Original Article

Carrier frequency of β-Thalassaemia in Twin-Cities of Islamabad and Rawalpindi

Muhammad Iqbal, Omar Awwab Khan , Abdul Ghani Waseem, Muhammad Tahir
Railway Hospital and Islamic International Medical College, Rawalpindi

Abstract

Background: To determine the prevalence of β-thalassaemia carriers in twin-cities of Islamabad and Rawalpindi. Methods: In this descriptive study, people were motivated to get screened for β-thalassaemia trait. Hemoglobin level, RBC count, MCV, MCH was measured. Hemoglobin electrophoresis was performed if MCH was less than 27pg, MCV less than 75fl and RBC count more than 5 million. If HbA2 was 3.5% or higher, thalassaemia trait was diagnosed.

Results: A total of 2101 people were screened in camps held at offices of Federal Ministry of Health & Pakistan Post, two villages (Terlai & Phoolgran) in the suburbs of Islamabad, Fatima Jinnah and Foundation Universities and other educational institutions. 85 (4.05%) were found to be carriers of β-thalassaemia.

Conclusion: β-Thalassaemia Trait has a carrier frequency of around 4% in twin-cities and suburbs.

Key Words: β-thalassaemia; Carrier frequency

Introduction

Thalassaemias are genetic disorders characterized by reduced synthesis of either alpha or beta chain of hemoglobin. Most of the patients with severe forms of β-thalassaemia i.e. thalassaemia major need lifelong treatment including blood transfusion, iron chelation etc. β-thalassaemia is a preventable disease through comprehensive strategy at community level and this fact has been proved by Cypriot and Greek experience with prevention of this disease.

Preventive strategies involving identification of carriers, genetic counselling and prenatal diagnosis have now almost eliminated the risk of new children with homozygous thalassaemia in countries which were once known for the highest prevalence of thalassaemia. WHO recommends countries with heavy burden of non-communicable disease to have national programs for prevention. A careful estimate of the carrier frequency in the community through a survey involving large population is required.

Subjects and Methods

In Rawalpindi and Islamabad, camps were held at Federal Government offices including Federal Ministry of Health and Pakistan Post, at two villages Terlai and Phoolgran and educational institutions. People were informed about the genetic nature of the disease, its severity and cumbersome lifelong treatment, the inheritance pattern and possibility of an individual to be carrier of the disease and a potential risk of having a child with thalassaemia major, if both parents are carriers. All people willing for screening were included in study irrespective of their age, gender or ethnic background.

Five ml blood sample was taken in EDTA bottle, labelled and saved for laboratory workup. Haemoglobin levels, RBC count, MCV, MCH were measured using an automated electronic haematology analyzer. Haemoglobin electrophoresis was performed if MCH was less than 27pg, MCV less than 75fl and RBC count more than 5 million. If HbA2 was 3.5% or higher, they were labelled as thalassaemic trait and others were advised to undergo iron studies. All individuals identified as thalassaemia carriers were invited to thalassaemia center, counselled regarding the risk they could face and given written information both in Urdu and English including inheritance pattern and prevention of thalassaemia.

Results

Over a period of four years, from 2006 to 2010 a total number of 2101 people were screened. Overall carrier rate was 4.05%, and was higher in rural population (Table 1).

Discussion

Identification of carriers is an essential part of prevention programs. Although a nationwide survey is required to know the real burden of disease, studies conducted on smaller number of subjects can be useful to estimate the burden of disease, particularly in a
country with limitation of resources and lack of efficient primary health infrastructure. 7

Table 1: Distribution of screened subjects and thalassaemia carriers

<table>
<thead>
<tr>
<th>Camp</th>
<th>Subjects Screened</th>
<th>β-Thalassaemia Trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>Federal ministry of health office, Islamabad</td>
<td>237</td>
<td>9(3.79)</td>
</tr>
<tr>
<td>Pakistan post, Islamabad</td>
<td>406</td>
<td>12(2.96)</td>
</tr>
<tr>
<td>Terlai village</td>
<td>206</td>
<td>21(10.19)</td>
</tr>
<tr>
<td>Phoolgran village</td>
<td>270</td>
<td>15(5.56)</td>
</tr>
<tr>
<td>Fatima Jinnah University</td>
<td>262</td>
<td>5(1.91)</td>
</tr>
<tr>
<td>Foundation University</td>
<td>307</td>
<td>8(2.61)</td>
</tr>
<tr>
<td>Other educational institutions</td>
<td>413</td>
<td>15(3.63)</td>
</tr>
<tr>
<td>Total</td>
<td>2101</td>
<td>85/2101(4.05%)</td>
</tr>
</tbody>
</table>

A study, in thalassaemia patients’ families, revealed frequency of consanguineous marriages in parents as first cousin marriages in 72%, second cousin marriages in 5%, distant cousins in 4% and unrelated in 19%. In our study 4.06% people were found to be carriers of β-thalassaemia which is comparable to the earlier reports (Table 2). Overall estimation of carrier rate at national level is around 5%. However there is variation in the prevalence of thalassaemia in different parts of country. There is, relatively, a high prevalence in Pathans (7.9%) as compared to Punjabis (3.26%). In a family having a patient of β-thalassaemia major (extended family) the prevalence of beta thalassaemia trait is higher (31%).

Our neighbouring countries also have more or less a comparable prevalence rate of β-thalassaemia carriers. Indian Punjab 3.5%, Saudi Arabia 3.4%, Southern China 2.54% and Hong Kong 3.4%.

Table 2: Carrier Frequency in Pakistan

<table>
<thead>
<tr>
<th>Population</th>
<th>Region</th>
<th>β-thalassaemia Trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathans 12</td>
<td>Peshawar/Rawalpindi</td>
<td>7.96%</td>
</tr>
<tr>
<td>Pakistani 8</td>
<td>Rawalpindi</td>
<td>3.9%</td>
</tr>
<tr>
<td>Punjabi 12</td>
<td>Rawalpindi</td>
<td>3.3%</td>
</tr>
<tr>
<td>Pakistani 9</td>
<td>Karachi</td>
<td>3.4%</td>
</tr>
<tr>
<td>Pakistani 12</td>
<td>Rawalpindi/Peshawar</td>
<td>5.4%</td>
</tr>
</tbody>
</table>

A carrier rate of 4% is considered to be among the high prevalence which can result in heavy burden of disease particularly in the absence of an effective preventive strategy. First part of such preventive program should be “targeted screening” involving families already having a patient with β-thalassaemia. It can be cost-effective, convenient and easily acceptable intervention to reduce the burden of disease. 15

References