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CASE REPORT

First observation of homozygote Hb Q-Iran (alpha 75 (EF4) Asp-His)

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ABSTRACT

The first observation of homozygote Hb Q-Iran (α1 75(EF4) Asp-His) is presented in this report. The clinical and hematological data of the index case, his father and mother showed that homozygous or heterozygous Hb Q-Iran has no clinical importance.

Key words: Hemoglobin, Q-Iran

ÖZET

Homozigot Hb Q İran'ın (alpha 75 (EF4) ASP-HIS) ilk kez belirlenmesi

Sivas kökenli anormal hemoglobini olan bir çocuğa moleküler analizle Homozigot Hb Q-Iran (α1 75(EF4) Asp-His tanısı konulmuştur.

Anahtar kelimeler: Hemoglobin, Q-İran

INTRODUCTION

During screening surveys for beta thalassemia and abnormal hemoglobins in İstanbul, a well-known city in Turkey, a hemoglobin variant was detected in a six–year- old boy from Sivas, a city in Mid-Anatolia, with a hemoglobin variant mobility similar to Hb S with a negative sickling test, with iron deficiency anemia. Further analysis of the variant revealed it as Hemoglobin Q-Iran (a1 75 (EF4) Asp to His).

Hemoglobin (Hb) Q-Iran is a rare hemoglobin variant, which was reported first in an Iranian, and then in Turkish, Chinese and Pakistani families [1-6]. However, our case is the first homozygous Hb Q-Iran.

CASE REPORT AND METHODS

The index case had a hemoglobin variant mobility similar to Hb S with a negative sickling test. Routine hematological methods were used. Cellulose acetate electrophoresis analysis revealed that the variant constituted 42.0% of the total Hb, and Hb F (<1%) values were within normal levels. His hematological results following oral iron therapy for three months are shown in Table 1. Family screening revealed the father and the mother as heterozygote carriers of the variant. Both had the variant as 20 and 22.2%, respectively. The propositus was in homozygous state. No hematological data was available for the parents.

Molecular techniques described previously for the common mutations did not reveal the variant [7]. For further variant analysis, DNA was obtained from all individuals following a written informed consent. Sequencing of the exon 1, 2 and 3 of the beta globin gene was performed according to our previous report, which did not reveal an abnormal variant (Beckmann Coulter, USA) [8]. The entire coding and intronic sequence of alpha-1 and alpha-2 globin genes was amplified as one amplicon each. While the forward primer was the same for the two genes, the reverse primers were specific to alpha-1 and alpha-2 genes. These amplicons were sequenced using internal primers. The reactions were done using Beckman DTCS kit and run on Beckman CEQ 8000 automated DNA sequence analysis system. The mutation located on codon 75 of alpha-1 globin gene was not present on the alpha-2 gene amplicon. Sequence analysis was repeated on an independent polymerase chain reaction (PCR) to confirm the presence of the mutation. Our analysis revealed an alteration of Asp to His at 75 (EF4) in homozygous state, which was named previously as Hb Q-Iran (Figure 1).

Table 1. Hematological and hemoglobin composition data of the propositus

Hb	RBC	PCV	RET	MCV	MCH	MCHC	Hb A2	RDW
g/dl	1012/1	L/l	%	fl	pg	g/dl	%	%
13.2	5.03	39.9	0.1	72.2	23.8	33	0.5	12.2

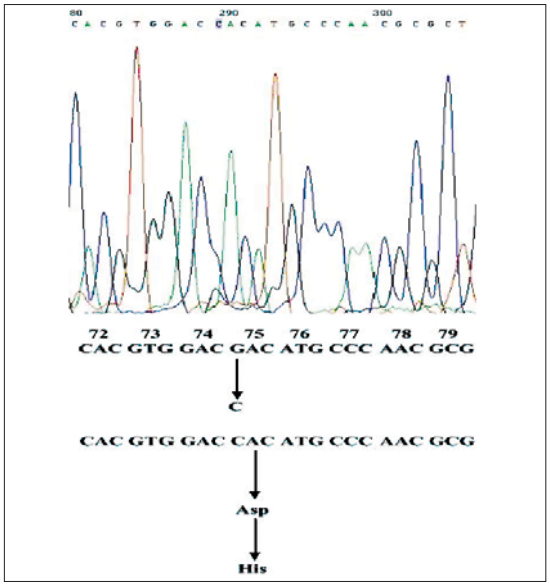

Figure 1. Sequencing data of Hb Q-Iran.

DISCUSSION

Two individuals from two Turkish cities, Adana and Ankara, were previously reported to have Hb Q-Iran. The latter was a 13-year-old girl with acute lymphoblastic leukemia with a combination of hemoglobin S and hemoglobin Q-Iran [4]. All the reported hemoglobin Q-Iran individuals were in heterozygote state. Here we describe a homozygous individual for hemoglobin Q-Iran without any clinical symptoms following iron treatment.

Turkey is situated at the meeting point of three continents of the world and stands as a crossroad between Asia and Europe. Due to its geographical location, Anatolia (Turkey) has historically been home to the various races and ethnic groups of three continents, a result of which is the reported presence of many kinds of hemoglobin variants in Turkey. Although the most important variant in the Turkish population is Hb S, there are also several examples of some rare variants [6,9].

Further observation of hemoglobin Q-Iran suggests that this variant is found sporadically in the Turkish population. With description of variants such as Hb Hamadan, Hb J Iran, Hb D Iran, Hb D Punjab and Hb Q Iran in the Turkish and Iranian populations, it is logical that the ancestral individuals probably immigrated from a region of Iran or back to Middle Asia [5-14].

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