Young Woman With Abdominal Pain and Anemia

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**History.** During a hospitalization for an asthma exacerbation 2 years earlier, the patient was told she had mild anemia; this had resolved by the time of a follow-up visit months later. During that hospitalization, she was also told that her potassium level was elevated, but it normalized spontaneously. Her only current medication is albuterol, which she uses as needed. Her sister and mother had "some liver problem", about which she does not have further information.

**Physical examination.** The patient appears in moderate distress; she is afebrile. Blood pressure is 100/60 mm Hg; heart rate is 72 beats per minute. Pertinent physical findings include scleral icterus, right upper quadrant tenderness to palpation, and a spleen tip palpable 3 cm below the left costal margin.

**Laboratory studies.** Total bilirubin is 6.4 mg/dL (normal, less than 1.3 mg/dL); direct bilirubin, 2.2 mg/dL (normal, 0 to 0.4 mg/dL); alkaline phosphatase, 278 U/L (normal, 38 to 110 U/L). The hemoglobin level fell overnight from 12.4 g/dL to 9.8 g/dL. Further evaluation of the patient's anemia reveals a mean corpuscular volume of 85 fL, a red blood cell distribution width (RDW) of 24% (normal, 11% to 16%), and a mean corpuscular hemoglobin concentration (MCHC) of 36 g/dL (normal, 31 to 37 g/dL). Additional laboratory findings include a reticulocyte count of 9.9%; serum haptoglobin, 24 mg/dL (normal, 26 to 226 mg/dL); and plasma lactate dehydrogenase, 205 mg/dL (normal, 94 to 200 mg/dL). The result of a Coombs test was negative. A sonogram of the abdomen confirms splenomegaly and reveals gallbladder sludge without cholecystitis. The patient's peripheral blood smear is shown here.

**What diagnosis do the clinical history and peripheral smear suggest?**

A. Malaria  
B. Glucose-6-phosphate dehydrogenase deficiency  
C. Hereditary spherocytosis  
D. Disseminated intravascular coagulation  
E. Thrombotic thrombocytopenic purpura

**A CASE IN POINT**

Hereditary spherocytosis (HS) is a disorder of the red blood cell membrane that results in fragile cells susceptible to hemolysis. In most cases, the defect is thought to be inherited in an autosomal dominant pattern. In mild cases, patients have no or only slight anemia. In severe cases, patients may be transfusion-dependent. HS may also cause hydrops fetalis. Clinical features of HS include anemia, jaundice, and splenomegaly. Bilirubin gallstone formation is a common complication; it affects nearly half of adults with this disorder. The presence of elevated direct bilirubin and alkaline phosphatase levels, accompanied by symptoms of biliary colic, led us to believe that our patient also had gallstone-induced obstructive liver disease. Laboratory findings include a hyperproliferative, normocytic, or microcytic anemia with an elevated MCHC, which reflects loss of red blood cell membrane and cell dehydration.\(^1\) Elevations of RDW are also common. One study found that elevations of MCHC greater than 35 g/dL in combination with RDW greater than 14 fL identified HS with a sensitivity of 64% and a specificity of 100%.\(^2\) Blood smears may show spherocytes that lack central pallor; this reflects membrane loss with cell aging. Inadvertent cooling or a delay in testing of serum samples may result in pseudohyperkalemia, because the cooling causes in vitro hemolysis of the sample and intracellular potassium leaks out. This phenomenon may explain this patient's spurious elevated potassium level during her previous admission. The differential diagnosis of spherocytes on peripheral smear includes HS as well as a wide range of disorders in which the final common pathway is the loss of red blood cell membrane. These disorders include glucose-6-phosphate dehydrogenase deficiency, autoimmune and mechanical hemolytic anemias, and
clostridial sepsis. Most other causes of spherocytosis are associated with a variety of dominant findings on a peripheral smear that point to the diagnosis. The exception is autoimmune hemolytic anemia, which can be distinguished by a positive result on a Coombs test.

**OUTCOME OF THIS CASE** Further questioning revealed that the "liver problem" in the family was recurrent jaundice. The patient's mother and sister had undergone splenectomy for this disorder, which suggested a familial process. A negative Coombs test and a positive result on incubated osmotic fragility testing confirmed the diagnosis of HS. Folate supplementation was initiated to support the patient's rapid cell turnover. She agreed to an outpatient elective cholecystectomy after being counseled about the high likelihood of symptom recurrence. Because her baseline anemia was mild, the patient elected to defer splenectomy.

**References:**


**Links:**

[1] [http://www.physicianspractice.com/authors/amber-shah-md](http://www.physicianspractice.com/authors/amber-shah-md)
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