Sickle Cell Trait as a Cause of Splenic Infarction While Climbing Mt. Fuji

Hiraku Funakoshi, Toshihiko Takada, Masahito Miyahara, Tomoko Tsukamoto, Kazutaka Noda, Yoshiyuki Ohira and Masatomi Ikusaka

Abstract

We report a 38-year-old mestizo man with the sudden onset of left upper abdominal pain while climbing Mt. Fuji, which is the highest mountain in Japan. Enhanced computed tomography showed splenic infarction. Although his peripheral blood smear was normal, a hemoglobin S level of 40% established the diagnosis of sickle cell trait (SCT). This trait is common worldwide, but is not well recognized by doctors in Japan because no Japanese patients with SCT have been reported. However, in Japan it is important to consider SCT when assessing foreign patients with splenic infarction.

Key words: sickle cell trait, splenic infarction, high altitude


Introduction

Splenectomy at high altitude is one of the characteristic problems that occur in patients with sickle cell trait (SCT), usually during travel to mountainous areas. Here we report a patient from Honduras who suffered from splenic infarction while climbing Mt. Fuji in Japan. He had no knowledge of his SCT, which was diagnosed by genetic testing.

Case Report

A 38-year-old mestizo man had lived in Japan for five years. While climbing Mt. Fuji, he complained of the sudden onset of severe pain in the left upper quadrant when he reached about 3,400 m above sea level. He was subsequently taken to a local hospital, where enhanced computed tomography showed splenic infarction. After about two weeks, he was referred to our hospital for further treatment because he lived in Chiba prefecture. His birth and development had been normal. He was a nonsmoker, did not use illicit drugs, and was not taking any medications. There was no family history of coagulopathy.

He was seen at our department on the 17th day after the onset. He complained of severe left hypochondrial pain exacerbated by breathing, particularly at the end of inspiration. Movement in all directions also worsened the pain. Although his pain had decreased from day 6 after the onset, it was still severe enough to cause difficulty with daily activities. His temperature was 36.0 °C, blood pressure was 114/66 mmHg, and pulse rate was 76/min with a regular rhythm. On examination, his breathing was shallow because his pain was exacerbated by deep respiration. There were no cardiac murmurs and no abnormal respiratory sounds were audible. His abdomen was tender in the left upper quadrant and his spleen was extremely enlarged to 4 cm below the left costal margin and its medial border extended to the mid-abdominal line. All other findings were normal.

Laboratory tests revealed mild anemia (Hb 13.3 g/dL), thrombocytosis (platelet count 882×10^3/μL), and a high serum level of lactate dehydrogenase. The results of a coagulation work-up were normal and his peripheral blood smear was unremarkable (Table 1).

Abdominal ultrasonography showed an enlarged spleen that was partially liquefied and also revealed the presence of a left-sided pleural effusion. Enhanced computed tomography revealed multiple non-enhanced regions in spleen, a finding that was compatible with splenic infarction (Fig. 1).

His peripheral blood smear morphology was normal, but...
Table 1.  Laboratory Profile

<table>
<thead>
<tr>
<th>Complete blood count</th>
<th>Chemistry</th>
</tr>
</thead>
<tbody>
<tr>
<td>WBC (10.3 \times 10^3/\mu L)</td>
<td>Aspartate Transaminase 28 U/L</td>
</tr>
<tr>
<td>Red blood cells (4.63 \times 10^7/\mu L)</td>
<td>Alanine Transaminase 43 U/L</td>
</tr>
<tr>
<td>Hb (13.3 \text{ g/dL})</td>
<td>Lactate Dehydrogenase 561 U/L</td>
</tr>
<tr>
<td>Hematocrit 38.9 %</td>
<td>Alkaline phosphatase 479 U/L</td>
</tr>
<tr>
<td>Platelet count (882 \times 10^3/\mu L)</td>
<td>Gamma-Glutamyltransferase 139 U/L</td>
</tr>
</tbody>
</table>

Figure 1. Enhanced computed tomography was performed on the fifth day after the onset at the local clinic that this patient first visited. It shows an enlarged spleen (arrow) with heterogeneous enhancement and a left-sided pleural effusion.

Discussion

Sickle cell disease is the most common structural hemoglobinopathy, especially occurring in African-Americans. This hemoglobinopathy is a compound heterozygous autosomal codominant trait. Sickle cell disease results from a single base substitution in the gene encoding the human \(\beta\)-globin subunit that changes the sixth amino acid from glutamic acid to valine. The resulting abnormal \(\text{HbS}\) polymerizes when deoxygenated and this leads to stiffening of the erythrocyte membrane that causes the characteristic sickle shape of red cells (1).

The prototype disease, sickle cell anemia, occurs in persons homozygous for \(\text{HbS}\). Most patients with sickle cell anemia suffer from hemolytic anemia. In addition, microinfarction caused by vascular occlusion is sometimes a fatal complication. The diagnosis is established by detecting elongated and crescent-shaped red blood cells on a peripheral blood smear and abnormalities of the complete blood count. In contrast, patients with SCT are usually asymptomatic and not anemic and their peripheral blood smear is normochromic. However, hypoxemia due to strenuous exercise or high altitude causes their red blood cells to become sickle-shaped. Because sickle-shaped red blood cells have less plasticity, thrombosis can lead to organ damage such as splenic infarction or pulmonary infarction.

The present patient did not know about his SCT and had never experienced any problems, although he often flew in airplanes and climbed mountains about 2,500 m high. The pressure in the cabin of an airplane is 0.75 atmospheres, which corresponds to 2,400 m above sea level. The red
blood cells of persons with SCT usually develop a sickle shape at about 3,500 m above sea level in the absence of heavy exercise. This is why he had no symptoms while flying or climbing other mountains, but splenic infarction occurred when he was about 3,400 m above sea level on Mt. Fuji.

Splenic infarction is usually a self-limiting condition. It is often associated with a left-sided pleural effusion, while laboratory tests typically show an increase of LDH and mild anemia. Whether the treatment of splenic infarction in patients with sickle cell disease should be splenectomy or conservative management is still under discussion (2), although many reports have indicated that conservative management can be effective (3, 4). The present patient responded well to conservative therapy and there were no complications. We advised our patient with SCT that he should avoid hypoxic situations such as heavy exercise or high altitude, use oxygen when flying in an airplane, and receive a vaccination for Streptococcus pneumoniae and Haemophilus influenzae infection (5).

The prevalence of SCT is especially high among African-Americans, while it is not seen among Japanese (6). Thus, SCT is not familiar to Japanese doctors, although numerous cases are reported overseas. The present case indicates that in Japan it is necessary to consider the possibility of SCT when examining foreign patients with splenic infarction. Every physician working in or around mountainous areas about 3,000 m above sea level, like Mt. Fuji, should be altered to this issue.

---

**Conclusion**

It is important to consider the possibility of SCT when a foreign patient presents with the acute onset of abdominal pain at a high altitude. We cannot rule out SCT, even when the peripheral blood smear is normal, because only hypoxemia causes the red cells of patients with SCT to become sickle-shaped. Accordingly, when SCT is strongly suspected, hemoglobin electrophoresis should be performed.

**Acknowledgement**

We are grateful to Dr. Keiko Harano, Department of Biochemistry, Kawasaki Medical School, Kurashiki, Japan for hemoglobin analysis.

**References**