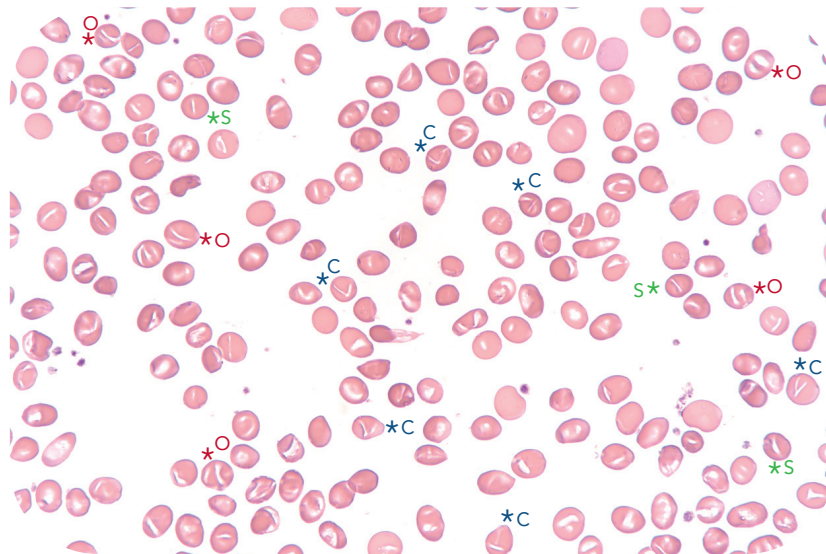


Four genetic red cell disorders in one

Sihem Tarfi and Bouchra Badaoui, Henri Mondor University Hospital



A 21-year-old Comorian woman was seen for counseling for sickle cell trait. Her mother has symptomatic sickle cell trait. On examination, the patient presented with cutaneous and mucosal jaundice. Laboratory evaluation showed moderate anemia (10.5 g/dL), increased total bilirubin (30 μ mol/L), and decreased haptoglobin. The blood smear revealed Melanesian ovalocytes, characteristic of Southeast Asian ovalocytosis (SAO) (original magnification, 63 \times objective; May-Grünwald-Giemsa stain). Representative cells (o), stomatocytes (s), and other poorly defined red blood cell (RBC) shapes (c) are shown, suggesting the presence of a combination of stomatocytes and ovalocytes with an abnormal hemoglobin distribution. The eosin-5-maleimide binding test was positive with a significant

decrease in fluorescence intensity. Molecular analysis revealed the unexpected association of the *HBB* mutation (HbS trait; rs334) with deletion in *SLC4A1* (heterozygous SAO; rs769664228), heterozygous *PIEZO1* mutation (xerocytosis; rs200970763), and heterozygous *SPTA1* variation (elliptocytosis; rs142775522). Ektacytometry showed only the flat profile, characteristic of SAO.

This case highlights the importance of analyzing the blood smear and molecular data in order to make a precise diagnosis. In our patient's case, xerocytosis was diagnosed; thus, splenectomy was contraindicated. This is a complex and unique case of 4 genetic RBC diseases: HbS trait, SAO, xerocytosis, and elliptocytosis.