First Observation of Hemoglobin Pyrgos [β83(EF7) Gly→Asp] in Turkish Population

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ABSTRACT

Hemoglobin Pyrgos [β83 (EF7) Gly→Asp] is a rare hemoglobin variant. This report describes the first observation of this variant in an 18-year-old Turkish girl living in Isparta.

Key Words: Hb pyrgos, Turkey.

ÖZET

Türk Toplumunda Gözelenen İlk Hemoglobin Pyrgos [β83(EF7) Gly→Asp]


Anahtar Kelimeler: Hb pyrgos, Türkiye.


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Hemoglobin Pyrgos [β83 (EF7) Gly→Asp] is a rare hemoglobin variant which was reported first in a Greek family, then from Japan, China, African Black from the republic of Mali, Italy, and Thailand[1-7]. It is a fast moving variant, reported in combination with Hb S, Hb E in association with a-thalassemia and Hb H disease with Hb Constant Spring[2,8,9]. The present report describes the first observation of this variant in an 18-year-old Turkish girl living in Isparta[10]. Routine hematological methods were used.

Cellulose acetate electrophoresis showed one abnormal band with a mobility faster than Hb A the amount of variant was 56.9%. Her hematological results are shown in Table 1.

PCR amplification was performed with the primers 5’TCATTCGTCTGGTCCATTCTAAC3’ and 5’GGTTGCGCAATCTACTCCAGGAG3’ and Inc., direct automated sequencing of the exon 2 of the beta globin gene (Beckmann Coulter, USA) revealed the variant as a missense mutation at codon 83 GGC-GAC.

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that leads to Gly to Asp substitution which was previously described as Hb Pyrgos (Figure 1). Isparta city, which the patient is originated, lies at the inner region of Mediterranean Area of Turkey. Detection of Hb Pyrgos in our population, fills the gap of the geographic area in which Hb Pyrgos was found i.e. far East to Italy.

REFERENCES


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Table 1. Hematological and hemoglobin composition data for Hb Pyrgos carriers

<table>
<thead>
<tr>
<th>Hb</th>
<th>RBC</th>
<th>PCV</th>
<th>RET</th>
<th>MCV</th>
<th>MCH</th>
<th>MCHC</th>
<th>Hb A2</th>
<th>RDW</th>
</tr>
</thead>
<tbody>
<tr>
<td>g/dL</td>
<td>L/1</td>
<td>%</td>
<td>fl</td>
<td>pg</td>
<td>g/dL</td>
<td>%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>13.2</td>
<td>5.03</td>
<td>41.2</td>
<td>0.1</td>
<td>82</td>
<td>26.2</td>
<td>32</td>
<td>1.5</td>
<td>15.1</td>
</tr>
</tbody>
</table>

Figure 1. Automatic DNA sequencing data of the patient showing Hb Pyrgos.