



First Observation of Hemoglobin Jabalpur [Beta 3 (NA3) Leu>Pro] in the Turkish Population

Türk Toplumunda İlk Gözlemlenen Hemoglobin Jabalpur [Beta 3 (NA3) Leu>Pro]

Ayfer Çolak¹, Burak Toprak¹, Kanay Yararbaş², Fatma Akyol¹, Cengiz Ceylan³

¹Tepecik Teaching and Research Hospital, Clinic of Clinical Biochemistry, İzmir, Turkey

²Düzen Laboratory Group, İstanbul, Turkey

³Tepecik Teaching and Research Hospital, Clinic of Hematology, İzmir, Turkey

To the Editor,

Hemoglobin Jabalpur [beta 3(NA3) Leu>Pro] is a rare hemoglobin variant previously described in the HBVar database of the Globin Gene Server [1]. In the present paper we report Hb Jabalpur identified in a Turkish family. This is the first report of Hb Jabalpur in the Turkish population.

The patient was an asymptomatic 19-year-old male who presented to the outpatient hematology clinic for further evaluation of mild anemia. On physical examination the patient's liver and spleen were not palpable. The patient's complete blood cell count showed a hemoglobin level of 116 g/L (Table 1). Cation-exchange high-performance liquid chromatography, which was performed using the Primus Ultra2 Hb variant analyzer (Trinity Biotech, Dublin, Ireland), showed abnormal hemoglobin amounting to 30.9% of total hemoglobin. Hb Jabalpur was eluted immediately before Hb A0 in the Hb A window.

Using direct Sanger sequencing (ABI 3130 Genetic Analyzer, Applied Biosystems, Foster City, CA, USA), we characterized the Hb variant as resulting from a CTG>CCG (c.11T>C, p.L4P, rs63750720, NCBI refSeq: NG_000007.3) replacement at codon 3 of the β -globin chain, corresponding to a Leu→Pro amino acid substitution (Figure 1). This mutation is not listed in the Human Gene Mutation Database as a disease-causing mutation, while it is present in the Hemoglobin Variation Database (HbVar: A Database

of Human Hemoglobin Variants and Thalassemias) as Hb variant Jabalpur. Informed consent was obtained.

The parents of the patient were also available for evaluation. DNA sequencing and chromatography analysis revealed that the mother also had Hb Jabalpur. The mother was found to have mild anemia (Hb: 107g/L) and no clinical symptoms were detected (Table 1).

The index patient and his mother were living in İzmir, in the Aegean region of Turkey. Hb Jabalpur was detected in another patient of Pakistani origin with compound heterozygous beta zero thalassemia, and moderate anemia (Hb: 97g/L) was also observed in that patient (Gallivan M 2010, personal communication). In the present report, both the index patient and his mother presented with mild anemia. It is known that the prevalence of the beta-thalassemia carrier state is high in Turkey; furthermore, several rare hemoglobin variants were previously reported. Hb Jabalpur and many rare hemoglobin variants generally do not produce clinical symptoms, but the consequences of the interaction between Hb variants and beta-thalassemia may be clinically important.

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.

Address for Correspondence: Burak TOPRAK, M.D.,
Tepecik Teaching and Research Hospital, Department of Clinical Biochemistry, İzmir, Turkey
Phone: +90 232 469 69 69 E-mail: beiot@hotmail.com

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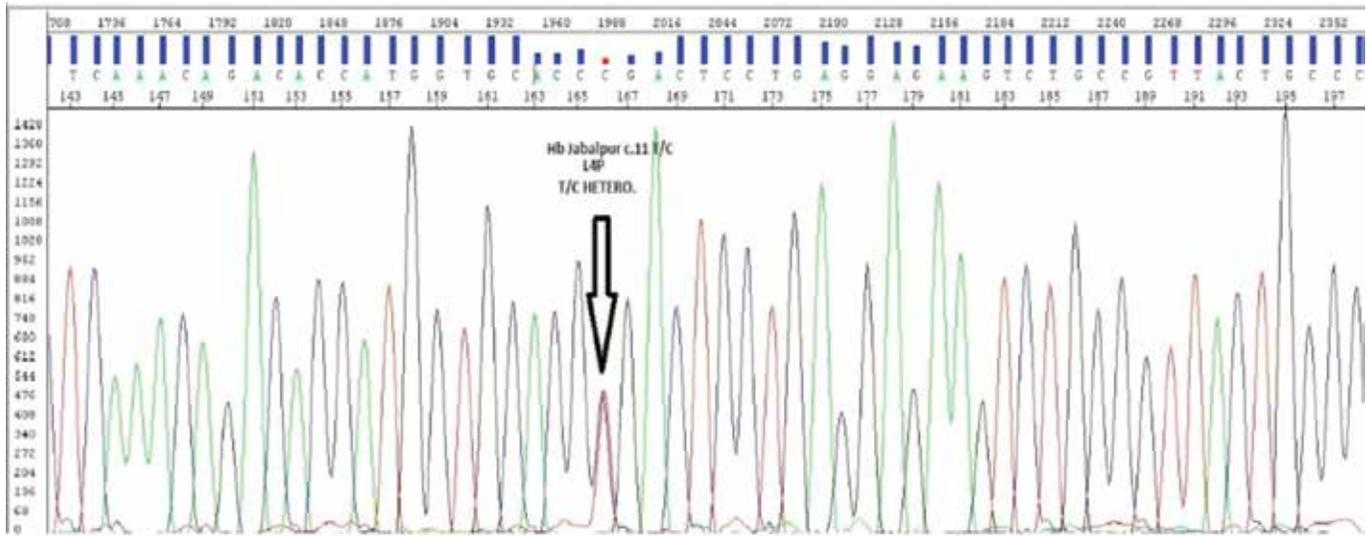


Figure 1. DNA sequencing data showing Hb Jabalpur.

Table 1. Hematological data of index patient and his mother.

	Hb (g/L)	RBC (10 ¹² /L)	MCV (fL)	MCH (pg)	MCHC (g/dL)	RDW (%)
Index patient	116	4.88	76	23.7	31.2	14.1
Mother	107	3.99	84.8	26.8	31.6	15

Key Words: Hemoglobinopathy, Hb Jabalpur, High-performance liquid chromatography

Anahtar Sözcükler: Hemoglobinopati, Hb Jabalpur, Yüksek performanslı sıvı kromatografisi

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