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Case Report

Diagnosis of a sickle cell carrier while investigating the cause of neutropenia in a child with short stature receiving growth hormone

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Abstract

Short stature requiring growth hormone treatment is a well-known complication of sickle cell anemia (SCA). Sickle cell carriers can experience a variety of complications similar to SCA; however, short stature has not been documented in sickle cell carriers yet. Diagnosing using a complete blood count in sickle cell carriers is also difficult. Thus, hemoglobin (Hb) electrophoresis is useful in diagnosing these carriers. We present a case in which a sickle cell carrier is identified using Hb electrophoresis while investigating the etiology of neutropenia in a child using growth hormone for short stature.

Key words: sickle cell carriers; short stature; growth hormone

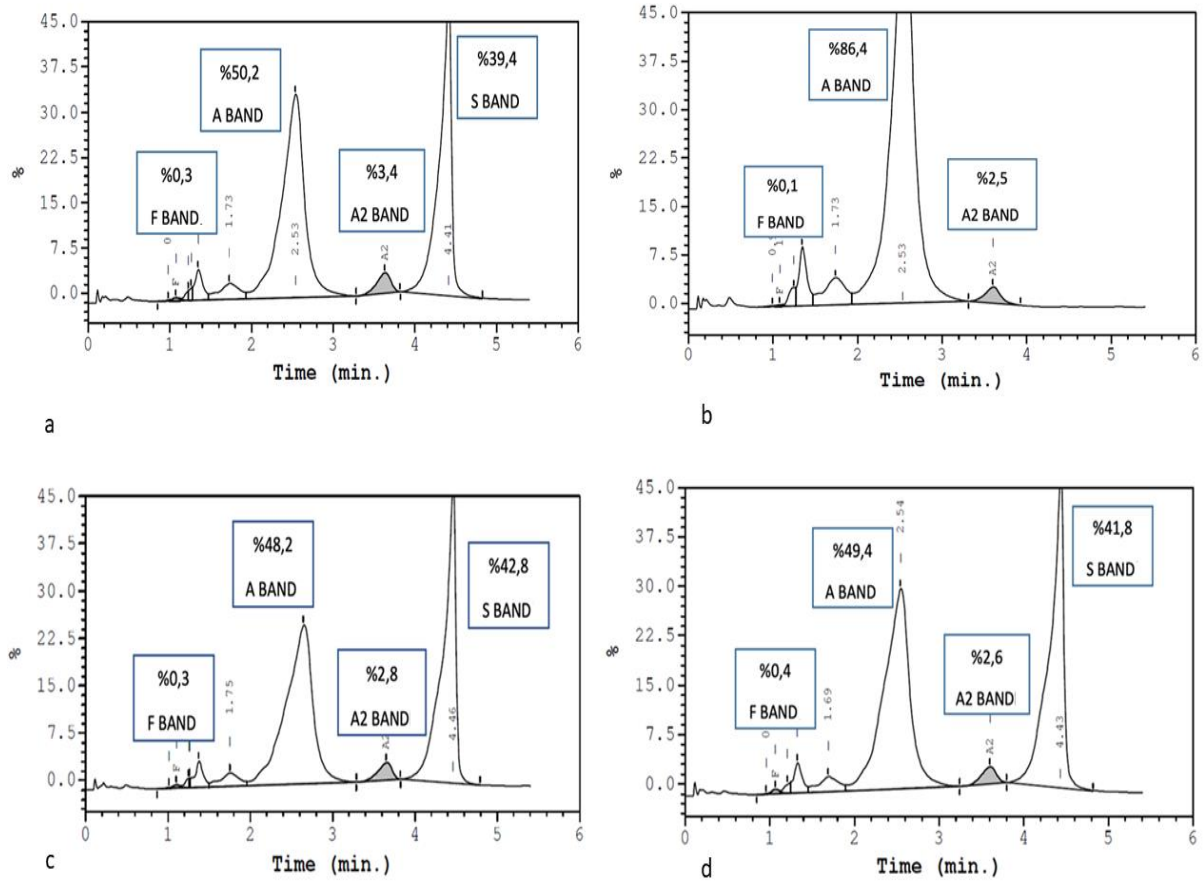
Introduction

Sickle cell trait occurs in around 300 million individuals worldwide [1]. The sickle cell trait affects approximately 13.6 percent of the population in Turkey's Mediterranean region [2]. In contrast to thalassemia carriers, sickle cell carriers can develop clinical symptoms similar to sickle cell anemia (SCA) patients; therefore, it is crucial to identify sickle cell trait individuals [3]. However, utilizing a complete blood count (CBC) to determine a sickle cell carrier is difficult. Hemoglobin (Hb) electrophoresis plays a crucial role in identifying sickle cell carriers. Thus, we present a child who was diagnosed with sickle cell carrier by coincidence while examining the cause of neutropenia and receiving growth hormone (GH) treatment for short stature.

Case Report

A 10-year-old girl was referred to our Hematology department for an investigation into the reasons for neutropenia. Her medical history revealed that she was diagnosed with GH deficiency due to short stature, GH treatment was started at another center three years ago, and neutropenia was found two weeks ago. When she was 7 years old, her height was 107 cm (3P), -3 SDS in specific anthropometric measurements, her annual growth rate was less than 4.5 cm, her bone age was behind according to calendar age, and GH stimulation tests were unresponsive in two different measurements. It was learned that the patient was examined for prolonged jaundice in the neonatal period, her maternal grandmother had gallstones, her father was from Hatay and her mother was from Mersin, but they were not related to each other. Laboratory examination revealed Hb 12.3 g/dL, mean corpuscular volume 80.7 fL, reticulocyte 1.4%, and normal erythrocyte morphology in peripheral smear. Hb electrophoresis revealed HbA 53.4%, HbA2 2.8%, HbS 42.8%, HbF 1%. Her vitamin B12 level (545 pg/mL), ferritin level (33 ng/mL), folic acid level (13 ng/mL), indirect bilirubin level (0.74 mg/dL), and lactate dehydrogenase level (223 IU/mL) were all within the normal range.

Her family members were tested for CBC and Hb electrophoresis. Although CBC values were found to be normal in her father and brother, the HbS band was detected in Hb electrophoresis (Figure 1).



The mother was found to be normal. A pathogenic heterozygous mutation in the HBB gene, c.20A>T(p.E7V) (p.Glu7Val) (HbS), was identified in the patient, her father, and her brother. During the follow-up, the neutropenia improved, and GH continued. It was thought that neutropenia secondary to viral infection developed in the patient Genetic counseling was given for sickle cell carriers.

Discussion

Thalassemia and SCA are both autosomal recessive hemolytic anemias. Patients with thalassemia or SCA require long-term, regular blood transfusions. The complication of transfusional hemosiderosis may develop over time. Advances in iron chelation therapy may prevent this problem. Allogeneic stem cell transplantation and gene therapy show promise as curative therapy for these patients [4]. However, it is essential to avoid the birth of thalassemia or SCA patients. Before marriage, our country's health officials performed a thalassemia trait test based on CBC levels. Although typical CBC may detect most thalassemia carriers early, sickle cell carriers are more difficult to detect with CBC. The gold standard in both carrier groups is Hb electrophoresis using high-performance liquid chromatography [5]. Preimplantation genetic diagnosis could allow for allogeneic stem cell donation from a tissue-typing matching healthy sibling [6]. Short stature has been reported in patients with SCA due to iron accumulation secondary to transfusion, intense inflammation caused by vaso-occlusive crises, and ischemic causes [7]. Although clinical findings similar to those observed in patients with SCA have been reported in sickle cell carriers, short stature requiring GH therapy has not been reported so far.

Our experience suggests that sickle cell trait can be considered in children with short stature who require growth hormones, and a Hb electrophoresis test may be ordered.

Informed Consent: The parents of all participants gave informed consent.

Authorship Contributions Design: Z.K., N.Ö.; Data Collection or Processing: Z.K., N.Ö., G.K., A.B.; Analysis or Interpretation: N.Ö., Z.K.,; Writing: Z.K and N.Ö.

Conflict of Interest: No conflict of interest was declared by the authors.

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