Confirming or Ruling out Hereditary Elliptocytosis

Rummens et al. [1] have published a report on two cases of myelodysplasia with elliptocytosis and schistocytosis. Though the patients’ history evokes an acquired elliptocytosis and the propounded diagnosis of myelodysplasia seems correct, the statement of the authors on p. 177 in the Discussion is not appropriate. Hereditary elliptocytosis (HE) cannot be excluded on the basis of a low percentage of elliptical red cells. We and others have shown that elliptocytes can vary from 10 to 100% in HE. Furthermore, in some rare situations, elliptocytes are totally absent (our case of HE diagnosed in a 25-week-old fetus having an heterozygous spectrin defect of the \( \alpha \), 74,000-dalton type).

Also, the absence of poikilocytes does not constitute a criterion for ruling out HE. Such fragmented cells are always found in the homozygous form of the disease as well as in a variety described in the heterozygous state, the so-called infantile pyknocytosis. Finally, HE should always be confirmed, or ruled out, by analysis of erythrocyte spectrin for qualitative defects and by electrophoresis of the membrane proteins (e.g. \( 4 \) deficiency). Rheology is a second tool for the confirmation of membrane abnormalities. We use Ektacytometry and always obtain abnormal curves in HE. In contrast, deformability was normal in our cases of myelodysplasia. We would be happy to test the erythrocytes of the second patient described by Rummens et al. [1] since poikilocytes and schistocytes are numerous, and some dacryocytes are present, a picture unusual in such pathological cases and suggestive of acquired thalassemia, a rare but already described situation.

Reference