

A Patient with a Previous Diagnosis of Hemoglobin S/C Disease with an Unusually Severe Disease Course

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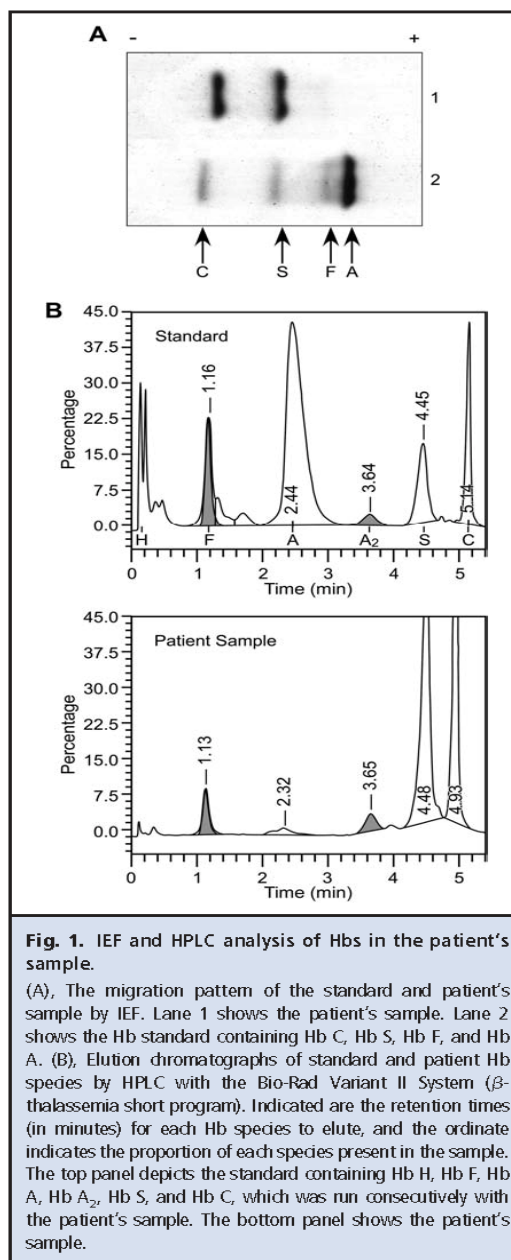
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CASE

A 17-year-old African American male presented to the hematology clinic for treatment of sickle cell disease (SCD). He had received the diagnosis of hemoglobin (Hb) S/C disease at an outside hospital at the age of 6 years; the diagnosis was confirmed in house at 11 years of age. His disease course had been severe, with frequent pain crises of increasing intensity and 2 episodes of acute chest syndrome requiring hospitalization and multiple blood transfusions.

The patient's physical examination was unremarkable: blood pressure, 120/64 mmHg; pulse, 83 beats/min; temperature, 36.9 °C. Laboratory results were as follows: white blood cell count, $11.8 \times 10^9/L$ [reference interval (RI), 3.9 – $10.3 \times 10^9/L$]; Hb, 6.39 mmol/L (RI, 8.68–10.8 mmol/L); packed cell volume, 0.28 (RI, 0.42–0.50); red blood cell count, $3.59 \times 10^{12}/L$ (RI, 4.5 – $6.0 \times 10^{12}/L$); platelet count, $417 \times 10^9/L$ (RI, 135 – $370 \times 10^9/L$); mean corpuscular volume, 78 fL (RI, 83–102 fL); mean corpuscular Hb, 28.7 pg (RI, 27–31 pg); mean corpuscular Hb count, 368 g/L (RI, 320–340 g/L); red cell distribution width, 17.3% (RI, 11.5%–14.5%); and absolute reticulocyte count, 0.115 (RI, 0.02–0.10). A peripheral blood smear showed scattered target and sickle cells, rare nucleated red cells, and mild anisopoikilocytosis. Results for the qualitative sickle cell solubility test were positive. Considering the severe disease course, Hb analysis by HPLC and isoelectric focusing (IEF) was ordered (Fig. 1).



Questions to Consider
• How do various hemoglobinopathies differ clinically?
• What laboratory tests should be used to distinguish different hemoglobinopathies?
• Why is it important to use at least two different Hb separations techniques for the diagnosis of a hemoglobinopathy?
• When should a patient with a diagnosis of Hb S/C be re-evaluated for alternative hemoglobinopathies?
• What treatment strategies are used for patients with different sickle cell diseases? Does this differ from other hemoglobinopathies?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the June 2009 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol55/issue6> and follow the link to the Clinical Case Study and Commentaries.

Educational Centers

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