First observation of hemoglobin Crete [Beta129(H7) Ala>Pro] in the Turkish population

Türk populasyonunda ilk gözlemlenen hemoglobin Crete [Beta129(H7)Ala>Pro]

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To the Editor,

Hemoglobin (Hb) Crete [β129 (H7) Ala>Pro] is a rare hemoglobin variant that was first reported in a Greek family living in USA, and then from Greece [1-4]. It was reported in combination with beta thalassemia and delta-beta-thalassemia [2], and also in a homozygous state [4]; however, this is the first report of Hb Crete in the Turkish population [5,6].

Herein we describe a 50-year-old Turkish man living in İzmir with Hb Crete. His family emigrated from Crete, Greece when he was 4 years old. He presented to the hospital with weight loss and hepatosplenomegaly. His routine hematological analysis results are shown in the Table 1. HPLC chromatography showed 1 abnormal band with 56.9% variance. Thoracoabdominal tomography showed hepatosplenomegaly and several multiple nodular lesions. Following posterolateral thoracotomy and paravertebral nodular excision, the histopathological diagnosis of the nodules was extramedullary hematopoiesis. Subsequently, the patient underwent splenectomy. We have the informed consent of the patient.

PCR amplification of the β-globin gene was performed using primers F:5'-GGTT GGCCAATCTACT CCCAGGAG-3' and R:5'-GCTCACTCAGTGTGGCAA AG-3' for exon 1-exon 2. For exon 3, PCR amplification was performed first with the primers F:5'-CAATGTATCATGCCTCTTTGCACC-3' and R:5'-GAGTCAAGGCTGA GAG ATAC AGGA-3' for a 861-bp fragment, and then 5'-TGCATATAAAT TGTAACTGAT-3' and 5'-CACT GACCTCCCACAT TCCC-3' primers were used for nested amplification. Direct automated sequencing of the all amplified regions of the β-globin gene was performed using an automatic sequencer (Beckmann Coulter, USA), and 2 different sets of PCR reactions with forward and reverse amplification were performed. The third exon amplification showed the variant as a missense mutation at codon 129 G change to C that leads to alanine substitution to proline, which was previously described as Hb Crete (Figure 1). Moreover, β-globin 5' UTR + 20 (C-T) change was observed, indicating a combination with beta thalassemia.

There are several reported 5' UTR mutations of the β-globin gene that cause the β-thalassemia

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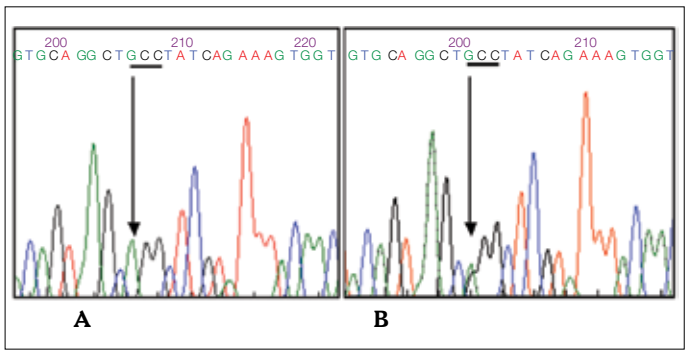

Figure 1. Automatic DNA sequencing data for the Hb Crete patient. Ais the normal sequence, B is the Hb Crete sequence

Table 1. Hematological and Hemoglobin Composition Data For the HB Crete Carrier

Hb	RBC	MCV	MCH	MCHC	RDW	RET
14.7	7.09	65.4	20.8	31.7	14.5	2.5

intermedia clinical phenotype or a combination with another abnormal Hb [7]. The presented patient had erythrocytosis, which was described previously and may be due to high oxygen affinity/ normal Bohr effect of Hb Crete [1], and may also account for the extramedullary foci observed in our patient.

We reported the first observation of Hb Crete in the Turkish population in combination with β-thalassemia. The presented patient was living in İzmir, on the Aegean coast of Turkey. Large numbers of Cretan Turks emigrated to İzmir at the beginning of the 20th century. Detection of Hb Crete in the Turkish population might be evidence of this historical event.

Conflict of interest statement

The authors of this paper have no conflicts of interest, including specific financial interests, relationships, and/or affiliations relevant to the subject matter or materials included.

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