A Case of Concurrent Sickle Cell Trait, Alpha Thalassemia, and G6PD Deficiency in a Pediatric Patient

Vinay Krupadev¹, Joshua Kirbens², Amina Rafique³

¹Tulane University Medical Center, Internal Medicine and Pediatrics (PGY3), New Orleans, Louisiana, United States; ²Department of Medicine, Tulane University School of Medicine (MS4), New Orleans, Louisiana, United States; ³Department of Hematology/Oncology, Tulane University School of Medicine and Children’s Hospital of New Orleans, New Orleans, Louisiana, United States

ABSTRACT

Objective: The aim of this study is to highlight the hospital course of a pediatric patient with concurrent sickle cell trait, alpha thalassemia, and G6PD deficiency.

Methods: The patient’s direct bilirubin remained less than 0.2 mg/dl throughout his hospitalization but his total bilirubin peaked at 18.7 mg/dl at 84 hours of life. While the patient’s bilirubin levels decreased after this, the decline was not as rapid as anticipated so a peripheral smear was performed which showed spherocytosis. Hemoglobin electrophoresis was also conducted just prior to discharge after the patient’s hyperbilirubinemia had resolved.

Results: The results of the patient’s hemoglobin electrophoresis revealed that the patient was a sickle cell trait carrier and also showed evidence of trace Hb Barts consistent with alpha thalassemia. In addition, given the patient’s peripheral smear showing spherocytosis, G6PD levels were also assessed and found to be low consistent with mild to moderate G6PD deficiency. The patient’s family was educated about precautions to take to reduce the risk of excessive oxidative stress that could precipitate acute hemolytic anemia episodes in the future.

Conclusion: The concurrent presentation of sickle cell trait, alpha thalassemia, and G6PD deficiency is rare and it is theorized that each trait respectively confers an evolutionary advantage against malaria.

Keywords: Sickle cell trait; Alpha thalassemia; G6PD deficiency; Hemoglobin electrophoresis

DESCRIPTION

Sickle Cell Trait (SCT) is an inherited blood disorder. Alpha thalassemia is a blood disorder that reduces the production of haemoglobin. G6PD deficiency is a genetic disorder that most often affects males. It happens when the body doesn’t have enough of an enzyme called glucose-6-phosphate dehydrogenase (G6PD). G6PD helps red blood cells work. It also protects them from substances in the blood that could harm them.

An estimated 3 million people in the U.S. have sickle cell trait but the exact prevalences of alpha thalassemia and glucose-6-phosphate dehydrogenase (G6PD) deficiency in the U.S. are not well known [1]. An estimated 10% of black males in the U.S. are thought to have G6PD deficiency which is an X-linked recessive disease more common in Africa, Asia, and the Middle East [2]. An estimated 5% of the world’s population is thought to carry a variant of the alpha thalassemia trait and prevalence in the U.S. is thought to be increasing in recent years [3]. While reports of sickle cell trait co-presenting with either alpha thalassemia or G6PD deficiency have been noted in previous studies, documented cases of all three inherited blood disorders presenting together are exceedingly rare.

CASE STUDY

A 4 months old boy born full-term was referred to the hematology clinic following persistent neonatal hyperbilirubinemia during the first week of life. His mother is a sickle cell trait carrier but had an otherwise unremarkable pregnancy course and received routine prenatal care. The baby was first admitted to the hospital after being found to have a total bilirubin level of 16.8 mg/dl at 76 hours of life. Because the infant had been breastfeeding and stooling appropriately for his age, he was initially thought to have either breast feeding or breast milk jaundice and admitted for phototherapy.

The patient’s direct bilirubin remained less than 0.2 mg/dl throughout his hospitalization but his total bilirubin peaked at 18.7 mg/dl at 84 hours of life. While the patient’s bilirubin levels decreased after this, the decline was not as rapid as anticipated so
a peripheral smear was performed which showed spherocytosis. Haemoglobin electrophoresis was also conducted just prior to discharge after the patient’s hyperbilirubinemia had resolved.

RESULTS
The results of the patient’s haemoglobin electrophoresis revealed that the patient was a sickle cell trait carrier and also showed evidence of trace Hb Barts consistent with alpha thalassemia. In addition, given the patient’s peripheral smear showing spherocytosis, G6PD levels were also assessed and found to be low consistent with mild to moderate G6PD deficiency. The patient’s family was educated about precautions to take to reduce the risk of excessive oxidative stress that could precipitate acute haemolytic anaemia episodes in the future.

Glucose-6-Phosphate-Dehydrogenase (G6PD) levels are shown in Table 1 and Haemoglobin Electrophoresis results are shown in Table 2. Overview of Hemoglobinopathies is shown in Figure 1.

Table 1: Glucose-6-Phosphate-Dehydrogenase (G6PD) levels.

<table>
<thead>
<tr>
<th>Type</th>
<th>Units</th>
</tr>
</thead>
<tbody>
<tr>
<td>G6PD</td>
<td>60 units/trillion RBCs</td>
</tr>
</tbody>
</table>

Table 2: Haemoglobin electrophoresis results.

<table>
<thead>
<tr>
<th>Hemoglobin type</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>A2</td>
<td>0%</td>
</tr>
<tr>
<td>F</td>
<td>79.3%</td>
</tr>
<tr>
<td>A</td>
<td>14.2%</td>
</tr>
<tr>
<td>S</td>
<td>6.5%</td>
</tr>
<tr>
<td>Hb Barts</td>
<td>Trace</td>
</tr>
</tbody>
</table>

CONCLUSION
The concurrent presentation of sickle cell trait, alpha thalassemia, and G6PD deficiency is rare and it is theorized that each trait respectively confers an evolutionary advantage against malaria. While the exact prevalence of alpha thalassemia and G6PD deficiency in the U.S is not well documented, each genotype often co-presents with sickle cell trait individually and should be considered in patients with persistent neonatal hyperbilirubinemia secondary to acute haemolytic anaemia.

REFERENCES


