The first case of a compound heterozygosity for hb e-saskatoon and hb s

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Hemoglobin E-Saskatoon (b22-Glu->Lys) was first described by Vella *et al.*[1] in a Canadian woman of mixed Scotish and Dutch origin and ever since the variant has been found in Greece, Scotland, Spain and Turkey.[2-6] Hb E-Saskatoon seems to be an innocuous variant but there are not any available data concerning the association of haemoglobin E-Saskatoon with other haemoglobinopathies. Few published data exist concerning compound heterozygosity of b-Thalassemia and Hb E-Saskatoon.[4-7] 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 haemoglobin 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.

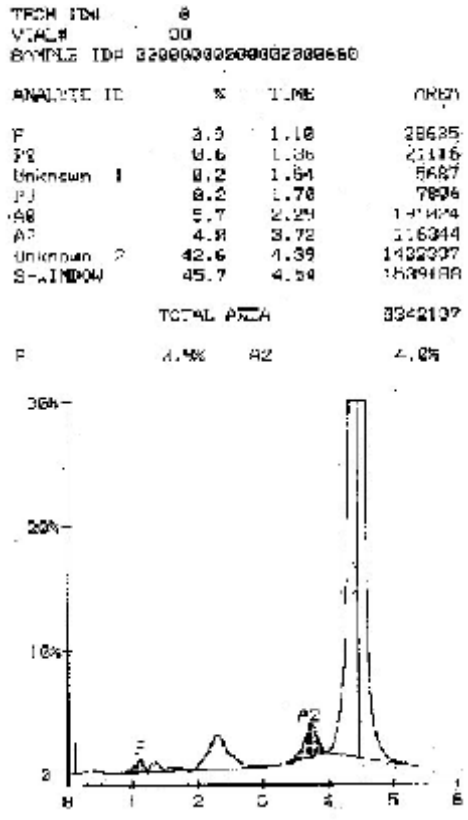

Figure 1. HPLC isolation of S and E-Saskatoon hemoglobins.

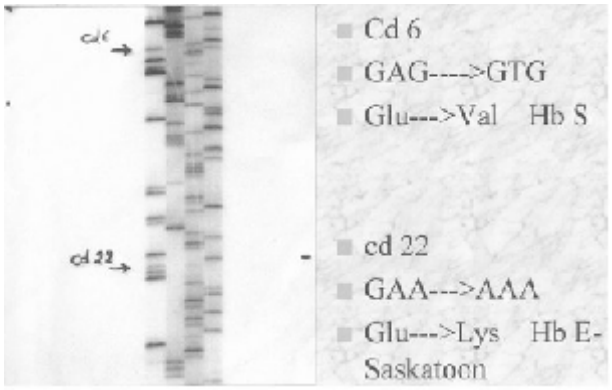

Figure 2. DNA sequencing.

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 haemoglobin S and E[8,9] reported to be symptomatic. This compound heterozygosity is the first description in Greece and in literature.

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