ORIGINAL RESEARCH PAPER

Obstetrics & Gynaecology

"PREVALENCE OF BETA-THALASSEMIA TRAIT IN PREGNANT WOMEN ATTENDING ANTENATAL CLINIC"

KEY WORDS: Beta-thalassemia, iron deficiency anemia, High-Performance Liquid Chromatography (HPLC)

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ABSTRACT

INTRODUCTION- Iron deficiency anemia and thalassemia syndromes, especially beta thalassemia trait (BTT), are the two most commonly ccuring microcytic hypochromic anemias highly prevalent in countries like India. Iron deficiency anemia is the first most common cause of anemia in pregnancy, beta thalassemia trait is the second most frequent cause of anemia of pregnancy.

Patients with thalassemia trait shows an increased incidence of anemia during the second trimester of pregnancy.

Beta Thalassemia Trait (BTT) patients are usually asymptomatic and ignorant of their carrier state unless diagnosed by testing.

Screening for thalassemia can be done by measuring HbA2 levels

MATERIAL AND METHOD- This is a retrospective study. The data were collected from a clinic and associated pathology in Shivpuri and a private maternity hospital in Gwalior from 1st January 2019 to 30 June 2021. 94 patients were included in the study. All pregnant women between the age of 18-45 years and at any gestational age with hemoglobin level <10g/dL and microcytic hypochromic anemia (report of CBC) were included in the study. High-Performance Liquid Chromatography (HPLC) and serum ferritin report were collected.

RESULTS- In our study the most common age group in which the anemia was found between 19-35 years (80.85%). Most of the patients were primigravida (44.68%), between 26-37 wks of gestational age (82.97%), from a rural area (70.21%). Only 2 patients had a family history (2.12%) of beta-thalassemia and 6 patients had a history (6.38%) of blood transfusion.

In our study moderate anemia was found in 55.31% of cases followed by mild anemia 25.53% and severe anemia 19.14%. In our study iron deficiency anemia was found in 87.23% only minor Beta-thalassemia was found to be 4.25%, of the patient, and the coexistence of iron deficiency anemia and beta-thalassemia in pregnant women is 8.51%.

In our study total of **12 patient**s (4 cases of only minor beta-thalassemia and 8 cases of minor beta-thalassemia associated with IDA) The prevalence rate of beta-thalassemia in our study was 7.8%.

CONCLUSION- the recommendation is to diagnose beta-thalassemia in pregnant patients is essential to give proper treatment. The screening of pregnant patients prevents unnecessary parental iron therapy and iron overload in case of anemia. further studies are needed in this field and less expensive and accurate methods should come in place.

1. INTRODUCTION-

Iron deficiency anemia and thalassemia syndromes, especially beta thalassemia trait (BTT), are the two most commonly occuring microcytic hypochromic anemias highly prevalent in countries like India.[1] Iron deficiency anemia is the first most common cause of anemia in pregnancy, beta thalassemia trait is the second most frequent cause of anemia of pregnancy. During pregnancy 30-50% of women become anaemic and the main reason is iron deficiency anaemia. The oxygen carrying capacity of red blood cells becomes less and it cannot fulfill the body requirements in anaemia which is a pathological condition. The most common hematological disease in developing countries is Iron Deficiency Anaemia (IDA) affecting 30% of world population. People residing in Indian subcontinent, middle east and central Asia are more likely to get affected by iron deficiency anemia. 3.5%-10% people are affected by Beta Thalassemia Trait (BTT) in India.[2] Patients with thalassemia trait shows an increased incidence of anemia during the second trimester of pregnancy. This does not result in any disability and does not require blood replacement during pregnancy. No adverse effects were observed in any of the patients or in any of their infants. Investigation should be done for the presence of thalassemia in all patients in whom the racial origin indicates a high incidence of the disorder. Pregnancy indicates the use of oral iron therapy but at the same time, it is mandatory to guard against excessive and parenteral iron therapy which may lead to a state of iron overload in patients with thalassemia.[2,3]

Many studies have reported the occurrence of iron deficiency anemia in patients with beta thalassemia trait. In clinical practice and routine OPD the identification, treatment and possible prevention for iron deficiency anemia with iron supplementation is uncomplicated and simple while BTT is rountinely diagnosed only after failure of treatment for IDA during pregnancy or after delivery. Beta Thalassemia is the most common type of hemoglobinopathies transmitted by heredity. Beta Thalassemia Trait (BTT) patients are usually asymptomatic and ignorant of their carrier state unless diagnosed by testing.[4]

However, both alpha and beta thalassemia traits are the reason behind microcytic, hypochromic anaemia and are most frequently misdiagnosed as IDA. Thalassemia trait (or minor) should be diagnosed Whenever there is a microcytic anaemia that is not improving with iron therapy or when anaemia fore date the pregnancy, especially in a patient from a high risk ethnic group, thalassemia trait(or minor) should be diagnosed. Also, certain findings on the complete blood count (CBC) indices can suggest thalassemia trait such as microcytosis out of proportion to the anaemia along with a raised RBC count.[5]

Screening for thalassemia can be done by measuring HbA2 levels. Thalassemia traits are associated with a reduced mean corpuscular volume (MCV), reduced mean corpuscular haemoglobin (MCH), and a normal mean corpuscular haemoglobin concentration (MCHC). Of all the above mentioned markers, the most reliable marker is MCH.[6]

Another way of diagnosing BTT is by using Mentzer index. It was first described by Mentzer in the year 1973. It is calculated from the CBC report. Mentzer index value (MCV/RBC count) of less than 13 may represent thalassemia trait, and greater than 13 often indicates IDA.[5,6]

Recognition of women with thalassemia trait is important for genetic counseling as well as avoiding unnecessary iron replacement therapy. Diagnosis and identification of thalassemia trait in a pregnant woman is of clinical benefit to her and potentially significant genetic benefit for her family. The diagnosis of thalassemia trait may provide an explanation for a microcytic anaemia which will, in concert with a normal serum ferritin, rule out iron deficiency and allow the patient to reduce iron supplementation.[7]

The genetic benefit occurs if the woman's partner carries a thalassemia mutation. If beta thalassemia mutations are carried by both the parents, their children will be at risk for beta thalassemia major, which carries with it significant lifelong morbidity. Parents having alpha thalassemia mutations may have children with Hemoglobin H disease or Hemoglobin Barts Hydrops Fetalis. The diagnosis of thalassemia trait in a pregnant woman identifies a potentially at risk pregnancy. Patients with thalassemia trait and well stocked iron stores (ferritin > 70 ng/mL) should not receive iron supplementation. Although these patients are not dependant on transfusion therapy, still they may develop iron overload later in life, which can lead to pulmonary hypertension and thrombosis.[8]

Despite the fact that the goal of treatment in iron deficiency in pregnancy is to raise the hemoglobin to 10mg/dl or above, this will not be possible in many women with thalassemia trait. The ferritin level should be utilized to measure iron status, and the risks of iron deficiency in pregnancy must be balanced with the risks of unnecessary iron therapy. Signs of iron deficient erythropoiesis start at a serum ferritin level of 25-40 mcg/l.[9]

Therefore when screening for iron deficiency in these patients, it is reasonable to use a conservative value such as 40-50 mcg/l of serum ferritin as a goal for iron replacement since there will be ongoing iron requirements in pregnancy. Unthinkingly referring patients for intravenous iron when their hemoglobin levels are not rising with oral iron replacement subjects them to the potential of future iron overload, unnecessary cost, and inconvenience.[8] Provided that a multidisciplinary team is available, pregnancy is possible, safe and usually has a favourable outcome in patients with thalassemia.[10]

Patients suffering from thalassemia trait require no treatment or long term monitoring. They usually do not have iron deficiency, so iron supplementation is unlikely to improve their anemia. A number of studies suggest that haemoglobin levels may be low in the patients with BTT.Beta-thalassemia trait could minimally, but significantly, increase risk of low birth weight.[8] BTT is associated with an increased risk to both mother and baby. To be precise, there are the issues surrounding cardiomyopathy in the mother due to iron overload and the increased risk of fetal growth restriction (FGR). Furthermore, with around 9 months of little or no chelation, women with thalassaemia trait may develop new endocrinopathies: in particular, diabetes mellitus, hypothyroidism and hypoparathyroidism due to the increasing iron burden.[10]

2. MATERIAL AND METHOD-

This is a retrospective study. The data were collected from a clinic and associated pathology in Shivpuri and a private maternity hospital in Gwalior from 1st January 2019 to 30 June 2021. 94 patients were included in the study. All pregnant women between the age of 18-45 years and at any gestational age with hemoglobin level <10g/dL and microcytic hypochromic anemia (report of CBC) were included in the study. High-Performance Liquid Chromatography (HPLC) and serum ferritin report were collected.

EXCLUSION CRITERIA- All pregnant women with hemoglobin above 10g/dL, patients with multiple gestations, patients in which Red Blood cell distribution width (RDW) and mentzer index were used to differentiate IDA and BTT, and patients who received a blood transfusion in the last one month.

The patient's age, parity, residence, occupation, gestational age, and family history of beta-thalassemia or blood transfusion were studied. Serum ferritin and hemoglobin electrophoresis reports of all patients were studies.

All the data were analyzed using IBM, SPSS Ver. 20 software. Cross Tabulation and frequency distribution were used to prepare tables. Data are expressed as numbers, percentages, and mean.

3. RESULTS-

Table 1. Age & Parity wised distribution of patients of anemia.

Maternal age	Number	Percent
<19 years	4	4.25
19-35 years	76	80.85
≥ 36 years	14	14.89
Total	94	100
Parity		
0	42	44.68
1	30	31.91
2-4	18	19.14
>5	4	4.25
Total	94	100
Place of residence		
Urban	28	29.78
Rural	66	70.21
Total	94	100
Gestational age		
26-37 wks	78	82.97
37-39	12	12.76
≥40	4	4.25
Family history		
Family history of beta-thalassemia	2	2.12
Family history of transfusion in pregnancy	6	6.38
Total	8	100

In our study the most common age group in which the anemia was found between 19-35 years (80.85%). Most of the patients were primigravida (44.68%), most of the patients were between 26-37 wks of gestation (82.97%), most are from rural areas (70.21%). Only 2 patients had a family history (2.12%) of beta-thalassemia and 6 patients had a history (6.38%) of blood transfusion.

Table 2. Grades of anemia in the study population.

Grades of anemia	**Number**	**Percentage**
Mild (9-11 g/dl)	24	25.53
Moderate (7-9 g/dl)	52	55.31
Severe (<7 g/dl)	18	19.14
Total	**94**	**100**

In our study moderate anemia was found in 55.31% of cases followed by mild anemia 25.53% and severe anemia 19.14%.

Table 3. Type of anemia in the study population.

Types of anemia	**Number**	**percentage**
Iron Deficiency Anemia	82	87.23
Minor Beta-thalassemia	4	4.25
Combined IDA (HPLC positive with low ferritin)	8	8.51
Total	**94**	**100**

In our study, the prevalence of only minor Beta-thalassemia was found to be 4.25%. iron deficiency was found in 87.23% of the patient and the coexistence of iron deficiency anemia and beta-thalassemia in pregnant women is 8.51%.

In our study total of 12 patients (4 cases of only minor beta-

thalassemia and 8 cases of minor beta-thalassemia associated with IDA) The prevalence rate of beta-thalassemia in our study was 7.8%.

4. DISCUSSION-

In our study the most common age group in which the anemia was found between 19-35 years (80.85%)., most of the patients were between 26-37 wks of gestation (82.97%). Our study is in agreement with study of **Bushra zulifiqar et al**[11]. and **Sedigheh amooee et al**[12].

In our study most of the patients were primigravida (44.68%) In our study most of the patients were Primigravida, our study was not in agreement with one another study in which 50% of patients were multigravida[11].

In our study moderate anemia was found in 55.31% of cases followed by mild anemia 25.53% and severe anemia 19.14%. A similar result was found in the study of **Neelam Swaroop et al**[13]**.** (moderate anemia 44.7%, mild 35.0%, and severe 20.3) and **Neha Tyagi et al**[14]**.** (moderate anemia 60.30%, mild anemia 32.5%, and severe anemia 7.23%).

In our study iron deficiency anemia was found in 87.23% which was similar to the study of **Neelam Swaroop et al**[13] (71.4%), In the study of **Bushra zulifiqar et al**[11], (91.4%) and **Sinha M et al**[15], (65%).

In our study, the combined anemia (IDA+BTT) was 8.51%, which was higher than the study of **Bushra zulifiqar et al**[11].

In our study the prevalence of miner Beta-thalassemia was found to be 7.8% which was comparable with the study of **Neelam Saroop et al**[13] (4.1%), **Bushra Zulfiqar et al**[11] (BTT 2.8%, Combined IDA+BTT 5.7%), **Sinha M et al**[15] (5.8%). Other studies in the antenatal period reported incidence of beta-thalassemia trait 8.45% and 3%[16,17] According to WHO Beta-thalassemia trait varies between 1.7-9% worldwide. The incidence of thalassemia is about 7% in Chinese.[11] In BangladeshBeta-thalassemia miner were 21.3%[18]

5. CONCLUSION-

Our study concluded that although iron deficiency anemia is the leading cause of anemia (Microcytic hypochromic anemia) minor beta-thalassemia is also contributes to about 7.8% of cases. The coexistence of iron deficiency anemia and beta-thalassemia in pregnant women is about 8.51% in our study. the recommendation is to diagnose beta-thalassemia in pregnant patients is essential to give proper treatment. The screening of pregnant patients prevents unnecessary parental iron therapy and iron overload in case of anemia. further studies are needed in this field and less expensive and accurate methods should come in place.

6. REFERENCES-

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