IDENTIFICATION OF SEVERAL FAMILIES WITH HEREDITARY SPHEROCYTOSIS IN A POPULATION FROM SOUTH-WESTERN POLAND

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Hereditary spherocytosis (HS) is the most common of the red blood cell (RBC) membrane disorders associated with defects of the erythrocyte membrane skeleton. The red cells present a varying degree of surface area deficiency resulting in spherocytic morphology and increased osmotic and mechanical fragility. Splenic destruction of these abnormal erythrocytes is the primary cause of the haemolysis experienced by HS patients. The primary biochemical defects in HS, either dominant or recessive in nature, reside in the proteins of the erythrocyte membrane, particularly those involved in the vertical interactions between the lipid bilayer and the membrane skeleton: spectrin, ankyrin, band 3 and protein 4.2. We present the results of electrophoretic analyses of red blood cell membranes isolated from 34 patients suffering from HS belonging to several families from south-western Poland. In three families, a defect in ankyrin was detected, and in one family, a band 3 (anion-exchanger protein) defect was discovered. The defects of the red blood cell membranes of the remaining families were not identified. Patients from 3 families with an ankyrin defect were characterised by a decrease in ankyrin 2.1 level, while the members of the single family with a band 3 defect showed a decrease in the content of this protein and a significant increase in electrophoretic mobility. All the changes were statistically significant, as confirmed by the results of the Student T test ($P<0.05$). Our analysis of haemolysis kinetics shows that this property was different from normal for HS cells.