

Case Report

Hereditary elliptocytosis discovered during work-up for infective endocarditis: About a case

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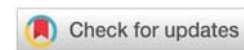
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Introduction

Hereditary elliptocytosis is a group of red blood cell membrane disorders that are characterized by elliptical-shaped erythrocytes and shortened red blood cell survival [1]. It is due to protein abnormalities involving the horizontal skeletal network of the red cell membrane, including the spectrin dimer-dimer interaction or the spectrin-actin-protein 4.1 junction complex [2]. It is often asymptomatic and may be complicated by constitutional hemolytic anemia due to an abnormality in the structure of the Red Blood cell (RB) membrane [2].

We report through this observation a case of elliptocytosis discovered by chance in a patient hospitalized for infective endocarditis and we discuss the clinical, biochemical, and genetic aspects of this condition.

Observation

A 17-year-old patient, from a non-consanguineous marriage, the only daughter of her parents and with no particular pathological history, presented to the emergency room for acute onset dyspnea at rest, with a fever of 38°C. Evolving in the context of the deterioration of the general state. Clinical examination revealed a heart murmur, splenomegaly with mucocutaneous pallor, and failure to thrive without any other associated clinical signs. Blood culture was used to isolate *Streptococcus pneumoniae*. Echocardiography confirmed the diagnosis of infective endocarditis. The patient was hospitalized

in the cardiology department and was treated with a double antibiotic therapy consisting of amoxicillin and ceftriaxone intravenously for 6 weeks. A blood count was performed showing isolated anemia at 7g/dl of normocytic hypochromic hemoglobin. The blood smear stained by the May Grunwald Giemsa (MGG) method revealed anisopoikilocytosis made up of 70% elliptocytes (Figure 1). The hemolysis assessment was normal. Ferritin was low. Membrane protein electrophoresis was not performed due to a lack of resources. The patient

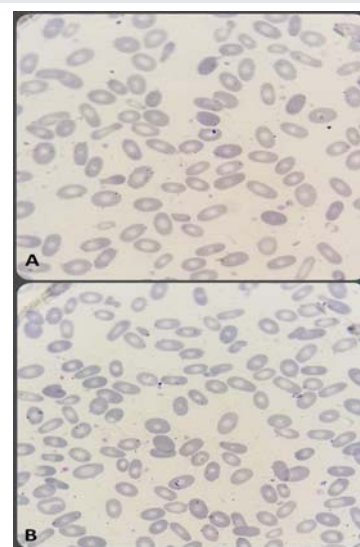


Figure 1: Blood smear showing elliptocytes.

received a blood transfusion and close monitoring. A family investigation based mainly on the blood smear was requested but the patient refused to cooperate.

Discussion

Hereditary Elliptocytosis (HE) is a group of disorders characterized by the presence of elliptical-shaped erythrocytes on peripheral blood film. It has a worldwide distribution but is more common in areas of endemic malaria, particularly in people of African and Mediterranean descent. [1]. The majority of HE-associated defects occur in spectrin, the major structural protein in the skeletal structure of the erythrocyte membrane. Spectrin is composed of two non-identical homologous proteins, alpha, and beta spectrin, encoded by separate genes [2].

HE and its related disorders are characterized by clinical, biochemical, and genetic heterogeneity. Manifestations range from asymptomatic carrier states to severe transfusion-dependent hemolytic anemia. In the healthy carrier, only detailed studies of the thermal stability of the red blood cell membrane as well as a detailed analysis of the protein composition of the cytoskeleton can highlight the anomaly. Mild forms include well-compensated hemolysis, without anemia, but which may be complicated by episodes of acute hemolysis, following a viral infection that stimulates the macrophage monocyte system (infectious mononucleosis, cytomegalovirus, hepatitis) [2]. In the chronic hemolytic form, there is frank hemolysis, anemia with a hemoglobin concentration between 8 and 10g/dl, hyperbilirubinemia, and splenomegaly. A particular entity is a neonatal and infantile poikilocytosis or pyropoikilocytosis. This is a very early presentation, with severe hemolysis responsible for neonatal jaundice often requiring exchange transfusion [1,2]. The reading of the blood smear is an important element of the diagnosis because usually, HE is clinically latent, it is characterized by the presence of more than 30% of elliptocytes [3]. In a normal blood smear and secondary elliptocytosis

associated with deficiency anemia or certain myelodysplastic syndromes, less than 5% of elliptocytes are found [3]. HE is an autosomal dominant disease that most often affects black people. The genetic abnormality is variable. In approximately 70% of cases, it is a mutation located in the SPTA1 gene coding for the alpha chain of spectrin leading to a point of weakness in the mesh constituting the erythrocyte skeleton. The remaining cases (25–30%) result from mutations in the EPB41 gene encoding protein 4.1R, which normally binds to actin and the spectrin beta chain. In very rare cases, the mutation is located on the SPTB gene coding for the spectrin beta chain [4].

Conclusion

HE is an easily diagnosed red blood cell membrane disease. An only careful reading of the blood smear provides a decisive element in the diagnostic process. HE is a very rare pathology whose precise frequency is difficult to estimate given the great clinical heterogeneity and the frequency of asymptomatic forms.

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