Original Article

FREQUENCY OF BETA-THALASSEMIA TRAIT AMONG PREGNANT WOMEN IN THEIR LAST TRIMESTER WITH HYPOCHROMIC MICROCYTIC ANEMIA

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Abstract

Background: Beta-Thalassaemia is a public health issue. Generally, very few people know about it in detail. Due to ignorance, government bear tremendous financial cost to care for it.

Methods: This was a cross sectional observational study conducted in an outdoor patient department (OPD) of Gynecology and obstetrics of a Postgraduate Medical Institute at Peshawar, from August 2012 to Jan 2013. A Total of 56 women in their last trimester of pregnancy were included. Required information's were recorded on predesigned proforma as per objectives of the study.

Results: Most of the participants were of age 20 to 49 years with mean+ SD of 34+3.4years. Majority (87.5%) of the Red Blood Cell (RBC) count was equal or less than 4600000/cmm³. A few (12.5%) of the participants had their RBC count more than 4600000/cmm³. More than two-thirds (76.79%) had RDW (Red Cell Width Distribution SD) below 45%, while less (20.21%) had RDW more than 45%. The seven cases that had high RBC count underwent Hb-Electrophoresis where in two cases (3.2%) were reported to be ? -thalassaemia trait.

Conclusion: Majority of the patient were having low RBC count that needed further investigations i.e Serum Ferritin to confirm the iron deficiency. Similarly those who have had high RBC count when subjected to Hb-electrophoresis, showed B.thalassaemia trait. We suggest all those cases having RBC count more than 4600000/cmm³ and RDW-SD less than 45% may further be screened for thalassaemia trait, that may help to counselling families for future.

Keywords: Thalassemia, blood disorder, low and middle income country.

Introduction

Pakistan has a population of 180 million plus people. Five percent of our country population carry beta-thalassaemia gene, and 0.5 to 1% carry Hb S and Hb E type genes on haemoglobin. The infants affected by Beta trait (BTT) are at a rate of 1.3 per 1000 live births, and about 6,000 to 7,000 children are born annually with beta-thalassaemia. Unfortunately, majority of the people are unaware of the disease, and a minimum of these cases are correctly diagnosed. Out of the affected cases, 80% of the thalassemia patients die due cardiac failure and iron overload, and transfusion related risks. Thalassemia is controlled by parental screening for trait, and by haemoglobin electrophoresis before marriages in couples with high risk of having children with thalassaemia (1,2).

It is reported that 5000 to 9000 of the children with Beta-thalassemia born each year in Pakistan. There are 9.8 million carriers of thalassaemia with a BTT rate of 5-7% in the country (3,4). Another study reported that around eight million Pakistanis are suffering from Beta-Thalassemia trait. Modell and Darlinson reported it as

4.6% in Pakistan during 2008(5). In our scenario a weak infrastructure of health care facilities and very few screening facilities for screening of BTT and cultural trends of cousin marriages of approximately 40% in our societies leads to high risk of Beta-Thalassemia trait (6).

Pakistan is a country with racial mix and a nation of considerable genetic diversity. Cultural preferences are stronger for consanguineous marriage that is high risk factor for prevalence of recessive disorders like Beta-thalassemia. To let these children healthy Beta-thalassemia patients require monthly blood transfusions along with iron-chelation therapy. To treat one annual birth cohort of thalassemia we need 90,000 blood units and 22 million dollars for deferoxamine for chelation therapy (7).

A study on the screening for Beta thalassemia trait in pregnancy, concluded that carrier screening for thalassemia should be done on all pregnant women attending antenatal clinics (8). Another national study on the frequency of Beta thalassemia trait in pregnant women reported prevalence of 8.5%. 17.6% cases husbands were also trait (9).

Present study was conducted to determine frequency of b-thalassemia trait in pregnant women in their last trimester with hypochromic microcytic anemia.

Methodology

This cross sectional study conducted in gynecology and obstetrics department of MTI, Lady reading Hospital Peshawar, from Aug 2012 to Jan 2013. Samples were collected via convenient (Non-Probability) sampling. A total of 56 women in their third trimester were enrolled. Inclusion criteria were all patients irrespective of age, or cause for attending the OPD. All patients taking regular iron therapy for anemia, and post-natal cases with other gynecological disorders were excluded. Patients with RBC count >4600000/cmm³ were subjected to Hb-Electrophoresis for b-Thalassemia trait. Patients with RBC <4600000/cmm³ were not subjected to Hb-Electrophoresis. Blood samples taken from all patients. 3ml of venous blood was collected by vein-puncture under aseptic techniques using disposable syringes. The blood was added to EDTA tubes and mixed gently. Complete blood picture was with hematology analyzer taken in the laboratory of Institute of basic medical sciences (IBMS) Khyber Medical University.

For Hb-Electrophoresis, hemolysate was prepared adding carbon tetrachloride to remove all other globin other than HB and distilled water also added. Hb electrophoresis done using Wealtech, machine, USA. Current flow of machine was adjusted as per instruction of the manual of device. Cellulose acetate strip was used for application of hemolysate and different bands of hemoglobin were recorded for reporting. Similarly the preliminary information's of the women regarding, age, gravida, para, address, weight and socioeconomic status were recorded on the questionnaire. The data was analyzed using MS-Excel program for outcome.

Results

A total of 56 women in their third trimester presenting to Gynecology and obstetrics department of Lady reading hospital Peshawar. The age range of the patient was from 20 years to 49 years with mean age of 34+3.4 years (Table 1). RBC count of 87.50% was equal or less than 4600000/cmm³. Seven (12.5%) of the women had their RBC count more than 4600000/cmm³ (Table 2). 76.79% had RDW (Red Cell Width Distribution SD) below 45%, while 20.21% had RDW more than 45% (Table 3). The seven cases that had high RBC count underwent Hb-Electrophoresis where in two cases (3.2%) were reported to be b-thalassemia trait (Table 4).

Table 1. Age range of patients.

Age	Total	Percentage
23-32	29	51.79
33-42	20	35.71
43-52	6	10.71
53-62	1	1.79
Grand Total	56	

Table 2. Red Cell count of the respondents

RBC(/UL)	Total	PERCENTAGE
3010000-3809999	18	32.14
3810000-4609999	31	55.36
4610000-5409999	7	12.5
Grand Total	56	

Table 3. Red Cell Width Distribution range.

RDW-SD(f)	Total	Percentage
17-26	1	1.79
37-46	42	75.00
47-56	10	17.86
57-66	2	3.57
67-76	1	1.79
Grand Total	56	

Table 4. Hb. Electrophoresis Reports of the target cases.

HB. Electrophoresis Report (7/56)	Number of patients (7/56)	Percentage %
Normal	5	8.92
B- Thalassemia Trait	2	3.5
Total	7(56)	

Discussion

Thalassemia incidence globally is very high, nearly 30 million people are reported with beta thalassemia trait. BTT incidence differs from 3 to 17 % globally. This creates heavy burden on the society in terms of finance and social liabilities. One of the most effective strategy to reduce the incidence of thalassemia is the implementation of carrier screening program especially among the pregnant ladies, genetic counseling, prenatal diagnosis of BTT and disease and therapeutic abortion or termination of pregnancies for the affected fetuses (10). Early diagnosis of Beta-thalassemia trait is necessary for two reasons; one is differentiation from iron deficiency and second is for genetic counseling if there is affected fetus. In Pakistan the BTT rate is 8-10% (11). There are over 4000 thalassemia children born in Pakistan yearly and the available infrastructure and health care facilities are not sufficient to deal with this number of children. If we start screening for these cases about 70% of hidden carriers can be diagnosed just by knowing percentage of HbA₂ by electrophoresis. There is need for to raise public awareness and to encourage people to go diagnose for screening of trait and to diagnosis risk of thalassemia major off spring and to avoid trait to trait marriages as seen in consanguinity marriages (12). We don't discourage cousin marriages but we discourage trait to trait marriages.

The microcytic hypochromic anemia can be either beta thalassemia trait (Beta-TT) or iron deficiency anemia (IDA). In present study RBC count of 87.50% respondents was equal or less than 4600000/cmm³. Seven (12.5%) of the women had their RBC count more than 4600000/cmm³. These patients were then selected

for Hb-Electrophoresis. Another study also reported that RBC count is lowest in cases of Beta thalassemia major and highest in Beta thalassemia minor or trait (13). So these high counts candidates were subjected to electrophoresis test for know their status for Beta.Thalassemia trait.

We evaluated patients according to red blood cell (RBC) count and red blood distribution width indices. In our study 76.79% had RDW(Red Cell Width Distribution-SD) below 45%, while 20.21% had RDW more than 45% which is not specific for differentiating IDA from Beta thalassemia trait, so we recommend Serum Ferritin level along with Hb-Electrophoresis to differentiate between the two disorders. A raised RBC count > 5.0 106/uL is a common feature of Beta-Thalassemia trait. The high RBC count is one of the accurate indices of Beta Thalassemia trait. It provides best sensitivity for Beta Thalassemia trait (94.8%) (13). RBC count has been considered a valuable index for further investigating a patient for b-thalassemia trait (14). But few other studies report that RBC count > 5.0 × 106/BetaL helps you diagnosis of Beta Thalassemia trait. But in IDA the frequency of raised RBC count was 29.4% as compared to Beta-TT. This confirms that only RBC count alone was not a reliable indicator for distinguishing Beta-TT from IDA. Elevated RBC count may be associated with other conditions like erythrocytosis. Same findings reported by Aslan and Altay, they reported a high RBC count in 61% of cases with iron deficiency anemia(13-15).

In present study the seven cases that had high RBC count underwent Hb-Electrophoresis where in two cases (3.2%) were reported to be Beta Thalassemia trait out of the total sampling. Our finding correlates with other national studies where they reported a carrier rate of 5-7% in Pakistan (3,4). Another study reported that national Beta-thalassaemia trait rate of 4.6% in Pakistan5.

In conclusion we found that a high RBC count in pregnancy status must be evaluated further for thassemia trait by doing HB-electrophoresis or high performance liquid chromatography. We suggest all those cases having RBC count more than 4600000/cmm3 and RDW-SD less than 45% may further be screened for B-Thalassemia trait, as these are the target group where if we further proceed with Hb Electrophoresis, we can find out b-Thassemia trait well in time and can help to counsel families on marriage and pregnancy status. We again insist on statement that trait should not marry trait its always not cousin marriages.

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