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ORIGINAL ARTICLE

Pre-Marital Screening: Thalassemia Trait and Iron Deficiency Anemia in Rahim Yar Khan, Pakistan

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ABSTRACT

Objective: β- thalassemia Minor (BTM) and Iron Deficiency Anemia (IDA) are the main causes of microcytic hypochromic anemias currently in Asian subcontinent. Healthcare facilities in Pakistan have to encounter massive number of patients with anemias every day, where we need to distinguish between (IDA) and (BTM). A number of discrimination indices have been suggested for rapid differentiation of these entities over parameters achieved from automated blood-count analyzers.
Objective. To determine the prevalence of β-thalassemia and IDA amongst the unmarried youngsters of Rahim Yar Khan.
Methods: A cross-sectional study performed in Sheikh Zayed Medical College/Hospital Rahim Yar Khan, Pakistan in 04 months from October 2021 to February 2022, 506 unmarried youngsters were included in the study. CBC, Serum Ferritin and Hb Electrophoresis were performed in Hematology Section, Pathology Department of SZMC Rahim Yar Khan.
Results: Out of 506 subjects, the prevalence of beta thalassemia trait was found 3.3% in males followed by 2.2% in females whereas the prevalence of IDA was 1.7% in males followed by 1.3% in females.
Conclusion: These results indicates that intensive education and awareness strategies are necessary to increase consumption of iron-rich dietary sources. This screening program is beneficial in prevention of high-risk marriages. Screening of spouses by pre-marital screening program is an actual way to control β-thalassemia major.
Keywords: βeta-thalassemia trait (BTT), iron deficiency anemia (IDA), premarital screening.

INTRODUCTION

Thalassemia is a recessive hereditary haemoglobinopathy that is the significant cause of morbidity and mortality[1]. Thalassemia genes are highly common and it is believed that these abnormalities are the most prevalent of all the genetic diseases in humans. Hemoglobin comprises two different alpha and beta chains. Beta chains are synthesized in balanced amount, separately from alpha chains under a specific genetic control. In the case of thalassemia, due to hemolysis Red Cells are incapable to carry sufficient oxygen. If the body is unable to produce the protein, then the blood cells formed are immature and their ability to transfer sufficient oxygen is decreased, resulting in a condition called anemia that manifests in childhood and continues till the life ends[2]. Thalassemia can be classified into two types. In α-thalassemia, α-chains are affected and vise-versa for β-thalassemia. A genetically determined reduction in the synthesis of β-globin chains of hemoglobin molecule is categorized under beta thalassemia[3,4]. According to severity beta thalassemia is classified as mild to severe. In thalassemia minor, protein deficiency is unable to disrupt the normal functions of hemoglobin and patient present with slight anemia. Symptoms of intermediate thalassemia and major thalassemia may vary. Patients diagnosed with intermediate thalassemia may need a blood transfusion. Major thalassemia (Cooley's anemia), a life threatening form with a severe deficiency of β-protein in hemoglobin leading to consistent medical care and blood transfusions. Repeated blood transfusions for life time lead to excessive iron accumulation that should be treated with chelating agents to prevent death and organ failure[5]. Patients having β-thalassemia major with HbA_2 (2%), HbF (98%) and HbA (Nil) need periodic transfusions while patients with β-thalassemia minor having HbA_2 (>3.5%), HbF (Nil) are clinically fine and detection done by routine hematological testing. Though, patients could inherit disorder to the kids if the patient's spouse also has beta-thalassemia minor[6].

In Pakistan, with 5–13% carrier rate, annually 5000–9000 children are born with β thalassaemia major, Screening of couples is mandatory by receiving appropriate advice to reduce the chances of producing affected child. Premarital screening programs have been widely accepted before 1970,s and many countries made it mandatory. High frequency of thalassemia may be detected in wide-belt ranging from the Mediterranean basin over Middle-East, Indian subcontinent and South East Asia[6,7]. About 15 million people present with thalassemia disorder and 100,000 babies born every year suffer from severe forms of thalassemia. World Health Organization (WHO) predicted that as a minimum (6.5%) of the world populations are carries of several inherited hemoglobinopathies. BTM is inherited disease with carrier-rate of about 08-10% in Pakistan[8]. Beta thalassemia trait (BTT) and (IDA) are the frequent cause of hypochromic microcytic anemia[9]. The differential diagnosis is the main concern to help physician to elude excessive iron therapy and false diagnosis of β-thalassemia minor, exclusively before marriage to counsel couples for the prevention of β-thalassemia major[10].

IDA is the most frequent cause of anemia due to inadequate dietary intake, decreased iron absorption, iron loss in blood due to uterine, intestinal and urinary sources[11]. Females are commonly affected because of blood loss during menstruation and poor iron containing diet. People suffering from IDA exceeds one billion[12]. Iron deficiency anemia in pregnancy occurs due to utilization of iron stores to increase blood volume and hemoglobin for the developing fetus. Presentation of iron deficiency anemia contributes pallor of skin, nail beds and conjunctiva, fatigue, vertigo, exertion, dyspnea progressing to breathlessness, headache, tachycardia, cardiac systolic flow murmur and hemodynamic instability[13]. In the diagnosis of IDA, reduction in mean corpuscular volume (MCV), mean cell hemoglobin (MCH), hemoglobin (Hb) and RBC count can be seen. Consequently, on blood film MCV is low with normal or increased RBC count will be appreciated in Beta Thalassemia Trait. HbA2 on Hb electrophoresis and serum-ferritin levels are confirmatory test to differentiate between these two entities[14]. Therefore current study has been carried out to determine the prevalence of beta thalassemia trait and Iron Deficiency Anemia amongst the unmarried youngsters of Rahim Yar Khan, Pakistan.

MATERIALS AND METHODS

This was a descriptive cross-sectional study including 506 young males and females aged 16 to 30 years. Duration of study was from October 2021 to February 2022 and the study was conducted in Hematology Section, Pathology Department of Sheikh Zayed

Medical College/Hospital Rahim Yar Khan, Pakistan. Blood samples from subjects were processed for CBC, Ferritin and Hb-Electrophoresis. On the basis of these tests Thalassemia trait and iron deficiency anemia was differentiated.

Methodology: 5 ml of blood was withdrawn under aseptic technique. Venous blood (3ml) was collected in (EDTA) coated tubes for CBC and Hb Electrophoresis while 2 ml was collected in a Gel tube for Serum Ferritin. The complete-blood count (CBC) was measured by an automated hematology analyzer (Sysmex KX 21; Sysmex, Tokyo, Japan). Hb Electrophoresis was performed on Mini Cap Fully Automatic Electrophoresis Machine. Serum ferritin level were measured by using "Cobas e 411 Rochi HITACHI". The Analysis of collected data was done on SPSS 21.

RESULTS

Subjects with Microcytic Hypochromic Anemia having (MCV) <80 fl with a ferritin levels <12 µg/dl were diagnosed as iron deficient, while Students with microcytosis, hemoglobin A2 ≥ 3.5% were reported to have beta-thalassemia trait with MCV less than 80 fL or MCH of less than 27 pg and a hemoglobin (A2) level of more than 3.5%.

Table I: Gender wise Distribution of Thalassemia Trait.

Gender		Frequency	Percentage
Male	Beta thalassemia trait	8	3.3
	Normal	234	96.7
	Total	242	100
Female	Beta thalassemia trait	6	2.3
	Normal	258	97.7
	Total	264	100

Out of 442 males, 8 were suffering from thalassemia trait (3.3% of total male population). Amongst 264 females 06 were suffering from thalassemia trait (2.3% of total female population).

Table II: Gender wise Distribution of Iron Deficiency Anemia.

Gender		Frequency	Percentage
Male	Iron deficiency anemia	4	1.7
	Normal	438	98.3
	Total	242	100
Female	Iron deficiency anemia	35	13.3
	Normal	229	86.7
	Total	264	100

Amongst 442 males, 4 were suffering from iron deficiency anemia which became 1.7%.Out of (264) females, (35) were diagnosed with IDA which became (13.3%) of total female.

DISCUSSION

Hemoglobinopathies are vital health problems in Pakistan. Thalassemia is a group of genetic disorder that occur primarily due to substandard formation of globin-chains of the hemoglobin moiety of the RBCs. These disorders lead to excessive RBCs destruction resulting anemia. The discrimination between IDA and beta-thalassemia has a significant clinical implication. Therefore, a reliable diagnosis would reduce excessive laboratory testing and elude inappropriate treatment. A variety of parameters can facilitate the differentiation between IDA and beta-thalassemia[15]. The prevalence of beta-thalassemia trait with increased hemoglobin A_2 and microcytic-hypochromic anemia were 3.3% (8/242) males and 2.3% (6/264) female's .The prevalence of iron deficiency anemia in 506 screened persons were 1.7% (4/242) males and 13% (35/264) females. Among the carriers of the β-thalassemia trait, 1.5% (4/264) female had high hemoglobin A2 and iron deficiency anemia. The overall prevalence of beta thalassemia in 506 were 2.7 % (14/ 506). In comparison Saleh et al, 2016 reported the prevalence of IDA in young girls in Egypt was 30.2% [16]. Aziz et al, 2015 reported prevalence of Beta thalassemia in general population of Quetta city was 6.5 % [17]. Abdul Rahman et al, 2007 screened 488,315 individuals in Saudi Arabia were for beta thalassemia and 3.22 had beta thalassemia trait. In Iran premarital beta thalassemia trait prevelance was 3.5% [18]. It is not uncommon for Egyptians to be carriers of the beta-thalassemia gene, which is found at a frequency of 0.03 percent. As many as 1,000/1.5 million Egyptian babies will be born with beta-thalassemia each year, according to an estimate[19]. It is not necessary that all screening programs share success. But many strong social factors such as religious beliefs, traditions, cultural norms, education level, literacy, governmental policies and the individual's attitudes may impact the acceptability of successful preventive program In Mediterranean countries, the number of patients with thalassemia has decreased significantly.

CONCLUSION

Pre-marital screening program is useful for the prevention of high-risk marriages. Pre-marital carrier couples screening is an actual way of monitoring beta-thalassemia due to strong tradition of consanguineous marriages in Pakistan. As majority of people do not accept termination of pregnancy. Therefore screening for thalassemia prior to marriage is hypothetically an easier and more cost-effective practice.

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