The first case of a compound heterozygosity for Hb E-Saskatoon and Hb S

Hemoglobin E-Saskatoon (b22-Glu->Lys) was first described by Vella et al. in a Canadian woman of mixed Scottish and Dutch origin and ever since the variant has been found in Greece, Scotland, Spain and Turkey. Hb E-Saskatoon seems to be an innocuous variant but there are not any available data concerning the association of hemoglobin E-Saskatoon with other hemoglobinopathies. Few published data exist concerning compound heterozygosity of β-Thalassemia and Hb E-Saskatoon. All cases are mild and with haematological features of a common thalassemia trait.

We report the first case of a compound heterozygosity for HbS and Hb E-Saskatoon in a four-year-old Greek girl. Carriers of hemoglobin E-Saskatoon were also found to be, her mother, brother, grand father and uncle. They all come from a village in Northern Greece.

The hematologic data of the propositus were: Hb=12.8 g/dL, Hct=37.8%, RBC=4980x10^3/mL, MCV=75.8 fl, MCH=25.7 pg. The blood films showed hypochromia, microcytosis, anisocytosis. Electrophoresis revealed a variant with electrophoretic properties of Hb E and Hb S. The sickle test was positive. Hemoglobin HPLC (Variant, Biorad) (Figure 1) isolation revealed the presence of Hb F, Hb A, Hb A2, Hb S (45.7%) and of an unknown variant (42.6%) eluting before S.

DNA studies that included gene amplification using the polymerase chain reaction (PCR), DGGE electrophoresis, DNA sequencing (Figure 2) and ASO hybridization showed a compound heterozygosity for Hb S and E-Saskatoon.

The propositus is a healthy girl with normal development, without pains and until now asymptomatic. The mildness of the condition was evident in the hemoglobin E-Saskatoon heterozygotes of the same family. In the literature there are few cases of compound heterozygosity for hemoglobin S and E reported to be symptomatic. This compound heterozygosity is the first description in Greece and in literature.

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References