Netherlands
The Journal of Medicine

PHOTO QUIZ

# A patient with haemolytic anaemia diagnosed after thirty years

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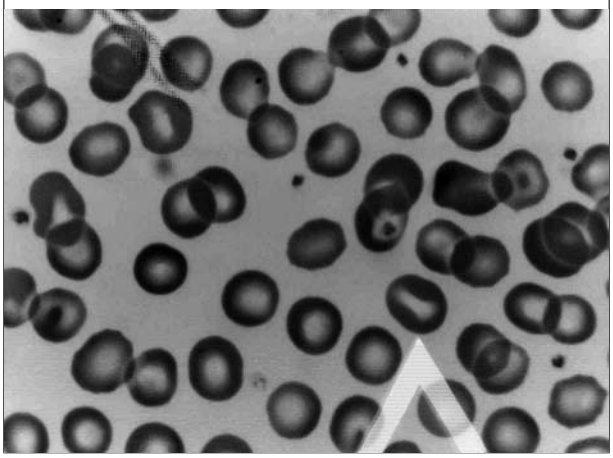
## CASE REPORT

Since 1977, a 37-year-old woman had been treated for mild non-autoimmune haemolytic anaemia (haemoglobin 6.2 mmol/l (normal 7.2 to 9.5)) with a $^{51}$Cr red cell half-life of less than 10 days (24 to 32 days). Mean corpuscular volume was 115 fl (80 to 100), mean cell haemoglobin concentration (MCHC) was 21.9 mmol/l (20.8 to 22.2), the reticulocytes fluctuated between 5.0 and 15.0% (0.3 to 2.0), unconjugated bilirubin was mildly elevated with normal lactate dehydrogenase. Splenectomy failed to restore the anaemia. Since splenectomy was also performed in her father and daughter because of anaemia, a hereditary disorder was suspected. The red cell spectrin content, the α and β haemoglobin chains and the activity of 13 intracellular-erythrocytic enzymes, however, were normal. The osmotic fragility was decreased with 50% lysis at 95.5 mOsmol/kg (127 to 159). On the blood smear (*figure 1*) a sporadic stomatocyte (arrow) was noted.

**Figure 1.** *The blood smear*



## WHAT IS YOUR DIAGNOSIS?

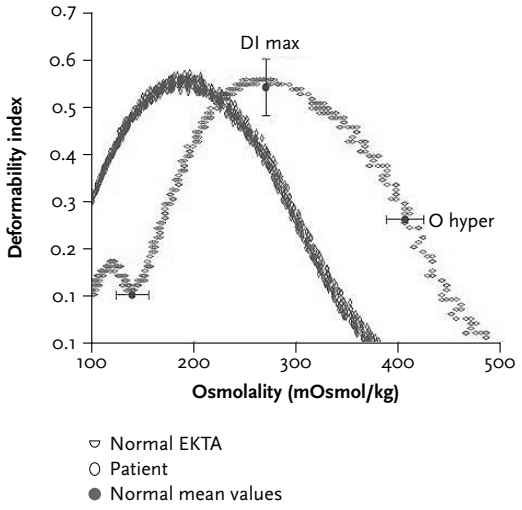
See page 311 for the answer to this photo quiz.

## DIAGNOSIS

The combination of mild familial haemolytic anaemia, macrocytosis, high MCHC and decreased osmotic fragility with sporadic stomatocytes on the (wet) blood smear is suggestive for hereditary dehydrated stomatocytosis (DHSt). In mild cases the morphological features may be overlooked for many years as in this patient.[1-3] The clinical course is usually mild. Splenectomy is not advocated in view of the minimal effects on the anaemia and the high incidence of thrombotic events. Despite the mild anaemia, in this patient the DHSt was complicated by biopsy-proven intrathoracic and abdominal sites of extramedullary haematopoiesis, which is a well-known event in red cell membrane disorders such as spherocytosis.

The pathogenesis of this autosomal disorder is not fully understood and involves a net loss of intracellular potassium content that is not fully compensated by increased sodium influx, leading to a state of dehydration. DHSt may be diagnosed by demonstrating these mild cation shifts within the erythrocytes. In this case DHSt was diagnosed by the characteristic left-shifted curve on ektacytometry with normal deformability index. Ektacytometry examines the deformability of the erythrocytes in various osmotic conditions under constant shear stress.[4] The deformability, reflected by the deformability index (DI), is assessed by optical laser diffraction. As shown in *figure 2*, the deformability of normal erythrocytes is maximal (DI max) at the physiological osmolality of 290 mOsmol/kg. Under these physiological conditions the DI of the patient's erythrocytes is decreased and the normal maximal DI is reached at a lower osmolality of 200 mOsmol/kg. These parameters indicate a dehydrated state of the patient's erythrocytes under physiological osmolality. Ektacytometry may be a diagnostic tool in patients with haemolytic anaemia especially caused by red cell membrane abnormalities.

**Figure 2.** *Ektacytometer osmolality curves of erythrocytes*




**The curve in grey resembles a normal curve. The black curve is the patient's curve. DI max = the maximal deformability index; O hyper = the osmolality at which 1/2 DI max is reached at the right site of the curve.**

## REFERENCES

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4. Johnson RM, Ravindranath Y. Osmotic scan ektacytometry in clinical diagnosis. J Pediatr Hematol Oncol. 1996;18:122-9.