Frequency of Iron Deficiency Anaemia in First Degree Relatives of Beta Thalassaemia Major Patients

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ABSTRACT

Objective: To find out frequency of iron deficiency anaemia in first degree relatives beta thalassaemia major patients.

Subjects & Methods: This is a comparative and non-interventional study. It was carried out in Fatimid Foundation and Sundus Foundation, Lahore between January 2008 to June 2008. A total of 300 subjects were taken. Out of which 200 subjects were first degree relatives of beta thalassaemia major patients and 100 were normal control subjects. Complete blood count with peripheral smear was done. Serum iron, serum TIBC, serum ferritin, haemoglobin electrophoresis and HbA2 estimation were carried out.

Results: There were 93 males and 107 females of thalassemia minor between 18-50 years of age. Normal or increased level of serum ferritin was found in 186 subjects and low level of serum ferritin (iron deficiency anemia) was found in 14 subjects with range <10 ng/ml.

Conclusion: The percentage of iron deficiency anaemia patients among the relatives of beta thalassaemia major patients was 7%.

Key words: Iron deficiency anaemia, First degree relatives, Beta thalassaemia major

INTRODUCTION

Thalassemia is the most common recessive single gene disease in human which is caused by inheritance of an affected allele from both parents. In the most severe forms, found in homozygotes or compound heterozygotes, the anemia is lethal within the first years of life in the absence of any treatment. Iron deficiency anemia is classically described as a microcytic anemia. The differential diagnosis includes thalassemia, sideroblastic anemias, some types of anemia of chronic disease, and lead poisoning. Serum ferritin is the preferred initial diagnostic test. It can cause reduced work capacity in adults and impact motor and mental development in children and adolescents.

In iron-deficiency anemia, storage iron declines until iron delivery to the bone marrow is insufficient for erythropoiesis. This can be monitored with clinical indicators, beginning with low plasma ferritin, followed by decreased plasma iron and transferrin saturation, and culminating in red blood cells with low haemoglobin content. When adequate dietary iron is provided, these markers show return to normal, indicating a response to the dietary supplementation.

Iron deficiency anemia and thalassemia minor are two of the most common causes of microcytic anemias worldwide. Because of similar red blood cell count parameters and blood picture, it was imperative to develop other measures that would differentially and correctly diagnose these two anemias. Iron deficiency is far commoner than iron overload. Uncomplicated iron deficiency anemia can be diagnosed easily by examination of the blood film and from the red cell indices. Thus, serum ferritin estimations can make an important contribution to the initial assessment of iron overload and can be used to follow the course of its treatment.

The most accurate initial diagnostic test for iron deficiency anaemia is the serum ferritin measurement. Serum ferritin values greater than 100 ng per mL (100 mcg per L) indicate adequate iron stores and a low likelihood of iron deficiency anemia. An accurate diagnosis is important because appropriate therapy may alleviate symptoms, inappropriate therapy may cause clinically important side effects, and the anemia may be a sign of underlying disease. It was recently found that the serum ferritin level is extremely useful in distinguishing between elderly anemic patients with and without iron deficiency and that no other standard laboratory test provides additional important information.

OBJECTIVE

The purpose of this study is to find out frequency of iron deficiency anaemia in first degree relatives of beta thalassaemia major patients.
MATERIALS AND METHODS

A total number of 200 apparently healthy first degree relatives of diagnosed beta thalassaemia major subjects of 18-50 years of age were included in the present study. Ten ml of venous blood was drawn. Tube containing 3mg of ethylene-diamine-tetra-acetic acid (EDTA) (disodium) was added in 2 ml of blood. Two blood smears from every sample were prepared and examined after staining with May Grunwald Giemsa Stain. Complete blood count was done by Sysmax analyzer Kx-21. Remaining blood was allowed to clot. Serum was separated by centrifugation at 3000 rpm for 5 minutes. Serum iron, serum TIBC were estimated on dimension AR chemistry analyzer. Haemoglobin electrophoresis using cellulose acetate was carried out for separating different haemoglobins. HbA2 estimation was carried out through column chromatography and serum ferritin was assed through ELISA (enzyme linked immunosorbent assay). The measurement of ferritin in serum is useful in determining changes in body iron storage.

RESULTS

The present study included 200 subjects who were first degree relatives of a beta-thalassemia major patient from Fatimid Foundation and Sundus Foundation, Lahore. Of these 200 subjects, the normal subjects were 78 (41 males being 20.5% and 27 females being 18.5% of the total); 108 were of beta thalassaemia trait (53 males, 26.5% and 55 females, 27.5%) and 14 subjects of iron deficiency anaemia (4 males, 2% and 10 females 5%) [Table 1]. According to age, there were 56 normal, 84 beta thalassaemia minor and 9 iron deficient subjects were found between the ages of 18-30 years. Seventeen normal, 20 beta thalassaemia minor and 2 iron deficiency subjects between 31-40 years of age and 5 normal, 4 beta-thalassaemia minor and 3 iron deficiency subjects were found between 41-50 years (Table 2). Out of 200 subjects, normal level of serum ferritin was found in 78 (39%), increased level i.e. in beta-thalassaemia minor was 108 (54%) and low level i.e. in iron deficiency anaemia was 14(7%) were seen (Table 3).

Table 1: Frequency distribution of genders according to normal, beta thalassaemia minor and iron deficiency anaemia

<table>
<thead>
<tr>
<th>Sex</th>
<th>Normal (%)</th>
<th>Beta thalassaemia minor (%)</th>
<th>Iron deficiency anaemia (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>41(20.5%)</td>
<td>55(26.5%)</td>
<td>4(2%)</td>
</tr>
<tr>
<td>Female</td>
<td>37(18.5%)</td>
<td>55(27.5%)</td>
<td>10(5%)</td>
</tr>
</tbody>
</table>

DISCUSSION

A study performed on the sibling of beta thalassemia major children have shown that the incidence was 58% with a male to female ratio of 0.9:1.15. In another study the prevalence of beta thalassemia minor with high HbA2 and microcytic hypochromic anaemia was 3.4% and includes 164 (53.4%) males and 143 (46.6%) females. Although beta thalassemia is an autosomally recessive disorder, there is a non-significant gender difference between males and females. A study was carried out in 88 patients there were 33 males and 55 females. The mean age was 42.9 years.

A study was carried out over beta thalassemia minor pregnant females had shown that ferritin concentrations were usually much higher and iron deficiency four times less common. Another study shown that low levels of ferritin were found in 61% of non-pregnant and in 32% of pregnant females beta thalassemia minor whereas male thalassemia carrier had normal iron stores.

A study was conducted in India over the relatives of severe transfusion-dependent beta thalassemia major children. Iron deficiency (serum ferritin <10.0 mcg/liter) was present in 6.3% of beta thalassemia minor males, 38.9% of control males. In females group, serum ferritin below 10 mcg/liter was present in 24.4% of beta thalassemia minor and 58.8% in control group. Thus the beta thalassemia minor had better iron nutrition. This also suggest that the beta thalassemia group has an advantage in maintaining iron balance.

In present study, the relatives of beta thalassaemia major had shown significant difference among males and females population. Out of two hundred subjects iron deficient subjects were 2% males and 5% females while among age group there was significant increase of 4.5% during 18-30 years of age. While serum ferritin had shown significant difference and 7% were iron deficient subjects.
CONCLUSION

Overall the study had shown that a significant number of iron deficient subjects are present among the relatives of beta thalassaemia major patients and the rate of females of 18-30 years of age are more iron deficient as compared to males. It is observed that out of 200 subjects, normal subjects are 78 (39%), beta thalassaemia minor subjects are 108 (45%) and iron deficiency anaemia subjects are 14 (7%).

REFERENCES