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Case Report

Erythrocyte deformability and hereditary elliptocytosis

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Hereditary elliptocytosis (HE) is a hereditary membranopathy that comprises a heterogeneous group of diseases, which share irreversible erythrocyte elongation with a prolonged longitudinal diameter in relation to the cross-section (elliptocytes and ovalocytes), which acts as a morphological sign to guide diagnosis. It is characterised by certain molecular alterations to cytoskeleton proteins, such as mutations of alpha-spectrin, beta-spectrin, protein 4.2 and, more rarely, of glycophorin C [3].

This disorder is considered rare, although its incidence is probably underestimated because most cases are pauci- or asymptomatic, and discovery is often fortuitous [8]. It can appear in several clinical forms (asymptomatic, mild, sporadic haemolysis, chronic haemolysis and hereditary pyropoikilocytosis), although most patients do not suffer from anaemia, and haemolysis is poorly marked if and when it occurs. Mild HE is more frequent with few clinical manifestations and with more than 15% of circulating elliptocytes found in peripheral blood [5].

Very few studies have dealt with rheological red blood cell behaviour in patients with HE [1, 2, 4, 6, 7] in which erythrocyte deformability has been determined by ectacytometric techniques.

We report a case of HE in a 37-year-old woman who was admitted to our hospital with clinical manifestations and analytic parameters compatible with marked iron deficiency anaemia in whom, after both treatment with iron therapy and correcting iron deficiency, a high percentage of elliptocytes was found in peripheral blood extension, which is compatible with HE.

The patient came to our centre presenting long-term weakness, hair loss, brittle nails and occasional heartburn. She sometimes noticed her skin took on a yellowish colour. She never had choluric urine, lower back pain nor haemorrhagic symptoms, save heavy periods lasting up to 2 weeks, nor fever, frequent infections or nightly itching.

The haematimetric test showed hypochromic microcytic anaemia: RBC count 4.86×10^6/ μL, Hb 9.9 g/dL, Ht 33.1%, MCV 68 fL, MCH 20.4 pg, MCHC 29.9 g/dL, RDW 17.2%. The iron metabolism test highlighted: iron 21 μg/dL, total iron-binding capacity 420 μg/dL, IS 5% and ferritin 8 ng/mL. The vitamin B12 and folic acid values were within the normal range (474 pg/mL and 10.13 ng/mL, respectively). Presence of thalassaemia trait was ruled out by Hb electrophoresis and molecular biology techniques, as

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Table 1
Haematimetric parameters and erythrocyte elongation index in hereditary elliptocytosis and the control

	Patient	Control
RBC ($\times 10^6$/ μL)	4.66	4.89
Haemoglobin (g/dL)	14.3	15.0
Haematocrit (%)	41.8	43.8
MCV (fL)	89.7	90.0
MHC (pg)	30.65	30.70
MCHC (g/dL)	34.16	34.20
RDW (%)	15.1	12.21
EEI 12 Pa (%)	33.10	44.48
EEI 30 Pa (%)	41.18	50.39
EEI 60 Pa (%)	46.36	53.40

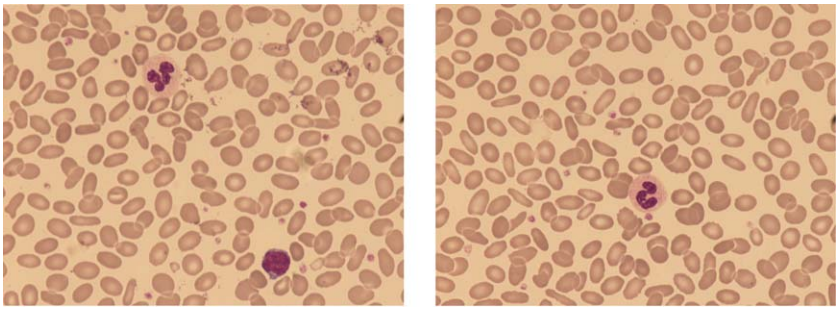

Fig. 1. Blood smears of elliptocytosis.

was presence of autoimmune haemolitic anaemia (haptoglobin 57%, free Hb 8.95%, reticulocytes 1.3% and negative Coombs test). Haemosiderinuria was not observed.

The patient was given iron treatment, 105 mg of ferrous sulphate (Ferro-gradumet®-Teofarma) for 3 months, and a check-up showed that her iron metabolism and haematimetric parameters had become normal (Table 1). Persistence of elongated or cigar-shaped erythrocytes (25% approximately) on blood smears was noted (Fig. 1), and HE was confirmed by a family-based study.

Erythrocyte deformability was determined by ectacytometry in a Rheodyn SSD [9], which measured the erythrocyte elongation index (EEI) at 12, 30 and 60 Pa. This test was simultaneously performed in the patient and in one control, who was sex- and age-matched (Table 1). The patient presented a very low EEI for the various shear stresses tested when compared with the control, which confirms the results previously obtained by other authors [1, 2, 6]. Interestingly in our case, after correcting the patient's iron-deficiency anaemia and correspondingly normalising the haematimetric indices, ectacytometry was able to detect the less deformable ellyptocytes although the percentage was only 25%.

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