Hereditary dehydrated stomatocytosis with new missense mutations in PIEZO1 through the use of next-generation sequencing panel

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Abstract

Dehydrated hereditary stomatocytosis (DHS) is characterized by alterations of ionic flux with increased cation permeability. Although the clinical presentation of DHS is commonly variable, all of them present hemolysis and anemia, which may range from mild to severe. Like all hemolytic anemias, jaundice, pallor, fatigue, splenomegaly, and gallstones are the key signs. The causative genes have been identified on ABCG5, ABCG8, PIEZO1, SLC2A1, ABCB6, KCNN4, and RHAG. In this case study, we report an 11-year-old male patient who had jaundice, hepatosplenomegaly, and chronic mild congenital non-autoimmune hemolytic anemia. In our patient, a novel homozygous missense mutation in the PIEZO1 gene was detected using a gene-targeted Next-Generation Sequencing panel: c.3364G>A (p.Glu1122Lys), confirming the diagnosis of hereditary dehydrated stomatocytosis. The reason we want to report this case is the detection of a new mutation due to hereditary stomatocytosis, which is a rare disease. Also, due to the limited number of research centers in developing countries and lack of financial resources, it is late to be diagnosed with rare diseases.

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Abbreviations

DHS	Dehydrated hereditary stomatocytosis
NGS	Next-Generation Sequencing
RBC	Red Blood Cell
MCV	Mean Corpuscular Volume
MCH	Mean Corpuscular Hemoglobin
MCHC	Mean Corpuscular Hemoglobin Concentration

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