

Publication

A rare case of coinheritance of Hemoglobin H disease and sickle cell trait combined with severe iron deficiency

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We present a case of a 40-year-old female from Turkey, who was referred to our outpatient clinic for an undetermined thalassemia and sickle cell trait. At first consultation hemoglobin was decreased (71 g/L) with microcytosis (MCV 55.1 fL), and hypochromia (MCHC 239 g/L). The patient had severe iron deficiency. Brilliant cresyl blue staining showed <50% of the erythrocytes with typical Hemoglobin H (HbH) inclusions. High-performance liquid chromatography (HPLC) revealed normal levels of HbA(2) and Hemoglobin F (HbF), and additionally a hemoglobin S (19%). Molecular diagnostics revealed the mutations $\alpha 2$ IVS-I donor site -5nt and a – MED II deletion in the alpha gene complex and confirmed the heterozygote mutation of the beta-gene at codon 6 (HBB:c.20A

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