Prevalence of Beta Thalassemia Trait in Denizli

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ABSTRACT

Beta thalassemia, characterized by the deficiency or the absence of beta globulin production, is the most widespread inherited disorder in the world and is also common in Turkey. To determine the prevalence of carriers for beta thalassemia, we screened the couples before their marriage. For this aim, from 1994 to 1999, a total of 14,200 people were screened. The complete blood count and red blood cell indices (hemoglobin: Hb, hematocrite: Hct, median corpuscular volume: MCV, median corpuscular hemoglobin: MCHb, median corpuscular hemoglobin concentration: MCHbC, concentration were measured by automated cell counter on the same day of collection. Then for the samples with MCV values of 78 fL or below, hemoglobin electrophoresis were employed. Testing for beta thalassemia was carried out by the conventional cellulose acetate electrophoresis at pH 8.4. People who have elevated HbA2 (≥ 3.5%) were accepted as beta-thalassemia carrier or patient. We detected 3300 people with MCV levels of 78 fL or below and 311 cases of beta thalassemia carrier and 11 cases of beta thalassemia. The prevalence of carriers for beta thalassemia in Denizli was 2.2%. This result indicated that the people with anemia in our region should be investigated for the existence of hemoglobinopathy.

Key Words: Anemia, Beta thalassemia, Prevalence, Hemoglobinopathy.


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INTRODUCTION

Beta thalassemia, one of the thalassemia syndromes, is an autosomal recessive inherited disease[1-3]. Thalassemia is one of the most frequent hereditary haematological disorders in the world[2-4]. When epidemiological distribution maps are reviewed, the incidence of beta thalassemia is greatest in people of Mediterranean and African origin[3,4].

High-HbA2 type beta thalassemia incidence in Turkey varies from one region to another. In various regions of Turkey, the incidence of high-HbA2 type beta thalassemias ranged from 1.1 to 10.8[5-15]. Until now, there have not been any studies in this city and total number of high HbA2 type beta thalassemia in Denizli is not precisely known. This program was planned in order to determine the prevalence of the high-HbA2 type beta thalassemia in the city of Denizli which is located in the Aegean region of Turkey.

MATERIALS and METHODS

This study was performed in the genetic laboratory for thalassemia of the Ministry of Health. To determine the prevalence of carriers for high-HbA2 type beta thalassemia, we screened couples before marriage. For this aim, from 1994 to 1999 a total of 14,200 people preparing for marriage were screened by haematological analysis. Participants were requested to complete a questionnaire about their sex, age, health status, place of birth, the place of immigration of their ancestors and the history of any type of hemolytic anemia in their families. We used the method reported by Cao et al., high HbA2 type beta thalassemia screening[16]. Venous blood was taken into both EDTA and citrate anticoagulated tubes (1 citrate versus 9 blood) and kept at 4°C in a refrigerator until studied. The complete blood count and red blood cell indices (MCV, MCHb, MCHbC) were measured by Cell-Dyn (Coulter Diagnostics) on the same day of collection. Hemoglobin electrophoresis on cellulose acetate at pH 8.4 (using Helena’s kit and Cliniscan II Dansitometer) was performed for those with MCV 78 fl or below, within one week of collection. People who had elevated HbA2 levels (≥ 3.5%) were diagnosed as beta thalassemia carrier or patient. In this study, we didn’t investigate the hemoglobin variants and thalassemia carriers with normal HbA2 levels.

RESULTS

We screened 14,200 participants (50% female and 50% male) ranging from 14 to 54 years of age. The prevalence of high HbA2 beta thalassemia trait was found to be 2.2% (311/14,200) and the prevalence of beta thalassemia intermedia was found to be 0.077%. When projected by this prevalence rate (2.2%), it is expected to have 22,000 potential beta thalassemia traits in Denizli. This rate was moderately higher compared to the whole of Turkey, which is 2%. The results are shown in Table 1.

DISCUSSION

Beta thalassemia, which is more frequently seen in the Mediterranean, Africa and Asia is an important problem for public health[1-3]. The aim of the survey on the genetic disorders is to identify the carriers of genetic diseases and then to establish preventive measures such as genetic and prenatal counseling of the affected couples to improve the health status of the population concerned[6,7]. The prevalence of beta thalassemia trait differs from country to country. This state can be related to the ethnicity of people. The prevalence of beta thalassemia trait was found to be 3.1% for Tunisia and Pado-

Table 1. The beta thalassemia status in Denizli

<table>
<thead>
<tr>
<th></th>
<th>Number of people</th>
<th>Prevalence (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total people</td>
<td>14,200</td>
<td></td>
</tr>
<tr>
<td>People with MCV≤ 78 fl</td>
<td>3,300</td>
<td>23.2</td>
</tr>
<tr>
<td>Beta thalassemia trait</td>
<td>311</td>
<td>2.2</td>
</tr>
<tr>
<td>Beta thalassemia intermedia</td>
<td>11</td>
<td>0.077</td>
</tr>
</tbody>
</table>
va (Italy), 5.4% for Pakistan, 3.4% for Hong Kong and 3.09% for Corsica\cite{17-21}. But it was lower for Sicilian, Fillipinios, Indian, Denmark, Marseille, Taiwan and England\cite{21-27}. The prevalence of beta thalassemia trait is also high in the immigrant populations from the endemic regions. For example; the prevalence of beta thalassemia trait for Chinese people living in Canada is high\cite{28}. Also it is high in the immigrant German population\cite{29}.

In Turkey, thalassemia was first reported by Aksoy et al. in the southern part of the country\cite{30}. Various authors had reported for high-hemoglobin A$_2$ type beta thalassemia from 1.2 to 10.8 in Turkey\cite{5-15}. The incidence of beta thalassemia is 1.2 to 3.06% in the inner Anatolia and the Marmara region\cite{5,7,8,14}. But, the incidence of beta thalassemia is higher in the east and the south coasts of Turkey. Especially in the south region of Turkey, the incidence of thalassemia trait may be 10% of total population\cite{9,10,12,13}. The general ratio of beta thalassemia carriers in Turkey was stated to be 2\%\cite{5-7,11}.

The Aegean region constitute the west of Turkey and it has big provinces like İzmir, Aydın, Denizli and Muğla. There is limited data confirming the prevalence of beta thalassemia trait in the Aegean region. Aydnok et al have shown that the prevalence of beta thalassemia trait in İzmir was 3 percent\cite{15}. The prevalence of beta thalassemia in Aydın and Muğla is not known. Denizli is the second highest populated city of the Aegean region. In this study; the prevalence of high-hemoglobin A$_2$ type beta thalassemia was found to be 2.2% in Denizli. The prevalence in Denizli was moderately higher than the prevalence of Turkey (2.2% versus 2%), while it was lower than that of İzmir (2.2% versus 3% respectively)\cite{5-8,11}. We didn’t investigate the hemoglobin variants and the thalassemia carriers with normal HbA$_2$ levels. The higher incidence of İzmir might be due to the presence of different ethnic groups in this city as a result of higher immigration rate.

In conclusion, the prevalence of beta thalassemia trait is moderately higher in Denizli than the other districts of Turkey. Also, a preventive genetic program aimed at controlling beta thalassemia in the Denizli population, carrier screening, genetic counselling and prenatal diagnosis should be applied.

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**REFERENCES**


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