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G6Pd deficiency with severe hemolytic anemia: a case report

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Abstract

A 3-year-old boy presented to our atoll hospital in H.A Alif Dhidhoo, with severe pallor, jaundice, easy fatigability and recurrent episodes of passage of dark-colored urine for past 3 days. He was born mature at 39 weeks of gestation with no past significant medical history. Recent history revealed the consumption of 2 cans of fava beans and application of some medicinal herbs. On admission, physical examination revealed fever of 101 degree Fahrenheit, severe pallor, jaundice, cervical lymphadenopathy and mild hepatomegaly. Laboratory investigation results showed a hemoglobin level of 5.4 g/dl with a hemolytic blood picture and serum Bilirubin of 6mg/dl. The patient's G6PD level was measured which showed marked deficiency. Other causes of hemolytic anemia were excluded. Patient required urgent packed RBC transfusion and antibiotics for infection. He responded well to the treatment and was discharged in a stable condition. Parents were appropriately advised on the condition and the importance of avoiding certain foods and medication. Folic acid was prescribed for maintaining normal hemoglobin concentration. This is a first case report in North Maldives of G6PD presenting with severe hemolytic anemia requiring blood transfusion.



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Introduction

Glucose-6-phosphate dehydrogenase (G6PD) is an essential enzyme in the hexose monophosphate oxidative pathway. It plays a pivotal role in the production of the cofactor NADPH which brings about the detoxification of oxidative radicals produced in the body. It is considered as the commonest enzyme disorder affecting approximately 400 million people worldwide [1, 2]. In classification by WHO, 5 different types have been mentioned and the most common is Type 2. Usually asymptomatic, however exposure to oxidative stress, they present with severe hemolytic anemia and its consequences, [3] due to severe enzyme deficiency. Such state can be precipitated by certain chemical agents, drugs, certain foods and infections [4-6]. Reports of hemolytic anemia upon exposure to drugs and favism have been published in older children and adults [7]. We are reporting this case of 3 years old boy who had consumed 2 cans of fava beans in the past 48 hours and developed frank hematuria and severe pallor. G-6PD estimation revealed severe deficiency. All other possible causes of hemolytic anemia were excluded. Further investigations revealed features of infection.

Case Report

A 3-year-old boy came to our hospital at North Maldives in HA. Alif Atoll of Dhidhdhoo Island along with parents with the complaints of recurrent episodes of dark-colored urine and severe pallor. He had developed palpitation and easy fatigability and was turning yellow day by day. The above complaints were preceded by fever. Upon history taking mother revealed that he is the most active child of the family and is on family diet. In addition to this he had consumed 2 cans of Fava beans around 3 days back. He has been applying some traditional herbal medicine with pungent smell mixed with henna (lawsonia inermis) leaves for some toe strain. On examination we found him well thriving with a bodyweight of 14.7 kg (50th percentile) and height of 105 cm (90th centile). Positive findings included fever (101Degree Fahrenheit), Heart rate of 120b/min, severe pallor, jaundice, hepatomegaly of 3.5 cm below the right costal margin. The spleen was not palpable. Lungs were clear and precordial examination revealed no cardiomegaly.

Complete blood count showed a hemoglobin level of 5.4 g/dl. Total white blood cell counts were raised (17000/cumm). Differential count and platelet count was within normal limits. The peripheral blood film showed a hemolytic picture. Hemoglobin analysis excluded other causes of hemolysis. G6PD screening by fluorescent spot test showed G6PD deficiency. Urinalysis shows 2-3 red blood cells per high power

field. Serum electrolytes were normal. Serum LDH was marginally raised and Serum Bilirubin was 6mg/dl and other liver enzymes were within normal limits. Hence, it was concluded that G-6PD deficiency is the cause of acute hemolysis caused due to high oxidative stress provoked by consumption of fava beans and application of herbal medicine. Patient was managed by urgent blood transfusion of packed Red blood cells and broad spectrum antibiotic for infection. Anti-pyretic and anti-emetic medication was given symptomatically. Patient responded well to the treatment. Follow-up investigations showed improved results with increasing HB levels, decreasing levels of serum bilirubin and Urine examination showed no RBC's along with visible improvement of high colored urine to normal straw color. Patient was discharged in a stable condition with advice of daily folic acid intake. Family was counselled and advised on G6PD deficiency and the importance of the patient taking folic acid regularly. At subsequent follow-ups, patient was noted to have fewer episodes of pallor and discolored urine.

Discussion

In 1958 a very crucial discovery was made where primaquine induced hemolytic anemia was observed in patients with G6PD deficiency [8]. Patient with G6PD deficiency usually presents with chronic hemolysis, however episodes of acute hemolysis are also seen when oxidative stress is more. Pallor and jaundice varies between mild and severe depending upon the amount of hemolysis and in few instances transfusion dependent hemolysis resembling thalassemia major is known to occur [9]. Majority of affected individuals gave history of neonatal hyperbilirubinemia often requiring exchange transfusion [10-12]. The patients also commonly have a history of hemolysis induced by drugs and/or infection or consumption of contraindicated foods and herbs [13]. Hepato-splenomegaly is usually present but not essential for diagnosis [14,15]. Compensatory mechanisms play role in balancing the HB concentration normal, however, severe oxidative stress leads to dramatic fall of hemoglobin level [16]. Studies have shown that both intravascular and extravascular hemolysis occur evidenced by chromium labelled studies on RBC's [17].

Our patient presented with all the clinical features mentioned above and to our support, there was a history of consumption of fava beans in abundance and application of herbal medicine which are both contraindicated in G6PD patients. This case highlights the importance of history taking and examination and subsequent relation between history and clinical findings, further supported by investigation. Differentially we thought of other causes of hemolytic anemia and acute lymphoblastic leukemia which were excluded with the help of meticulous investigation. Counselling of parents about the pros and cons of the disease and importance of avoiding certain foods, drugs and herbs were explained with importance. Folic acid plays a very important role in ameliorating the problem of anemia and it was stressed upon to continue.

Conclusion

G6PD deficiency is a rare but manageable disease. It requires strict avoidance of certain foods and drugs which can trigger a life threatening episode of hemolysis requiring urgent intervention and transfusion. History plays a pivotal role in diagnosis and avoids bothersome referrals and invasive investigations.

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