diagnosed with Gaucher disease type 1. This case illustrates the diagnostic pitfalls and management barriers encountered in sub-Saharan Africa.

Case Presentation

A one-year-old female presented to Moi Teaching and Referral Hospital (MTRH), Kenya, with progressive abdominal distension over four months, accompanied by intermittent low-grade fevers, poor weight gain, and easy fatigability. There was no history of recurrent infections, bleeding tendencies, or developmental delay. The child was an only child of non-consanguineous parents, with no significant family medical history. Antenatal and perinatal periods were uneventful. Examination revealed a pale, underweight child with gross abdominal distension. The liver was palpable 6 cm below the costal margin, and the spleen extended to the right iliac fossa, firm and non-tender. There was no jaundice, lymphadenopathy, or dysmorphic features. Anthropometric measurements indicated growth faltering. Laboratory investigations showed hemoglobin of 7.4 g/dL, leukocytes at 3.1×10^9/L, and platelets at 82×10^9/L, consistent with pancytopenia. Peripheral blood film revealed normocytic normochromic anemia with occasional tear-drop cells but no blasts. Liver function tests and renal profile were within normal limits.

Differential diagnoses at this stage included acute leukemia, lymphoma, chronic hemolytic anemia (such as sickle cell disease), storage disorders, and hypersplenism from infectious causes. Malaria rapid diagnostic test and HIV serology were negative. Chest radiograph and abdominal ultrasound confirmed massive hepatosplenomegaly without focal lesions or lymphadenopathy. Bone marrow aspiration revealed large macrophages with abundant fibrillary cytoplasm resembling "wrinkled tissue paper," characteristic of Gaucher cells. No malignant blasts were identified. A diagnosis of Gaucher disease type 1 was made. Unfortunately, confirmatory glucocerebrosidase enzyme activity assay and *GBA* gene sequencing were unavailable locally. Lysosomal storage disorder panel testing could not be accessed due to financial constraints. The family received counseling regarding the condition, prognosis, and therapeutic limitations. The child was managed with supportive care, including blood transfusions, nutritional support, and monitoring for infectious complications, and was referred to Kenyatta National Hospital—a national referral facility better equipped in both human and infrastructural resources. The aim of the referral was to further solidify the diagnosis through enzyme assays and access ERT.

Discussion

This case underscores the diagnostic complexity of Gaucher disease in low-resource settings, where its rarity and non-specific manifestations frequently lead to misdiagnosis. The child presented with splenomegaly, cytopenias, and growth faltering—features that overlap with more prevalent pediatric conditions such as hematologic malignancies, sickle cell disease, malaria, and chronic infections [1,6]. In our patient, acute leukemia was the leading initial suspicion given the pancytopenia; however, the absence of blasts on peripheral smear and bone marrow examination helped redirect the evaluation.

Splenomegaly in African children is most commonly attributed to malaria, schistosomiasis, or hematologic malignancies [7]. Negative malaria testing and the absence of parasitic exposure history excluded infectious causes. Pancytopenia further supported the possibility of bone marrow infiltration. Lymphoma

and leukemia remained strong considerations until bone marrow aspiration revealed Gaucher cells.

The cytopenias observed in Gaucher disease are multifactorial, arising from hypersplenism and direct marrow infiltration by Gaucher cells [8,9]. This explains the anemia, leukopenia, and thrombocytopenia in our patient, which initially misled clinicians toward marrow failure syndromes. Hepatomegaly, though present, was less pronounced than splenomegaly—consistent with the typical Gaucher phenotype [10].

Definitive diagnosis requires demonstration of deficient glucocerebrosidase activity and/or *GBA* gene mutation analysis [11]. However, such tests remain inaccessible in most African settings due to cost and limitations in laboratory infrastructure [12]. Reliance on bone marrow morphology, while helpful, carries a risk of misclassification, as pseudo-Gaucher cells may appear in hematologic malignancies such as multiple myeloma and chronic myeloid leukemia [13,14]. In our case, the absence of malignant cells strengthened the diagnosis.

Delayed diagnosis is a recurring theme in resource-limited contexts, with studies documenting diagnostic delays of several years in African patients [15,16]. Such delays permit the development of irreversible complications, including bone disease, growth retardation, and increased morbidity. Our case illustrates this gap, as the child underwent multiple investigations and transfusions before a storage disorder was considered.

Enzyme replacement therapy (ERT) has transformed outcomes for Gaucher disease, leading to reversal of cytopenias, reduction of organomegaly, and improved growth and survival [17–19]. Substrate reduction therapy (SRT) is an alternative for selected patients, while bone marrow transplantation remains rarely utilized [20,21].

Despite these therapeutic advances, access in sub-Saharan Africa remains negligible. ERT is prohibitively expensive, with annual costs exceeding $200,000, and is not covered under public health insurance schemes [22]. In our case, the child could only receive supportive care, consisting of transfusions, nutritional rehabilitation, and infection prevention. Splenectomy, once used to control hypersplenism, is now reserved for refractory cases where ERT is unavailable, though it carries risks of overwhelming sepsis and progressive skeletal disease [23,24].

This case emphasizes systemic gaps in rare disease management in Africa: limited diagnostic infrastructure, lack of awareness among clinicians, and absence of sustainable therapeutic access. Similar cases have been reported in Kenya, Uganda, and South Africa, all highlighting misdiagnosis and lack of therapy [15,16,25]. A coordinated approach integrating clinician education, regional diagnostic hubs, and advocacy for subsidized therapies is urgently needed.

Conclusion

This case of Gaucher disease in a Kenyan infant, initially misdiagnosed as a hematologic malignancy, illustrates the diagnostic and therapeutic barriers encountered in resource-limited settings. Clinical vigilance is essential when evaluating children with massive splenomegaly and cytopenias that are unresponsive to standard treatments. Strengthening laboratory diagnostic capacity, subsidizing access to enzyme replacement and substrate reduction therapies, and establishing national rare disease registries would significantly improve patient outcomes. Lessons from this case reinforce the importance of early clinical suspicion, comprehensive differential diagnosis, and systemic reforms to address inequities in rare disease care across sub-Saharan Africa.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

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Conflict of Interests

The authors have no conflict of interest to declare.

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