

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

¹NAFISA FATIMA, ²SHAHIDA AMJAD, RIAZ SHAH, ³AALIA HAMEED

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 thalassaemia 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

Thalassaemia is the most common recessive single gene disease in human which is caused by inheritance of an affected allele from both parents.¹ 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.² Iron deficiency anemia is classically described as a microcytic anemia. The differential diagnosis includes thalassaemia, sideroblastic anemias, some types of anemia of chronic disease, and lead poisoning. Serum ferritin is the preferred initial diagnostic test.³ It can cause reduced work capacity in adults⁴ and impact motor and mental development in children and adolescents.⁵

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.⁶

Iron deficiency anemia and thalassaemia 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 anaemias.⁶ 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.⁷

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.⁸ 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.⁹

OBJECTIVE

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

¹Senior Medical Officer, ²Professor & Incharge Blood Bank, ³Senior Demonstrator Shaikh Zayed Hospital Lahore
Correspondence to Dr. Nafisa Fatima, Department of Hematology, Shaikh Zayed Hospital, Lahore

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 was prepared and examined after staining with May Grunwald Giemsa Stain.¹⁰ 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 assayed 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-thalassaemia 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

Sex	Normal	Beta thalassaemia minor	Iron deficiency anemia
Male	41(20.5%)	53(26.5%)	4(2%)
Female	37(18.5%)	55(27.5%)	10(5%)

Table 2: Frequency distribution of ages according to normal, beta thalassaemia minor and iron deficiency anaemia

Age (years)	Normal	Beta thalassaemia minor	Iron deficiency anemia
18 – 30	56(42%)	84(42%)	9(4.5%)
31 – 40	17(8.5%)	20(10%)	2(1%)
41 – 50	5(2.5%)	4(2%)	3(1.5%)

Table 3: Frequency distribution of serum ferritin (ng/ml)

Serum ferritin	Frequency	%age
Normal	78	39.0
Thalassaemia minor	108	54.0
Iron deficiency anemia	14	7.0

DISCUSSION

A study performed on the sibling of beta thalassaemia major children have shown that the incidence was 58% with a male to female ratio of 0.9:1.15.¹¹ In another study the prevalence of beta thalassaemia minor with high HbA₂ and microcytic hypochromic anemia was 3.4% and includes 164 (53.4%) males and 143 (46.6%) females.¹² Although beta thalassaemia 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.¹³

A study was carried out over beta thalassaemia minor pregnant females had shown that ferritin concentrations were usually much higher and iron deficiency four times less common.¹⁴ Another study shown that low levels of ferritin were found in 61% of non-pregnant and in 32% of pregnant females beta thalassaemia minor whereas male thalassaemia carrier had normal iron stores.¹⁵

A study was conducted in India over the relatives of severe transfusion-dependent beta thalassaemia major children. Iron deficiency (serum ferritin <10.0 mcg/liter) was present in 6.3% of beta thalassaemia minor males, 38.9% of control males. In females group, serum ferritin below 10 mcg/liter was present in 24.4% of beta thalassaemia minor and 58.8% in control group. Thus the beta thalassaemia minor had better iron nutrition. This also suggest that the beta thalassaemia group has an advantage in maintaining iron balance.¹⁶

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

1. Borgna-Pignatt C, Galanello R. Thalassemsias and related disorders: quantitative disorders of hemoglobin synthesis. In: Lee GR, Foerster J, Lukens JN, Eds. *Wintrobe's Clinical Hematology*. 10th ed. Philadelphia: Lippincott Williams & Wilkin, 2004; 1319-65.
2. Cooley T, Lee P. A series of cases of splenomegaly in children with anemia and peculiar bone changes. *Trans Am Pediatr Soc* 1925; 37: 29-33.
3. Killip S, Bennett JM, Chambers MD. Iron deficiency anemia. *Am Family Phys* 2007; 13-21.
4. Haas JD, Brownlie T IV. Iron deficiency and reduced work capacity: a critical review of the research to determine a causal relationship. *J Nutr* 2001; 131: 676S-88S.
5. Halterman JS, Kaczorowski JM, Aligne CA, Auinger P, Szilagyi PG. Iron deficiency and cognitive achievement among school-aged children and adolescents in the United States. *Pediatrics* 2001; 107: 1381-6.
6. AlFadhli SM, Al-Awadhi AM, Alkhalidi D. Validity assessment of nine discriminant functions used for the differentiation between iron deficiency anemia and thalassaemia minor. *Trop Pediatr* 2007; 53: 93-7.
7. Jacobs A, Worwood M. Ferritin in serum: clinical and biochemical implications. *N Engl J Med* 1975; 292: 951-6.
8. Guyatt GH, Oxman AD, Ali M, Willan A, McIlroy W, Patterson C. Laboratory diagnosis of iron-deficiency anemia: an overview. *J Gen Intern Med* 1992; 7: 145-53.
9. Guyatt GH, Patterson C, Ali M, et al. Diagnosis of iron-deficiency anemia in the elderly. *Am J Med* 1990; 88: 205-9.
10. Dacie JV, Lewis SM. *Practical haematology*. 7th ed. Edinburgh: Churchill Livingstone, 1991; 236-41.
11. Khattak ID, Khattak ST, Khan J. Heterozygous beta thalassaemia in parents of children with beta thalassaemia major. *J Med Sci* 2006; 4: 103-9.
12. Al-Suliman A. Prevalence of beta-thalassaemia trait in premarital screening in Al-Hassa, Saudi Arabia. *Ann Saudi Med* 2006; 26: 14-6.
13. Yang Z, Chaffin CH, Easley PL, Thigpen B, Reddy VVB. Prevalence of elevated haemoglobin A2 measured by the capillary's system. *Am J Clin Pathol* 2009; 131: 42-8.
14. White JM, Richards R, Jelenski G, Byrne M, Ali M. Iron state in alpha and beta thalassaemia trait. *J Clin Pathol* 1986; 39: 256-9.
15. Economidou J, Augustaki O, Georgiopolou V, Vrettou H, Parcha S, Loucopoulos D. Assessment of iron stores in subjects heterozygous for beta-thalassaemia based on serum ferritin levels. *Acta Haematol* 1980; 64: 205-8.
16. Mehta BC, Pandya BG. Iron status of beta thalassaemia carriers. *Am J Hematol* 1987; 24: 137-41.