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Pediatric Gaucher Disease with Secondary Malnutrition (Kwashiorkor): Case Report

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Abstract

Gaucher Disease (GD) is a rare lysosomal storage disorder caused by mutations in the GBA gene. It results in glucocerebrosidase deficiency, leading to the accumulation of glucocerebrosides in macrophages, causing systemic manifestations such as splenomegaly, anemia and growth retardation. This case report describes a 1-year and 5-month-old female diagnosed with GD, whose condition was further complicated by protein-energy malnutrition (Kwashiorkor). This report highlights the challenges of managing GD in resource-limited settings and emphasizes the need for a multidisciplinary approach.

Keywords: Gaucher disease; Pediatric malnutrition; Kwashiorkor; Enzyme replacement therapy; Genetic disorders; Resource-limited settings

Introduction

Gaucher Disease (GD) is the most common lysosomal storage disorder, caused by biallelic mutations in the GBA gene that lead to deficient activity of the enzyme glucocerebrosidase. This deficiency results in the accumulation of glucocerebrosides in macrophages, which disrupts normal cellular function and leads to systemic manifestations such as hepatosplenomegaly, anemia and developmental delays [1,2]. Malnutrition, specifically Kwashiorkor, exacerbates the disease's clinical course due to its impact on immune and metabolic functions [3,4]. This report describes a pediatric patient with GD complicated by Kwashiorkor, underscoring the importance of early diagnosis and nutritional management.

Case Presentation

- **Age:** 1 year and 5 months
- **Sex:** Female

Chief complaint

Generalized body swelling for one month before admission.

History of present Illness

The patient, a known case of gaucher disease diagnosed four months ago, presented with progressive generalized edema that began in the lower limbs and extended to involve the entire body over the course of a month. Symptoms were associated with decreased appetite, reduced activity and intermittent low-grade fever relieved by antipyretics. There was no respiratory distress, gastrointestinal symptoms or urinary abnormalities.

Prenatal and birth history

- **Maternal health:** Regular prenatal follow-ups and adequate nutrition reported
- **Delivery:** Full-term spontaneous vaginal delivery without complications
- **Birth weight:** Appropriate for gestational age

Nutritional history

The patient was breastfed on demand and received formula milk until six months of age, after which she was introduced to diluted cow's milk. Recently, she was intermittently given dried milk, which may have

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contributed to the development of Kwashiorkor [4].

Developmental history

- **Motor skills:** Delayed, patient could take steps but often fell
- **Language:** Limited to basic words such as "dada" and "mama"

Family history

No documented cases of gaucher disease in the family, though two cousins exhibited similar symptoms but lacked formal diagnoses. Both parents are healthy, with no known genetic or autoimmune disorders.

Physical examination

Vital signs

- **Temperature:** 98.6°F (37°C)
- **Heart rate:** 120 bpm
- **Respiratory rate:** 30 breaths/min
- **Oxygen saturation:** 98% on room air
- **Weight:** 7.58 kg (below the 3rd percentile)
- **Height:** 68 cm

- **General appearance:** Mild pallor, signs of malnutrition and generalized edema
- **Abdominal exam:** Palpable spleen 5 cm below the left costal margin; liver size normal
- **Neurologic exam:** Age-appropriate gross motor function, no focal deficits

Laboratory and imaging findings

- **Ultrasound:** Enlarged spleen (12.8 cm); normal liver size
- **Bone marrow aspiration:** Gaucher-like cells present [2]
- **Complete Blood Count (CBC):** Mild anemia (Hb 10.4 g/dL), elevated white blood cell count (20.3 × 10^3/mm^3)
- **C-Reactive Protein (CRP):** Positive, indicating inflammation
- **Albumin:** Low (2.8 g/dL), consistent with severe malnutrition (Kwashiorkor) [3]

Diagnosis

The patient was diagnosed with gaucher disease (confirmed *via* bone marrow findings) complicated by severe malnutrition (Kwashiorkor).

Discussion

Gaucher disease results from deficient glucocerebrosidase activity, causing lipid accumulation in macrophages and systemic manifestations such as organomegaly and anemia [1]. Malnutrition, specifically Kwashiorkor, exacerbates these symptoms by impairing metabolic and immune functions, leading to hypoalbuminemia and edema [3].

The co-occurrence of gaucher disease and malnutrition presents diagnostic and therapeutic challenges. While Enzyme Replacement Therapy (ERT) is the gold standard for GD treatment, its unavailability in resource-limited settings necessitates supportive care, including nutritional rehabilitation [5].

In this case, the patient's nutritional status was addressed with high-protein dietary supplementation and close monitoring. Multidisciplinary care, including hematology and dietitian consultations, was essential to improve her condition.

Conclusion

This case highlights the complex interplay between gaucher disease and Kwashiorkor, emphasizing the need for early diagnosis, nutritional support and multidisciplinary care. Addressing malnutrition in children with chronic conditions is critical to improving their quality of life and overall outcomes.

Consent Statement

Informed consent was obtained from the patient's parents for the publication of this case report.

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