Haemolytic Anemia In A 4-Year-Old Boy Due To Glucose-6-Phosphate Dehydrogenase Deficiency: A Case Report

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Abstract

A 4-year-old boy of presented with fatigue, pallor, and jaundice after ingesting fava beans, leading to a diagnosis of haemolytic anemia due to glucose-6-phosphate dehydrogenase (G6PD) deficiency. This case illustrates the significance of recognizing dietary triggers in the management of G6PD deficiency, emphasizing the importance of prompt and accurate diagnosis and the critical role of dietary management. The findings confirm the necessity of educational interventions for families at risk and highlight the need for heightened clinical vigilance and preventive care in children with G6PD deficiency. This report adds valuable insight into the management and clinical course of pediatric G6PD deficiency, contributing to a deeper understanding of the condition and its triggers.

Keywords: Glucose-6-Phosphate Dehydrogenase Deficiency, Hemolytic Anemia, Oxidative haemolysis.

INTRODUCTION

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common enzyme deficiency worldwide, affecting over 400 million people globally.[1] It is an X-linked recessive genetic disorder that primarily affects red blood cells, leading to episodes of haemolytic anemia triggered by specific stressors such as infections, certain foods, and drugs.[2]

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The condition is highly prevalent in populations from Africa, the Mediterranean, Middle-East, and Southeast Asia due to a protective effect against malaria.[3]

The pathophysiology of G6PD deficiency involves reduced activity of the glucose-6-phosphate dehydrogenase enzyme, which is crucial for protecting red blood cells from oxidative damage. Without adequate enzyme activity, red blood cells break down prematurely under oxidative stress, leading to haemolytic anemia. This condition can range from asymptomatic to life-threatening, depending on the severity of the enzyme deficiency and the nature of the precipitating cause.[4]

In pediatric cases, G6PD deficiency can manifest with jaundice, pallor, and significant fatigue during haemolytic episodes. Diagnosis is often made following an acute haemolytic event and is confirmed by demonstrating reduced G6PD enzyme activity in red blood cells. It is critical to identify the condition early in children to manage potential triggers and prevent haemolysis.[5]

CASE REPORT

A 4-year-old boy, previously healthy, who presented to the emergency department with symptoms of severe fatigue, pallor, and jaundice. His parents reported that he had a recent history of consuming fava beans, which was unusual in his diet. On examination, he was found to have scleral icterus and splenomegaly.

Initial laboratory investigations showed hemoglobin of 6.8 g/dL, an increased reticulocyte count of 15%, and elevated indirect bilirubin, indicating an ongoing haemolytic process. A peripheral blood smear demonstrated the presence of bite cells and blister cells, typical of oxidative hemolysis. Given the clinical presentation and the hematological findings, an acute haemolytic episode induced by G6PD deficiency was suspected.

Further diagnostic testing included a quantitative G6PD assay, which confirmed a significantly reduced enzyme activity level, consistent with G6PD deficiency. Based on these findings, the child was diagnosed with haemolytic anemia

secondary to G6PD deficiency, triggered by the ingestion of fava beans.

The management involved supportive care, including hydration and monitoring for worsening anemia or hyperbilirubinemia. The child was transfused with one unit of packed red blood cells and was started on folic acid supplementation. Education was provided to the parents regarding the avoidance of known triggering substances, including certain drugs and foods like fava beans.

Laboratory Test	Result	Normal Range
Haemoglobin (g/dL)	6.8	11.5 - 13.5
Reticulocyte Count (%)	15	0.5 - 1.5
Indirect Bilirubin (mg/dL)	2.4	0.2 - 0.8
Direct Bilirubin (mg/dL)	0.3	0 - 0.3
Total Bilirubin (mg/dL)	2.7	0.3 - 1.0
LDH (U/L)	480	120 - 246
Haptoglobin (mg/dL)	<10	30 - 200
G6PD Assay (U/g Hb)	1.2	4.6 - 13.5
Peripheral Smear Findings	Bite cells, Blister cells	N/A

Table 1:- Lab investigation in studied case.

DISCUSSION

Glucose-6-phosphate dehydrogenase (G6PD) deficiency, a genetic disorder characterized by the absence or reduced activity of G6PD enzyme, predisposes affected individuals to haemolytic anemia under stressors such as infections, certain medications, or foods. The case of a 4-year-old boy diagnosed with G6PD deficiency exemplifies the typical clinical presentation and complications associated with this condition, underscoring the need for awareness and prompt diagnosis.[6]

The pathophysiology of G6PD deficiency involves the vulnerability of red blood cells to oxidative damage due to the lack of G6PD enzyme, which is crucial for maintaining the integrity of red blood

cell membranes. This case mirrors findings from previous studies, such as the one by Cappellini and Fiorelli, which provides an extensive review of the enzymatic defect and its hematological implications.[7] Similarly, a case report by Richardson SR et al. discusses the typical oxidative stressors and their avoidance in the management of G6PD deficiency, highlighting the importance of education for the patient's family.[8]

Further analysis of our case compared to other documented instances reveals a common challenge in the early recognition of G6PD deficiency, especially in settings without neonatal screening. A similar case discussed by Frank et al. demonstrates the severe consequences of unrecognized G6PD deficiency in early childhood, where patients experienced significant haemolytic episodes before a definitive diagnosis was established. These reports emphasize the necessity for early and accurate diagnosis through neonatal screening and the potential role of genetic counseling for families at risk.[9]

This case also brings to light the essential aspect of managing G6PD deficiency, primarily through the avoidance of known oxidizing agents and prompt treatment of infections. The importance of this management strategy is supported by a study by Nannelli C et al , where they outline the global distribution of the gene variants and suggest tailored community-based approaches for managing the disease in high-prevalence areas.[10]

CONCLUSION

This case of G6PD deficiency-induced haemolytic anemia in a pediatric patient following dietary exposure highlights the importance of awareness and education about this genetic disorder. Early diagnosis and management are crucial to prevent severe complications and ensure the well-being of affected individuals.

Conflict of interest

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