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Frequency of Thalassemia Minor and Iron Deficiency Anemia in Pregnant Women Presenting with Anemia

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ABSTRACT

Background: Anemia in pregnancy is a significant public health concern, affecting maternal and fetal health outcomes. Among its causes, iron deficiency anemia and thalassemia minor are critical conditions requiring early diagnosis and management. Identifying the prevalence and associated factors of these anemia types is essential for improving antenatal care and preventing complications. **Objective:** To determine the prevalence of thalassemia minor and iron deficiency anemia in pregnant women with anemia. **Study Design:** Cross-sectional study. **Duration and Place of Study:** This study was conducted from September 2024 to December 2024 in the Obstetrics and Gynaecology Department of Saidu Group of Teaching Hospitals, Swat. **Methodology:** A total of 130 pregnant women aged 18–40 years with a confirmed singleton pregnancy beyond 12 weeks of gestation and diagnosed with anemia were enrolled using a non-probability consecutive sampling method. Patients with hypertensive disorders, gestational diabetes, autoimmune diseases, or hematological disorders other than anemia were excluded. Demographic and clinical data, including hemoglobin (Hb) concentration, serum ferritin, and mean corpuscular volume (MCV), were recorded. **Results:** The mean age of participants was 30.21 ± 3.25 years, with a gestational age of 17.15 ± 3.07 weeks. The prevalence of iron deficiency anemia was 27.7%, while 6.2% of participants were diagnosed with thalassemia minor. **Conclusion:** Thalassemia minor and iron deficiency anemia are prevalent among anemic pregnant women, with distinct demographic and clinical associations.

INTRODUCTION

Anemia is a physiologic condition of pregnancy that occurs due to the reduction in the number of red cells or hemoglobin level, causing a reduced oxygen-carrying capacity of the blood.[1] Increased iron and other nutrient demand during pregnancy is due to the rise in maternal blood volume and requirements of the fetus. Anemia of pregnancy occurs as a result of many etiologic factors such as diet-deficiencies, chronic diseases, and genetic abnormalities.[2] Pregnancy anemia is also linked with harmful effects to mother and child such as prematurity, low birth weight, and increased maternal morbidity.[3] Early detection of anemia in its early stage is very crucial so that the maximum advantage can be derived by both the child and the mother.

Iron deficiency anemia (IDA) is the most prevalent form of anemia in pregnancy, resulting largely from inadequate dietary supply or absorption of iron.[4] The body requires more iron in pregnancy to sustain higher blood volume and fetal development. With a deficiency of dietary supply to support such higher demands, body reserves of iron get depleted, leading to IDA.[5] The symptoms of IDA include weakness, tiredness, pallor, and shortness of breath.[6] The diagnosis is established with blood tests to determine hemoglobin concentration and ferritin, a marker of body reserves of iron. The treatment is in the form of intravenous or oral iron supplements in conjunction with dietary modifications to supply more dietary sources of heme and non-heme iron.[7] Adequate treatment of IDA is paramount to prevent complications in pregnancy.

Thalassemia minor is a genetic blood disorder that also presents in pregnancy in the form of anemia, though it is less common in nature compared to iron deficiency anemia.[8] Thalassemia minor is a condition in females when one of their genes responsible for producing hemoglobin is mutated, resulting in mild anemia due to a reduction in the synthesis of normal hemoglobin.[9] Thalassemia minor is not treatable with iron supplements, and inappropriately administered iron can lead to iron overload. Thalassemia minor in pregnant women requires careful control and monitoring, as such women are more likely to be at risk of complications such as intrauterine growth restriction, gestational diabetes, and hypertension.[10] Genetic counseling is essential in such women, especially if their partner is also a carrier, to assess the risk of having a child afflicted with thalassemia major, a more severe form of the disease.[11] Proper antenatal checkups in conjunction with consultation with hematologists can control the condition and avert complications.[12]

A study conducted by Susanti AI et al. reported that the prevalence of thalassemia minor among pregnant women with anemia was 5.7%, while iron deficiency anemia was found in 21.9% of the cases.[13]

This research is necessary in order to have a better idea of the incidence and effect of various forms of anemia, including thalassemia minor and iron deficiency anemia, among pregnant women. With the serious health consequences for both mother and child, it is important to know the exact etiology of anemia among pregnant women. The results could inform specific interventions, enhance prenatal care, and assist in the mitigation of the risk of pregnancy-related complications from anemia. In addition, knowledge of the distribution of these conditions may be used to inform public health policy and resource allocation for maternal health.

METHODOLOGY

This cross-sectional study was conducted between September of 2024 to December of 2024 in the Obstetrics and Gynaecology Department of Saidu Group of Teaching Hospitals in Swat. There were a total of 130 participants, of whom the sample size was calculated using WHO sample size software, at a 95% confidence level, a 3% margin of error, and a hypothesized frequency of thalassemia minor of 5.7% in anemic pregnant women.[13] Participants were recruited via a non-probability consecutive sampling method. Inclusion was females aged between 18 to 40 years old with a confirmed ultrasound singleton pregnancy, a gestation of more than 12 weeks via the last menstrual period, and having anemia. Exclusion was females with a history of hypertensive complications of pregnancy, gestational diabetes, intrahepatic cholestasis of pregnancy, aplastic anemia, idiopathic thrombocytopenic purpura, or autoimmune disease. Demographic and clinical details were collected, i.e., age, gestational age, parity, body mass index (BMI), education, socioeconomic status, residential status, profession, and concentration of hemoglobin (Hb). Informed written consent was taken after hospital ethics committee approval was given to all participants. Venous blood was sampled and stored at a temperature of 2–8°C prior to transportation to the laboratory for analysis. Diagnoses of iron deficiency anemia and thalassemia minor were confirmed. In particular, a diagnosis of iron deficiency anemia was confirmed when Hb was <11 g/dL, serum ferritin was <15 µg/L, and mean corpuscular volume (MCV) was <82 fL. Thalassemia minor was diagnosed on chronic hemolytic anemia, low Hb, low MCV, high red cell distribution width (RDW), and confirmed using Hb electrophoresis showing absent HbA1, high HbA2 (above 3.5%), and HbF (above 90%).

IBM SPSS software version 26 was utilized to process the data.

RESULTS

The mean ± standard deviation (SD) values for the continuous variables, based on Table-I, include: age (30.21 ± 3.25 years), gestational age (17.15 ± 3.07 weeks), parity (1.73 ± 0.99), BMI (25.03 ± 1.75 kg/m²), duration of anemia (7.46 ± 3.88 months), serum ferritin levels (14.86 ± 3.18 µg/L), MCV (81.36 ± 1.87 fL), and hemoglobin (HB) level (10.67 ± 0.40 g/dL). Regarding educational background, 45.4% of the participants were uneducated, 27.7% had primary education, 20.8% had secondary education, and 6.2% had higher education. Socioeconomically, 45.4% of the participants were from low-income backgrounds, 48.5% from middle-income, and 6.2% from rich backgrounds. Additionally, 73.1% of the women lived in rural areas, while 26.9% resided in urban areas. The majority of the participants (86.9%) were housewives, and 13.1% had jobs.

Table I

Patient Demographics (n=130)

Demographics		Mean ± SD
Age (years)		30.207±3.25
Gestational Age (weeks)		17.153±3.07
Parity		1.730±0.99
BMI (Kg/m²)		25.031±1.75
Duration of Anemia (months)		7.461±3.88
Serum Ferritin Levels (µg/L)		14.861±3.18
MCV (fL)		81.361±1.87
HB Level (g/dL)		10.673±0.40
Education	Uneducated n (%)	59 (45.4%)
	Primary n (%)	36 (27.7%)
	Secondary n (%)	27 (20.8%)
	Higher n (%)	8 (6.2%)
Socioeconomic Status	Low n (%)	59 (45.4%)
	Middle n (%)	63 (48.5%)
	Rich n (%)	8 (6.2%)
Residential Status	Rural n (%)	95 (73.1%)
	Urban n (%)	35 (26.9%)
Profession	House wife n (%)	113 (86.9%)
	Job n (%)	17 (13.1%)

As shown in Table-II, the prevalence of iron deficiency anemia was 27.7% (36 women), while 6.2% (8 women) were diagnosed with thalassemia minor.

Table II

Prevalence of Thalassemia Minor and Iron Deficiency Anemia in Pregnant Women

Anemia Types	Frequency	% age
Iron Deficiency Anemia	36	27.7%

Thalassemia Minor	8	6.2%

Table-III presents the association of these conditions with demographic factors. Iron deficiency anemia was significantly associated with age ($p < 0.001$), BMI ($p = 0.012$), and residential status ($p = 0.047$), while no significant association was found with socioeconomic status ($p = 0.061$). Thalassemia minor showed significant associations with BMI ($p = 0.007$) and socioeconomic status ($p = 0.010$), with no significant association with age ($p = 0.725$) or residential status ($p = 0.107$).

Table III

Association of Thalassemia Minor and Iron Deficiency Anemia with Demographic Factors

Demographic Factors		Iron Deficiency Anemia		p-value
		Yes n(%)	No n(%)	
Age (years)	18-30	31 (41.9%)	43 (58.1%)	<0.001*
	>30	5 (8.9%)	51 (91.1%)	
BMI (Kg/m2)	≤25	13 (18.6%)	57 (81.4%)	0.012
	>25	23 (38.3%)	37 (61.7%)	
Socioeconomic Status	Low	21 (35.6%)	38 (64.4%)	0.061*
	Middle	15 (23.8%)	48 (76.2%)	
	Rich	0 (0%)	8 (100%)	
Residential Status	Rural	31 (32.6%)	64 (67.4%)	0.047*
	Urban	5 (14.3%)	30 (85.7%)	

Demographic Factors		Thalassemia Minor		p-value
		Yes n(%)	No n(%)	
Age (years)	18-30	4 (5.4%)	70 (94.6%)	0.725*
	>30	4 (7.1%)	52 (92.9%)	
BMI (Kg/m2)	≤25	8 (8.4%)	62 (88.6%)	0.007*
	>25	0 (0%)	60 (100%)	
Socioeconomic Status	Low	8 (13.6%)	51 (86.4%)	0.010*
	Middle	0 (0%)	63 (100%)	
	Rich	0 (0%)	8 (100%)	
Residential Status	Rural	8 (8.4%)	87 (91.6%)	0.107*
	Urban	0 (0%)	35 (100%)	

***Fischer Exact Test**

Lastly, Table-IV provides the correlations between various factors. Notable findings include a significant positive correlation between age and parity ($r = 0.619$, $p < 0.001$), a strong negative correlation between the duration of anemia and serum ferritin level ($r = -0.662$, $p < 0.001$), and a significant positive correlation between MCV and hemoglobin levels ($r = 0.895$, $p < 0.001$). Additionally, iron deficiency anemia had a significant positive correlation with age ($r = 0.220$, $p = 0.012$) and thalassemia minor had a negative correlation with serum ferritin ($r = -0.158$, $p = 0.072$).

Table-IV

Correlation Analysis of Demographic and Clinical Factors with Anemia Types in Pregnant Women

		Age	BMI	Duration of Anemia	Serum Ferritin Level	MCV	HB Level	Iron Deficiency Anemia	Thalassemia Minor
Age	Pearson Correlation	1	.308**	-0.126	0.117	0.084	0.096	.220*	-0.063
	Sig. (2-tailed)		0	0.153	0.184	0.34	0.276	0.012	0.479
BMI	Pearson Correlation	.308**	1	0.007	-.238**	-.236**	-0.154	-.220*	0.078
	Sig. (2-tailed)	0		0.937	0.006	0.007	0.081	0.012	0.378
Duration of Anemia	Pearson Correlation	-0.126	0.007	1	-.662**	-.538**	-.736**	-.602**	-.598**
	Sig. (2-tailed)	0.153	0.937		0	0	0	0	0
Serum Ferritin Level	Pearson Correlation	0.117	-.238**	-.662**	1	.894**	.850**	.818**	0.15
	Sig. (2-tailed)	0.184	0.006	0		0	0	0	0.088
MCV	Pearson Correlation	0.084	-.236**	-.538**	.894**	1	.895**	.719**	.187*
	Sig. (2-tailed)	0.34	0.007	0	0		0	0	0.033
HB Level	Pearson Correlation	0.096	-0.154	-.736**	.850**	.895**	1	.712**	.365**
	Sig. (2-tailed)	0.276	0.081	0	0	0		0	0
Iron Deficiency Anemia	Pearson Correlation	.220*	-.220*	-.602**	.818**	.719**	.712**	1	-0.158
	Sig. (2-tailed)	0.012	0.012	0	0	0	0		0.072
Thalassemia Minor	Pearson Correlation	-0.063	0.078	-.598**	0.15	.187*	.365**	-0.158	1
	Sig. (2-tailed)	0.479	0.378	0	0.088	0.033	0	0.072	

** Correlation is significant at the 0.01 level (2-tailed).
* Correlation is significant at the 0.05 level (2-tailed).

DISCUSSION

The results of the analysis found that iron deficiency anemia was more common (27.7%) than thalassemia minor (6.2%) in the group that was studied. This is in keeping with the general observation that one of the most common types of anemia during pregnancy is iron deficiency anemia due to increased needs of iron for fetal development and expansion of blood in the mother. In contrast, a lower incidence of thalassemia minor is accounted for by its genetic nature in that many of those suffering often carry a mild condition of anemia that is not diagnosed except when screened for specifically.

The significant relationship between age ($p < 0.001$) and iron deficiency anemia is explained by younger pregnant women, especially those younger than 30, having a higher demand for iron due to their high needs of growth and development, making them more susceptible to deficiency in iron. The relationship between BMI ($p = 0.012$) shows that more women who are overweight would be having a higher response to inflammation or dysregulation of their metabolism of iron, hence having lower reserves of iron. The relationship between residential status ($p = 0.047$) shows that there is a healthcare and nutrition problem, with fewer resources in rural areas to manage nutritional deficiencies.

In contrast, BMI correlation ($p = 0.007$) with thalassemia minor is explained by body composition's effect on blood hemoglobin in that patients with a higher BMI would be more likely to be more severely anemic due to the increased need for oxygen transport. Socioeconomic status correlation ($p = 0.010$) with thalassemia minor is explained by higher disease incidence in lower socioeconomic groups in whom genetic counselling or health services would be more limited.

Our study, conducted in 130 pregnant women, found a prevalence of 6.2% of thalassemia minor, a result that is in keeping with a number of studies that have been done in Pakistan and other regions. In Samreena Akhter Chohan et al.'s work[14] for example, a diagnosis of the beta-thalassemia trait was established in 9% of anemic pregnant women, a higher percentage in cases of moderate anemia (4%) and in cases of severe anemia (3.5%). In Asma Mustafa et al.'s work[15] also, 4.9% of subjects presented with the trait, a percentage slightly lower than our work's but in keeping with that of other studies. The similarity of these results shows that the beta-thalassemia trait is a common condition in cases of pregnant women with anemia, a reflection of a wide problem in regions of high consanguineous marriage incidence.

Our results also agree with those of Maimoona Qadir et al.[16] who found that 56.7% of pregnant women with anemia in Peshawar were carriers of the beta-thalassemia trait. The greater prevalence in their study can be explained, however, considering their greater number of participants (194 participants) and possibly regional differences, including genetic vulnerabilities in the Khyber Pakhtunkhwa province. Furthermore, our study showed that the majority of our group of women (45.4%) were of low socioeconomic status, and this also may be an explanation for the greater prevalence in our study and in comparable research, in that healthcare services and genetic counseling might be more limited in these situations.

A comparison of our work to that of Hamzullah Khan et al. [17] also indicates that there is similarity in results. In their work, two females (3.2%) carried the beta-thalassemia trait after high RBC count screening, in contrast to our work in which the trait was carried in 6.2% of the population. The difference in prevalence can be attributed to our large sample size and that all females in our work underwent screening for anemia types and their demographic variables. The work also indicates that screening based on RBC count, RDW, and further electrophoresis (for subjects having more than 4,600,000/cmm³ of RBC counts) is of paramount importance, just like in Khan et al.'s work.[17]

Also, Kiran Iqbal et al. [18] in Ghurki Teaching Trust Hospital reported that 7.5% of pregnant women with hypochromic microcytic anemia were carriers of beta-thalassemia. This is consistent with our findings, suggesting that beta-thalassemia is an important cause of anemia in pregnant women in the area. Both studies highlight the importance of early diagnosis to prevent thalassemia major in future generations. Furthermore, the recommendation of routine screening is consistent with our study, in that individuals with elevated RBC counts and RDW of less than 45% should be investigated for thalassemia.

We also observed that iron deficiency anemia (IDA) was also prevalent in our population, in which 27.7% of women suffered from iron deficiency anemia. This is in conformity with Hamzullah Khan et al.'s studies [17] and Samreena Akhter Chohan et al.'s studies [14] in which it was observed that IDA was a co-morbid condition prevalent in women suffering from beta-thalassemia trait. As a point in case, it was observed that 92.5% of subjects in Iqbal et al.'s study [18] suffered from IDA, and 63.7% of women in Chohan et al.'s studies [14] also suffered from mild to severe anemia. In our studies, it was also observed that women suffering from beta-thalassemia had a mean of 14.86 ± 3.18 µg/L of serum ferritin, indicating that even though thalassemia minor is a cause of anemia, many of these women also suffer from deficiency of iron.

Finally, in Wang et al.'s paper [19] IDA was more common in women having alpha-thalassemia and beta-thalassemia minor genotypes, careful supervision of iron supplement in women having thalassemia was recommended in the study. This is of concern in view of our results and that of previous studies indicating that despite the importance of iron supplement in managing anemia, women having a thalassemia trait can be vulnerable to iron overload if not monitored correctly.

The overlap of thalassemia trait with iron deficiency anemia also underscores the importance of a more integrated diagnostic approach, i.e., hemoglobin electrophoresis, to accurately diagnose and treat these two entities. The observed prevalence rates combined with demographic factors point towards a strong need for more genetic counseling programs and increased awareness to restrict the burden of thalassemia to generations to be born in the future.

However, our work is not without limitations. The work was a one-center one in a given geographic location, a limitation that would restrict its applicability to other regions of the world that differ in their demographic and genetic structure. The sample was also small in size compared to other studies, a limitation that would affect precision of estimates. The work also did not control for the long-term effects of such a pregnancy or determine the impact of different methods of managing anemia. Such studies in multiple centers using large samples would be helpful to crosscheck these results and provide more information on the interaction between thalassemia trait and iron deficiency anemia in pregnancy.

CONCLUSION

Our study reveals that beta-thalassemia trait is prevalent in pregnant women suffering from anemia, suggesting regular screening in high-risk groups is required. The findings support detection of thalassemia trait in conjunction with iron deficiency anemia to allow accurate diagnosis and treatment. Early detection through screening and genetic counseling can significantly reduce complications in future pregnancies, particularly in high consanguinity areas. This underscores the key role of total anemia screening in preventing thalassemia major and improving mother and perinatal health outcomes.

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Author Contributions

The authors have made significant contributions to this manuscript, as outlined below.

Dr. Maria Nawaz was responsible for the study's conceptualization, drafting the article, and gathering hospital data.

Dr. Parveen Naveed played a key role in developing the article, conceptualizing the study, and performing data analysis and interpretation.

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