

# Frequency of Betathalassemia Trait in Pregnant Females

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## ABSTRACT

**Aim:** To study frequency of Thalassemia trait in pregnant females presenting with hypochromic microcytic anemia in first trimester.

**Study design:** A descriptive Cross sectional study.

**Place and duration of study:** Ghurki Teaching Trust Hospital during May 2016 till April 2017.

**Methods:** This study was conducted in Ghurki Teaching Trust Hospital during May 2016 till April 2017. 120 pregnant patients presenting in outpatient department with hypochromic microcytic anemia for antenatal checkup in first trimester included in this study. Complete blood counts, Serum Ferritin and Hemoglobin (Hb) electrophoresis were done in all case. Beta-thalassemia trait was diagnosed in case of Hb A2 level >3.5%.

**Results:** Pregnant women age ranged from 17 to 42 with mean  $26.4 \pm 4.3$  years. Hemoglobin ranges from 5 to 10.5g/dl with mean  $8.5 \pm 0.5$ g/dl. With Hemoglobin electrophoresis, 9(7.5%) women found to have thalassemia trait.

**Conclusion:** Screening for thalassemia should be mandatory for all antenatal women presenting with hypochromia and microcytosis to prevent thalassemia major in successive generations.

**Keywords:** Thalassemia trait, Hemoglobin electrophoresis.

## INTRODUCTION

The most common causes of hypochromia and microcytosis include iron deficiency anemia followed by beta thalassemia trait<sup>1</sup>. Beta-thalassemia is a heterogenous genetic disorder caused by mutations in genes responsible for beta chains production causing diminished or absent beta chains leading to decreased haemoglobin in erythrocytes, reduced erythropoiesis and anemia<sup>2</sup>. In thalassemia, inheritance pattern is mostly autosomal recessive. Thalassemia has equal gender distribution in males and females and incidence is reported to be 4.4 of 10,000 live births worldwide<sup>3,4</sup>. Iron deficiency anemia is the most prevalent type of anemia in pregnant women in Pakistan<sup>5,6</sup>. In Iron deficiency anemia, the production of Hb-A<sub>2</sub> is also affected causing reduction in Hb-A<sub>2</sub> levels in patients with thalassemia trait<sup>7</sup>.

Almost 9.8 million people in Pakistan are the carriers of Beta thalassemia with 5-7% estimated carrier rate. 5000-9000 children are born every year with thalassemia major, of which only 10-15% receive optimal treatment<sup>8</sup>. The cost of treating properly one thalassemic child amounts to Rs. 6000\$ annually<sup>9</sup>. The only cure which is available today is bone marrow transplantation, which is beyond the affordability of most of the patients. Thus, the emphasis should be on the prevention of such births. The most effective approach to minimize the disease burden in society is application of a comprehensive screening program to identify carriers and providing counseling regarding disease. Thalassemia has high prevalence in Pakistan and screening is an important way to prevent its inheritance in successive generations.

The objective of this study was to determine the prevalence of  $\beta$  thalassemia trait in pregnant women presenting with hypochromic microcytic anemia in Pakistan.

## METHOD

Inclusion criteria was all pregnant women of any parity and age with hemoglobin value of less than 10.5g/dl presenting in first trimesters with hypochromia and microcytosis blood picture. Exclusion criteria was already diagnosed cases of haemoglobinopathies. Of these subjects, 120 pregnant females with microcytic hypochromic blood picture were included in the study. The written consent was taken in all cases. Detailed history and clinical examination were done in all women. Complete blood count was performed on blood collected in EDTA (anticoagulant). The hematological parameters, hemoglobin along with RBC indices (PCV, MCV, MCH and MCHC) were analyzed by three-part hematology analyzer Sysmex KX2100. Peripheral blood smears were also made. Serum ferritin were done on all the samples. Serum ferritin level of <15 $\mu$ g/dl was taken as cut off for diagnosis of iron deficiency anemia. Serum ferritin was done by the principle of chemiluminescence immunoassay. Cellulose acetate Hb electrophoresis was used for thalassemia carrier screening.

Women with HbA<sub>2</sub> (>3.5%) were labelled as carriers of beta thalassemia. Data was recorded on a pre-designed proforma. The collected data was organized and SPSS 20 was used for statistical analysis. Quantitative variables were calculated by Mean and standard deviation values. Frequencies and percentages were presented for qualitative variables.

## RESULTS

Pregnant women age ranged from 17 to 42 with mean  $26.4 \pm 4.3$  years. Hemoglobin ranges from 5 to 10.5 g/dl with mean  $8.5 \pm 0.5$ g/dl, Mean value for mean corpuscular volume (MCV) was  $64.32 \pm 1.56$ fl and for mean corpuscular hemoglobin (MCH) was  $21 \pm 0.56$  g/dl.

Among 120 women 20(17%) had mild anemia (Hb 10–11 gm%), 85(71%) moderate anemia (Hb 7–10gm%), 15(12%) severe anemia (Hb <7gm%). With Hemoglobin electrophoresis, 9(7.5%) women found to have thalassemia trait with Hb A<sub>2</sub> more than 3.5%. In women with mild anemia group 2(20%), in women with moderate anemia

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group 4(4.7%) and in severe anemia group 20%(n=3) women were found to have thalassemia trait. Three females had thalassemia with iron deficiency anemia. 111(92.5%) patients diagnosed with iron deficiency anemia.

Fig 1: Distribution of patients depending upon severity of anemia.

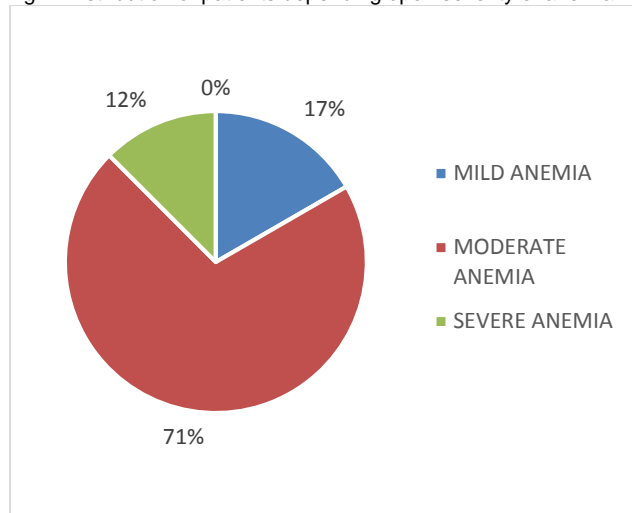
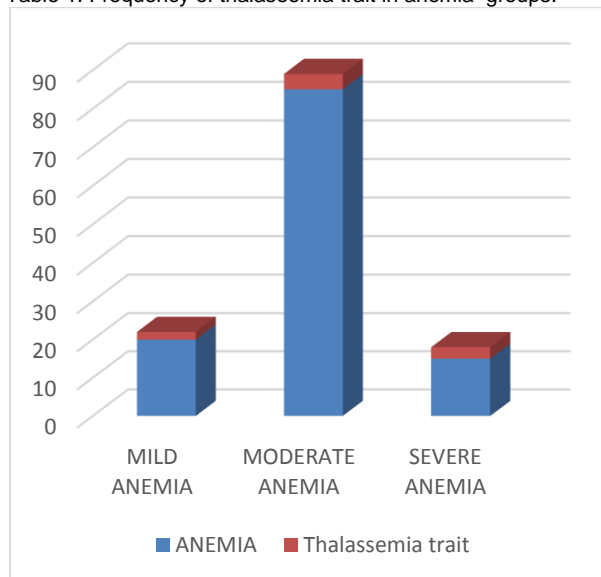


Table 1: Frequency of thalassemia trait in anemia groups.



## DISCUSSION

Thalassemia is characterized by decreased or absent production of one or more globin chains. Severity of disease depends upon affected globin chains. Beta Thalassemia is considered to be one of the most common single gene disorders in world. Majority of thalassemia major children are born in underdeveloped countries due to lack of national preventive program<sup>10,11</sup>.

If both parents are carriers, there are 50% chances of a child being born as  $\beta$  thalassemia minor, 25% chances of a child inheriting  $\beta$  Thalassemia major and 25% chances for a normal healthy child.  $\beta$  thalassemia minor patients are

usually asymptomatic present with only mild anemia. Physicians mostly mistakenly diagnose it simply iron-deficiency anemia and suggest hematinic. Blood picture shows mild anemia, hypochromia and microcytosis. Hemoglobin Electrophoresis can be used to detect carrier status in most of cases. Few carriers with silent mutations and normal hematological profile will require DNA analysis.<sup>12</sup> Thalassemia carriers in case of iron deficiency result in falsely low or borderline A2 levels. Thalassemia is a genetic disorder causing huge financial and psychological trauma to society, its transmission in subsequent generations can be reduced by preventive measures like carrier screening in early pregnancy<sup>13</sup>.

In our study 7.5% pregnant anemic patients were diagnosed as  $\beta$  thalassemia carriers. It was most frequent in patients with moderate anemia. Mean hemoglobin level was recorded as  $9.43 \pm 0.40$  g/dl. Higher frequencies of thalassemia trait in pregnant women have been reported in our region.

Hafeez M et al in their study in Lahore observed that 53.1% of the pregnant ladies presented with the diagnosis of beta thalassemia trait<sup>14</sup>. Sarda H et al, studied 209 pregnant anemic women and beta thalassemia trait was found in 108(51.6%)<sup>15</sup>.

Similarly studies showing results in accordance with our study have also been reported in our region. Kulkarni P et al in 2015, studied 210 pregnant women and 18(8.5%) were thalassemia carriers<sup>16</sup>. Qamar-ur-Nisa et al study on pregnant women shows 8.5% carrier rate<sup>17</sup>.

Sukrat et al carried out a study in pregnant females in 2010 including anemic and non-anemic pregnant females. The 39.7% females were found to be thalassemia carrier in the anemic pregnant cases and frequency of thalassemia trait was 24.4% in the other group.<sup>18</sup> Sur D et al in 2016, screening of thalassemia was done in 1,083 women and prevalence of thalassemia carriers was 4.61%<sup>19</sup>.

In Pakistan, lower literacy rate, higher trend of consanguineous marriages and lack of implementation of preventive program is resulting in substantive number of thalassemia cases born every year. The effective strategy to deal with magnitude of thalassemic problem is prevention. Increased awareness campaigns and Premarital screening of couples should be offered. This strategy has been adopted effectively in other parts of world like in Cyprus resulting in no thalassemia major birth between 2002 and 2007<sup>20</sup>. Saudi Arabia also successfully reduced the prevalence of beta-Thalassemia by 70% in six years<sup>21</sup>.

In our study, higher prevalence of thalassemia recommends effective implementation of screening in all antenatal patients. Screening should be done before marriages or early in the pregnancy to prevent thalassemia major in successive generations.

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