Survey on haemoglobin variants, β thalassaemia, glucose-6-phosphate dehydrogenase deficiency, and haptoglobin types in Turks from Western Thrace

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SUMMARY A total of 102 apparently healthy and randomly selected Turks who either immigrated from Western Thrace or were still living there were studied for haemoglobin variants, high Hb A₂ β thalassaemia, G6PD deficiency, and haptoglobin types. The incidence of haemoglobins S and O Arab were 2.9 and 3.9% respectively. The incidence of high A₂ β thalassaemia was 10.8% and that of G6PD deficiency 5%. The gene frequencies of Hp1 and Hp2 were 0.326 and 0.674, respectively.

The occurrence of numerous haemoglobinopathies, such as sickle cell anaemia, sickle cell β^+ or β^0 thalassaemia, sickle cell-haemoglobin O Arab disease, and β^0 or β^+ thalassaemia-haemoglobin O Arab combinations, prompted us to conduct a survey on abnormal haemoglobins, high A₂ β thalassaemia, glucose-6-phosphate dehydrogenase (G6PD) deficiency, and the distribution of haptoglobin types in Turks from Western Thrace, a district in Greece close to the Turkish border.

The purpose of this paper is to report the results of this study.

Material and methods

A total of 102 apparently healthy and randomly selected Turks who either immigrated from Western Thrace to Turkey or still lived there were studied. Mostly they were from Komitine (Gümülcine), Thessalonikis (Selanik), and Xanthe (Iskeçe). The majority were students from several faculties of Istanbul University and a few were healthy relatives of Turkish immigrants from Western Thrace with different acquired haematological disorders. The group comprised 60 males and 42 females, whose ages varied between 13 and 68 years (mean 34 years). The methods for blood counts, electrophoretic analyses of the haemoglobin by starch gel and

cellulose acetate, quantitative estimations of haemoglobins A₂ and F, haptoglobin typing, and the screening method for G6PD activity have been described elsewhere.¹ For the differentiation of abnormal haemoglobins with similar electrophoretic mobility, such as haemoglobins S and D or haemoglobins O Arab and E, the sickling test with 2% Na-metabisulfite and citrate-agar gel electrophoresis (Helena plates at pH 6.2) was used.²

Results

The results of haemoglobin analyses are given in table 1. Three subjects with sickle cell trait were found among these people (Hb S: 36 to 41%; Hb A₂: 2.1 to 2.6%; Hb F: 0 to 0.1%). Four subjects with haemoglobin O Arab (32 to 35%) were also found. Structural analysis of the Hb E-like fraction of one of the patients, performed by liquid chromatography (HPLC) in the Protein Chemistry Laboratory of

TABLE 1 Distribution of abnormal haemoglobins and high A₂ β thalassaemia in 102 Turks from Western Thrace.

	Subjects	%
Normal	82	80.4
Sickle cell trait	3	2.9
Heterozygous haemoglobin O Arab	4	3.9
Heterozygous high A ₂ β thalassaemia	11	10.8
Heterozygous normal A ₂ β thalassaemia or α thalassaemia I	2	2.0

TABLE 2 Distribution of haptoglobin types in 101 Turks from Western Thrace*

Haptoglobin types	Observed	Expected	%
1-1	10	10.6	10.0
2-1	45	43.9	44.5
2-2	45	45.5	44.5
0 [‡]	1	1.0	

$\chi^2=0.061$, Hp1=0.326, Hp2=0.674.

*Haptoglobin typing was performed in 101 out of 102 Turks.

[‡]The subject with haptoglobin O did not exhibit any findings of increased haemolysis.

the Medical College of Georgia, identified a lysyl residue at the aminoterminal of the tryptic peptide 13 of the β chain where it replaced the normally occurring glutamic acid residue. This Glu→Lys substitution is characteristic of Hb O Arab. The incidence of Hb O Arab among Turks from Western Thrace was 3.9%.

There were 11 heterozygotes with high $A_2\beta$ thalassaemia. The incidence of this type of β thalassaemia among Turks from Western Thrace was 10.8%. In addition, two subjects showed a mild microcytic anaemia with normal levels of haemoglobins A_2 and F and serum iron. Unfortunately, we did not perform globin chain synthesis studies and gene mapping of DNA. Therefore, we are unable to state whether these two subjects have heterozygous normal $A_2\beta$ thalassaemia or α thalassaemia.

The incidence of G6PD deficiency among 60 males was 5%. None of them was heterozygous for high $A_2\beta$ thalassaemia or haemoglobins S and O Arab.

The results of the haptoglobin types are given in table 2. Proceeding from this, the gene frequencies were calculated as Hp1 0.326 and Hp2 0.674. One subject had HpO.

Discussion

In this survey 20 subjects among 102 apparently healthy Turks from Western Thrace had either an abnormal haemoglobin variant or a heterozygous form of β thalassaemia.

HAEMOGLOBIN S

Apart from Eti-Turks, an Arabic speaking population living in south western Turkey, the incidence of Hb S in Turks is generally very low, ranging between 0.20 and 0.37%, according to three studies performed.³⁻⁵ Despite this, there are some foci of this abnormal haemoglobin in Turkey.^{1,6} In the present study an incidence of Hb S of 2.9% was found among Turks from Western Thrace.

HAEMOGLOBIN O ARAB

The incidence of Hb O Arab among Turks from Western Thrace is 3.9%. Hb O Arab has been found in Arabs living in Israel,⁷ in Egypt,⁸ in Aden,⁹ in Sudan,¹⁰ in Turks,¹¹ in Turkish Cypriots,¹² and in Negroes from Jamaica,¹³ and the United States.¹⁴ Furthermore, a total of 17 families with Hb O Arab, mostly associated with heterozygous β thalassaemia, and one with the homozygous form, have been reported from Bulgaria.¹⁵ Fourteen of them lived in Burgas, Eastern Thrace, and the remaining three were from other regions of Bulgaria. Additionally, a few families with Hb O Arab have been found in Romania,¹⁶ Yugoslavia,¹⁷ and Hungary.¹⁸ Sharma *et al*¹⁹ described a family with Hb O Arab β^0 thalassaemia of Greek origin living in Australia, but they noted that the family was of Turkish descent and came from Komitine. Therefore, this family was similar to those presented here. Up to the present, we have studied nine families with Hb O Arab β^0 or β^+ thalassaemia, one family with sickle cell-Hb O Arab disease,²⁰ and one family with heterozygous Hb O Arab. All were Turkish immigrants from Western Thrace. It seems most likely that at present there are two foci of Hb O Arab in Eastern Europe: one comprises Turks from Western Thrace and the second comprises Bulgars mainly living in the district of Burgas. However, it is difficult to explain the absence of Hb O Arab in Greeks living in Western Thrace. This problem needs further investigation.

HIGH $A_2\beta$ THALASSAEMIA

The incidence of this type of β thalassaemia among Turks from Western Thrace is as high as 10.8%. According to Arcasoy and Çavdar²¹ the incidence of high $A_2\beta$ thalassaemia in Turkey is 2.1%. Furthermore, as pointed out by Dinçol *et al*,²² although high $A_2\beta$ thalassaemia has been found in every region of Turkey, it is rather more frequently encountered in the western part of the country. The present study confirms this.

HAPTOGLOBIN TYPES

The distribution of haptoglobin types among Turks from Western Thrace was a little different from that found in Turkish people in general (table 2). The percentage of Hp 1-1 was a little higher than found in Turks in general: 10% and 7.5% respectively.²³ Furthermore, Hp1 gene frequency among Turks from Western Thrace was also high when compared with those of other Asiatic populations, including Turks in general: 0.326 and 0.265 respectively. Hp1 gene frequency among Asiatic people varies between 0.21 and 0.31.²⁴

G6PD DEFICIENCY

The incidence of G6PD deficiency in Turks from Western Thrace was found to be moderately high at 5%. Contrary to this, the overall incidence of G6PD deficiency in Turks in general is low at 0.6%,¹ but there are some foci of comparatively high incidence of this enzyme deficiency.¹

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