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FREQUENCY OF BETA THALASSEMIA TRAIT IN ANEMIC PREGNANT WOMEN OF A TERTIARY CARE SETUP OF PAKISTAN

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Abstract:

Introduction: *The most common causes of hypochromia and microcytosis include iron deficiency anemia followed by beta thalassemia trait.* ***Objectives of the study:*** *The main objective of the study is to find the frequency of beta thalassemia trait in anemic pregnant women of a tertiary care setup of Pakistan.* ***Material and methods:*** *This cross-sectional study was conducted in Pakistan Institute of Medical Sciences, Islamabad (PIMS) during January 2019 to December 2019. All pregnant women age 18-45 years and at any gestational age having Hb% level 7-10gm/dl were included in the study. Non pregnant patient and pregnant women with haemoglobin 10-11gm/dl were excluded.* ***Results:*** *The data was collected from 120 patients. The age of the women included ranges from 18-45 years Table 1 show the presence of moderate anemia in relation with the age of pregnant female in which most of the patients 75 (53%) belongs to 18-30 years of age group.* ***Conclusion:*** *It is concluded that Iron deficiency is the leading factor for anemia found in this study, but thalassemia trait also a significant contributing factor.*

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INTRODUCTION:

The most common causes of hypochromia and microcytosis include iron deficiency anemia followed by beta thalassemia trait. 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. In thalassemia, inheritance pattern is mostly autosomal recessive [1]. Thalassemia has equal gender distribution in males and females and incidence is reported to be 4.4 of 10,000 live births worldwide. Iron deficiency anemia is the most prevalent type of anemiain pregnant women in Pakistan. In Iron deficiency anemia, the production of HbA2 is also affected causing reduction in Hb-A2 levels in patients with thalassemia trait [2].

The capacity of oxygen carrying of red blood cells decreased to fulfil the body requirements in anaemia which is a pathological condition. The haematological disease in developing countries is Iron Deficiency Anaemia (IDA) affecting 30% of world population [3]. People living in Indian subcontinent, middle east and central Asia are more affected by iron deficiency anemia. 3.5%-10% quoted prevalence of Beta Thalassemia Trait (BTT) in India [4].

Adult male or female carriers of either a- or, B-thalassaemia trait are not usually seriously anaemic, although there is some variation between ethnic groups, sexes, and individual patients [5]. The anaemia usually worsens at times of clinical stress, notably infection and commonly (or always) pregnancy. Indeed, female patients may present or be diagnosed for the first time in pregnancy as cases of refractory hypochromic anaemia. Iron deficiency anaemia commonly present in Pakistan especially in female and young children, 70-80% and 6% of pregnant population having iron deficiency anaemia and β-Thalassemia Trait (BTT) respectively [6].

Objectives of the study

The main objective of the study is to find the frequency of beta thalassemia trait in anaemic pregnant women of a tertiary care setup of Pakistan.

MATERIAL AND METHODS:

This cross-sectional study was conducted in Pakistan Institute of Medical Sciences, Islamabad (PIMS) during January 2019 to December 2019. All pregnant women age 18-45 years and at any gestational age having Hb% level 7-10gm/dl were included in the study. Non pregnant patient and pregnant women with hemoglobin 10-11gm/dl were excluded. 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.

Data was analysed through the software SPSS-V.20. The entire continuous variable was presented as Mean+SD. The entire categorical variables were shown in frequency and percentages.

RESULTS:

The data was collected from 120 patients. The age of the women included ranges from 18-45 years Table 1 show the presence of moderate anemia in relation with the age of pregnant female in which most of the patients 75 (53%) belongs to 18-30 years of age group.

Table 01: Number of patients having iron deficiency anaemia only, β-Thalassemia trait and combined iron deficiency anaemia & β-Thalassemia.

Type of anemia	Number of Patients
Iron Deficiency Anemia (IDA)	108 (91.4%)
B-Thalassemia Trait only (BTT)	4 (2.8%)
Combined IDA + BTT	8 (5.7%)

DISCUSSION:

β-Thalassemia is extremely heterogeneous in terms both of genotype and phenotype, depending on the nature of β-gene mutation and the extent of impairment in β-globin chain production. As a rule, heterozygous carriers of β-thalassemia (one affected allele), are asymptomatic, and only altered laboratory values (low, normal, or slightly subnormal hemoglobin levels, slightly low mean cellular hemoglobin, low mean cell volume, low β:α-globin chain ratio on biosynthesis, HbA_2 ≥3.5%) are observed [7].

Identifying high-risk populations for thalassemia is the main step for reducing incidence. Screening programs may differ throughout the world, depending on population needs, culture, and/or ethics, and although antenatal diagnosis remains a personal choice, policies are focused on education and counselling [8]. In Greece, where carriers

account for 7.5% of the general population, such a program has been in place since the 1970s, raising awareness and drawing attention to this inherited disease. Traditionally, thalassemia was known of higher prevalence in populations of the Middle East, Eastern Mediterranean, India, and Africa, but freedom of movement and subsequent immigration of populations, as well as interethnic mixing, has altered trends [9]. However, according to Hussein et al, there is a lack of randomized trials of preconception genetic risk assessment, and evidence for current policy recommendations is limited to nonrandomized studies [10-11].

CONCLUSION:

It is concluded that Iron deficiency is the leading factor for anemia found in this study, but thalassemia trait also a significant contributing factor. The recommendation is to give proper treatment for only IDA, only β-thalassemia and co-existent cases.

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