Original Article

Assessment of the Frequency of Beta Thalassemia Trait in Pregnant Anaemic Patients

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Background: To determine the frequency of beta thalassemia trait in anaemic pregnant women in order to assess the gravity of this disease and to identify high risk group, to prevent complications of thalassemia in newborn

Material & Methods: Three hundred forty (340) anaemic pregnant women, anaemia confirmed on complete blood count laboratory report, were selected. Their peripheral blood smear, RBC indices and haemoglobin electrophoresis were sent to assess for the presence of beta thalassemia trait.

Results: Frequency of beta thalassemia trait in pregnant anaemic women was found to be 5.3% using haemoglobin electrophoresis as gold standard for detection of beta thalassemia trait.

Conclusion: Study showed that 5.3% of pregnant anaemic women had beta thalassemia trait.

Keywords: Anaemia, Thalassemia and Pregnancy.

Introduction

Thalassemia is a type of inherited blood disorder which is the second most common cause of hypochromic microcytic anaemia in Pakistan. Iron deficiency anaemia and beta thalassemia trait are the most common forms of microcytic anaemia[1]. In beta thalassemia trait one of the beta globin genes is defective. The defect can be a complete absence of beta globin protein or reduced synthesis of beta globin protein. It is not a sex linked genetic trait.[2]

World Health Organization (WHO) has suggested that about 5% of the world population is carrier for different inherited disorders of hemoglobin. The percentage of beta thalassemia trait in pregnant women is 16.7 %.[3]

Detection of Beta thalassemia trait is important during pregnancy for the mother and fetus. The physiological stress of pregnancy[4] may exacerbate symptoms of thalassemia. Beta thalassemia is usually symptom free but anaemia is common in pregnancy. There is also possibility of an increased risk of neural tube defects.[5]

Beta thalassemia trait is significantly associated with oligohydromnios, intrauterine growth retardation, low APGAR score, congenital malformation and perinatal mortality. Patients with beta thalassemia minor are most likely to have cesarean deliveries.

Early diagnosis is of paramount importance in order to prevent birth of children with thalassemia.

Approximate 100,000 babies are born worldwide with severe forms of thalassemia each year; however this condition occurs more frequently in people of South East Asia because their prevalence and severity are population dependent.[7]

Diagnosis of Thalassemia is based on a complete blood count and red blood indices. Mean corpuscular volume (MCV) less than 80fl, mean corpuscular hemoglobin (MCH) less than 25pg were used for screening for thalassemia trait.[8] This was confirmed on Protein electrophoresis, the gold standard to detect thalassemia.

Inclusion Criteria

1) Pregnant woman.
2) Haemoglobin <10.0 gm/dl on complete blood count.

Exclusion Criteria

1) Multiple gestations determined by Ultra-sonography.
2) Co-morbidities of chronic illness like chronic renal failure, congestive cardiac failure determined by history/ previous investigations.
3) Bleeding disorder determined by history/ previous investigations.

Data Collection

Three hundred forty (340) anaemic pregnant women visiting Gynae Unit 3 were enrolled as per inclusion and exclusion criteria. Their peripheral blood smear and RBC,s indices was sent. Blood samples were sent for haemoglobin electrophoresis to same hematologist, who assessed for presence of beta thalassemia trait. Women, who had increased level of Hb A2 > 3.5%, determined on haemoglobin electrophoresis were considered to have beta thalassemia trait.woman, who had increased level of Hb A2s>% determined on hemoglobin electrophoresis were considered to have bete thalassemia trait.

Data Analysis

Data was analyzed using SPSS version 12.

Results

Three hundred and forty pregnant anaemic patients with Hb <10.0 gm/dl were selected at their 1st antenatal visit. Beta Thalassemia Trait was detected in 5.3% (18) patients and 322 (94.7%) patients were negative for this **(table 1).**

Table1: Distribution of subjects according to beta thalassemia trait presence n=340.

Thalassemia	Frequency	Percentage
Present	18	5.3%
Absent	322	94.7%
Total	340	100.0%

Majority of the patients with beta thalassemia had mild to moderate anaemia. 72% had Hb 7.1-9.0 gm/dl and 22% had Hb 9.1-10.0 gm/dl. Only a single patient had severe anaemia with Hb <7.0 gm/dl and no patient had Hb <5.0 gm/dl **(table 2).**

Table2: Distribution of beta thalassemia trait subjects according to Hb levels n=18.

Hb(gm/dl)	No. Of Patients	Percentage
<5.0	0	0.00%
5.1-7.0	1	5.55%
7.1-9.0	13	72.22%
9.1-10.0	4	22.23
Total	18	100.0

The age of patients with beta thalassemia trait ranged between 18 to 35 years with a mean age of 28.33 with SD of 5.66. So majority of the patients were less than 30 years of age.

Regarding the duration of pregnancy, maximum number of patients i.e 72% were detected during 2nd trimester and only 11% patients were detected during last trimester.While only 17% patients were detected during 1st trimester.

Table3: Distribution of thalassemia trait subjects according to duration of pregnancy n=18.

Trimester	Frequency	Percentage
1st	03	16.66%
2nd	13	72.23%
3rd	02	11.11%
Total	18	100.0%

Discussion

Beta Thalassemia is one of the most common inherited single gene disorder in Pakistan. It is highly prevalent in our part of world. Due to high prevalence, it has become important for health care providers to encourage people to utilize lab facilities for carrier and prenatal genetic testing[8-9].

The overall frequency of beta thalassemia trait was found to be 5.3% in this study. This means that among 340 anaemic pregnant women, 18 were beta thalassemia trait positive.

In comparison, a study conducted at Liaqat University Hospital Hyderabad reported the frequency of beta thalassemia trait to be 8.5% in pregnant women. In 17.6% cases husbands were also carriers. This would justify routine screening for thalassemia, to identify at-risk couples due to a risk as high as 25% for affected offspring[10].

Another study from India reported that the overall carrier frequency of beta thalassemia trait is 4.05%. The birth incidence calculated thereof for homozygous thalassemics would be 11,316 per year, which are added each year to the existing load of patients, which is very high.[11]

Screening of antenatal patients in a multiethnic community for beta thalassemia trait was done at Saint Marys Hospital, London.696 women with a MCV<83fl were investigated further by Hb electrophoresis. Beta thalassemia trait was diagnosed in 8% of microcytic women[12].

The frequency of beta thalassemia gene is population dependant and is most prevalent in African, Southeast Asian and Mediterranean descendants. In the African American population, 4% of people carry beta thalassemia gene[13].

All this data emphasizes the importance of screening for beta thalassemia trait in pregnant patients. Our study used a very simple and routine test to detect anaemia in pregnant patients. These were then subjected to further tests to detect beta thalassemia trait.

A shortcoming of this study was that, because of very strict selection criteria, only patients with anaemia were included in this study. This resulted in selection of those patients who already had anaemia detected on complete blood count test. That didn't allow us to see the incidence of anaemia among all pregnant women.

Second disadvantage may be cost because

haemoglobin electrophoresis was performed in all patients. This resulted in very high cost of the study and a burden on the health budget.

As, there is already a lot of research going on to develop a screening criteria in anaemic patients to check for beta thalassemia trait, MCV and MCH values have proved to be more sensitive and specific tools for this purpose. So, any anaemic pregnant patient with MCV <80fl and MCH <27pg should be checked for beta thalassemia trait, by performing more costly "haemoglobin electrophoresis"[14-17].

National policy/ guidelines should be in place all over Pakistan to ensure that all pregnant anaemic women are properly investigated for cause of anaemia. And, these patients are given proper advice regarding prevention and treatment of anaemia. Especially the patient with beta thalassemia trait, so that complications can be prevented in the newborn.

t the moment, it is difficult to predict the benefits of national guidelines on testing all anaemic population for presence of thalassemia trait. But, it is clear that it will definitely benefit those, who have the disease.

National and international guidelines make recommendations on the most appropriate ways in which genetic carrier screening programmes should be conducted[18].

Strict implementation of collective measures including carrier identification, genetic counseling and prenatal diagnosis are required for preventing beta thalassemia trait in Pakistan.

Conclusion

This study shows that anaemia is very common in pregnant women. So every pregnant woman should be screened for anemia and thalasemia on her first antenatal visit, since a considerable population of anaemic pregnant women have β-thalasemia trait (5.3%). This is important to prevent complications in the mother and fetus, as well as in the newborn in later life. Also the husband should be checked for thalasemia trait and proper counseling of the couple should be done.

Similar studies should be carried out and encouraged in all teaching hospitals and at a national level so that suitable control measures can be taken in Pakistan to lessen the burden of thalassemia.

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