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Letter to the Editor

Compound heterozygosity for hemoglobin E-Saskatoon and β-thalassemia

Dear Editor,

In our previous paper[1] we discussed hemoglobin (Hb) E-Saskatoon and the combinations with thalassemia or other hemoglobinopathies during pregnancy and genetic counseling in such couples. The second couple referred to has three children. Their second boy was 4 years old when he was found to be a compound heterozygous carrier of β-thalassemia and Hb E-Saskatoon. As already reported, his mother, grandfather and uncle were also found to be carriers of Hb E-Saskatoon. They all come from a village in northern Greece. His father was a heterozygote for β-thalassemia and carried the IVSI-110/N thalassemic mutation. The hematological data of the propositus were: Hb = 10.5 g/dL, hematocrit = 33.4%, red blood count = $5460 \times 10^3/\mu L$, mean cell volume = 61.1 fl, and mean cell hemoglobin = 19.3 pg. The blood films showed hypochromia, microcytosis, anisocytosis and basophilic stippling. Electrophoresis revealed a variant with electrophoretic properties of the Hb E and β-thalassemic trait. Hemoglobin high-pressure liquid chromatography (Variant, Biorad) isolation revealed the presence of Hb F = 1.1%, Hb A = 10.9%, Hb A2 = 5.4% and Hb S = 78.2%. A sickle cell test was negative as expected. DNA studies showed a compound heterozygosity for β-thalassemia (IVSI-110/N) and E-Saskatoon. The propositus is a healthy boy with normal development. In the literature there are few cases of compound heterozygosity for Hb E-Saskatoon and β-thalassemia reported to be asymptomatic.[2,3] Genetic counseling that was provided to the parents was not to undergo prenatal diagnosis due to the mildness of the rare combination previously reported in the literature. This compound heterozygosity is very rare. We were happy to have a mild phenotype as predicted and we believe that communication of such data could be useful for genetic counseling. Detection of thalassemia and structural Hb variants is important in clinical laboratories in countries such as Greece, where there is a high percentage of carriers. Genetic counseling is particularly important in medical genetics because of the often predictive nature of genetic information and the implications for other families.

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