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Hematological Characterization of Beta Thalassemia in Sudanese Patients

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Abstract

Thalassemia is common inherited disorder among humans, and they represent a major public health problem in many areas of the world. The study aimed to measurement of hematological characterization of beta thalassemia in Sudanese patients. Blood samples from 61 beta thalassemic patients were collected after written consent form obtained from all participants. The frequency of Adults (>18 years) were 45 (73.8%), and Children's (<18 years), were 16 (26.2%) the frequency of male was 27 (44.3%) and 34 were females (55.7%). Hemoglobin estimation and red cell indices were carried out using the automatic blood cell counter Sysmex KX21N. The results showed Hb and RBCs indices were vared between mild to moderate and severe decreasing, Hemoglobin concentration (Hb) with the mean value of 9.6 g/dL, with minimum value of 6.1g/dL and maximum of 11.9g/dL, while RBCs was increased in all patients, mean value 5.2c/L, Mean corpuscular volume (MCV) mean was 58.9 fL, hematocrit was 30.4, mean corpuscular hemoglobin (MCH) 18.8 pg, mean corpuscular hemoglobin concentration (MCHC) was 31.7pg and RDW was 18.8%. The method used for hemoglobin electrophoresis was cappilary electrophoresis, Hb pattern shows increased HbA2 and HbF, the mean of HbA is 78.3%, HbF is 2.3%, and HbA2 is 6.5% with the min value of 3.6% and max of 12.2%. While the mean of serum iron was 82.75ug/dL, 7 patients showed low level, 19 high level and 35 was normal level. Comparison of hematological analysis (HbA2) in thalassemic patients coexisted with Iron deficiency and without result was insignificant difference (p = 0.645) this result disagree with references that say iron deficiency masking HbA2. Nevertheless the association between HbA2 and HbF revealed a statistically significant difference (p<.013) and HbA2 with Hb was insignificant (p =.260).

Keywords: Thalassemia; RBCs indices; Hb electrophoresis; CBC; Iron; Sudan

Abbreviations: CBC: Complete Blood Count; Hb: Hemoglobin; MCHC: Mean Corpuscular Hemoglobin Concentration; MCV: Mean Corpuscular Volume; HbA2: Hematological A2; MCH: Mean Corpuscular Hemoglobin;

RDW: Red Cell Distribution Width; TIF: Thalassemia International Federation.

Introduction

Thalassemia is a Mendelian autosomal recessive heritable blood disorder it's a group of genetically determined microcytic, hypochromic anemia's resulting from a decrease in synthesis of one or more globin chains in the hemoglobin molecule [1]. The most common types are alpha and beta thalassemia according to which globin chain is reduced [2]. Beta thalassemia is classified into three types depending on the severity of symptoms: thalassemia major also known as Cooley's anemia [3,4]. Thalassemia intermediate and thalassemia minor, thalassemia major is more severe. The signs and symptoms of thalassemia major appear within the first 2 years of life, Children develop life-threatening anemia, and they do not gain weight and grow at the expected rate (failure to thrive) and may develop yellowing of the skin and whites of the eyes (jaundice) [5]. Affected individuals may have an enlarged spleen, liver, heart, and their bones may be misshapen. Some adolescents with thalassemia major experience delayed puberty. Many people with thalassemia major have such severe symptoms that they need frequent blood transfusions to replenish their red blood cell supply over time, an influx of iron-containing hemoglobin from chronic blood transfusions can lead to a buildup of iron in the body, resulting in liver, heart, and hormone problems. Thalassemia intermedia are milder than thalassemia major [6]. The signs and symptoms of thalassemia intermedia appear in early childhood or later in life. Affected individuals have mild to moderate anemia and may also have slow growth and bone abnormalities [7]. The disorder may occur in the homozygous or heterozygous state. Heterozygotes may be asymptomatic but Homozygotes typically have a severe, often fatal, disease. It involves increased (HbA2) and decreased production of normal adult hemoglobin (Hb A), the predominant type of hemoglobin from soon after birth until death [8]. Mostly the patients are diagnosed on routine blood examination. Beta Thalassemia carrier it is commonly not diagnosed until adolescence or adult life, and may be detected in a routine hematological screening examination. The red Cell indices, Hb Electrophoresis and molecular studies give more reliable diagnosis. In thalassemia trait MCV and MCH are low while MCHC is marginally reduced or normal. Hemoglobin electrophoresis and molecular study is essential for definite diagnosis of β-thalassemia cases. Normally Hb A2 is less than 3.2% but in Beta- thalassemia trait it is more than 3.5% [9].

Materials and Methods

Across-sectional descriptive study was carried out to detect hematological characterization of Beta-Thalassemia Sudanese patient in Khartoum State Sudan, during the period of July 2017 to July 2019. From each patient, 2.5 ml of venous blood sample was collected in sterile EDTA container. The blood samples were analyzed for Complete blood count (CBC) using the automated hematology analyzer Sysmex KX21N, (manufactured by Sysmex corporation Kobe, Japan) within 24 hours of blood collection. On the same day itself the blood samples were screened for Haemoglobinopathies by Hb electrophoresis method (Sebia, France) [10]. The inclusion criteria Patients were diagnosed as Beta – thalassemia, availability of patient demographic data and laboratory reports, (CBC, Hemoglobin electrophoresis, and peripheral blood picture, Iron studies), Patients not diagnosed as Beta-thalassemia or coexisted with other hemoglobin variants or with other hematological malignancy excluded from the study. Permission of this study was obtained from the local authorities in the area of the study. The objective of the study explained to all individuals participating in this study. An informed written consent obtained from all participants.

Results

Out of 61 beta-thalassemic patients the frequency of Adults (>18 years) was 45(73.8%), and 16 (26.2%) was Childrens (18 years) and the frequency of male to female was 27 of the patients were males (44.3%) and 34 were females (55.7%) as shown in (Table 1) Hemoglobin estimation and red cell indices were carried out using the automatic blood cell counter Sysmex KX21N. The results obtained were as follow: Hemoglobin concentration (Hb) with the mean value of 9.6 g/dL, with minimum value of 6.1g/dL and maximum of 11.9g/dL, while RBCs was increased in all patients with mean value of 5.2c/L, Mean corpuscular volume (MCV) mean was 58.9 fL, hematocrit was 30.4, mean corpuscular hemoglobin (MCH) 18.8 pg, mean corpuscular hemoglobin concentration (MCHC) was 31.7pg and RDW was 18.8%, as shown in (Table 2). The method used for hemoglobin electrophoresis was cappilary electrophoresis the Hb pattern shows increased HbA2 and HbF, the mean of Hb A is 78.3%, HbF is 2.3%, and HbA2 is 6.5% with the min value of 3.6% and max of 12.2% as shown in (Table 3). The mean of serum iron was 82.75ug/dL, 7 patients showed low level, 19 high level and 35 was normal level, (Table 4) and (Figures 1-3).

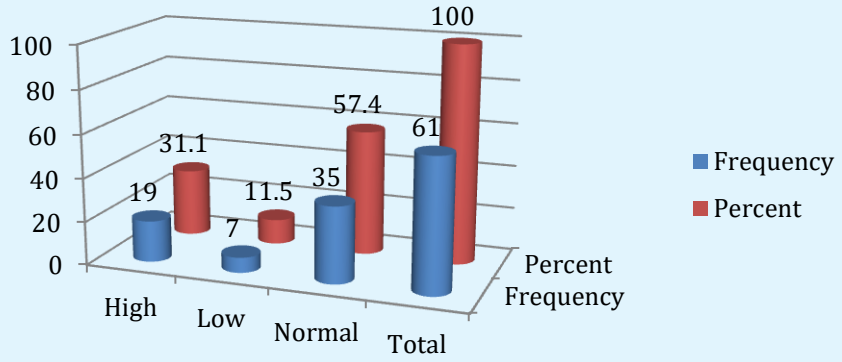

Figure 1: Frequency of Iron level in study group.

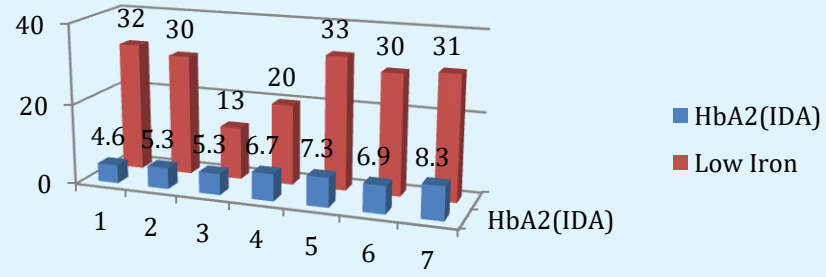

Figure 2: HbA2 in thalassemic patients co-existed with iron deficiency.

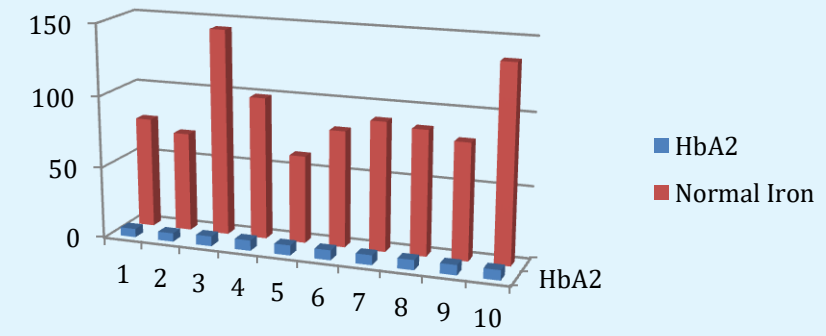

Figure 3: HbA2 in normal iron group.

	Frequency	Percent
Gender		
Male	27	44.3
Female	34	55.7
Age group(y)		
<18	16	26.2
>18	45	73.8

Table 1: Gender and Age distribution in the study group.

Parameters	Hb/g/d	HCT	RBCs	MCV	MCH/pg	MCHC/pg	RDW%
Mean	9.618	30.364	5.197	58.882	18.761	31.657	18.825
Std. Deviation	1.3214	3.6247	0.7394	6.5545	2.9306	1.7615	2.9644
Minimum	6.1	18.8	3.2	44.2	13.3	27.2	12.5
Maximum	11.9	36.2	6.7	76.4	27.2	35.6	23.3

Table 2: Mean and standard deviation of Hb and RBCs indices in the study group.

Hb electrophoresis parameters	HbA2/%	HbF/%	HbA/%
Minimum	3.6	0.4	70.1
Maximum	12.2	10.4	92.7
Mean	6.498	2.277	78.289
Std. Deviation	1.3895	1.9511	3.2221

Table 3: Hb electrophoresis pattern in thalassemic patients.

	Iron result
Mean	82.75
Std. Deviation	29.112

Table 4: Mean and standard deviation of serum iron in study population.

Discussion

For 61 beta-thalassemic patients, Hemoglobin estimation and red cell indices were carried out using the automatic blood cell counter Sysmex KX21N. The results obtained were; overall mean haemoglobin concentration (Hb) was decreased 9.6 g/dL. Red blood cell count (RBCs) 5.19×1012 cell/L was found to be raised. Red blood cell indices were found to be low (packed cell volume mean cell volume (MCV) 58.9 fL, mean cell haemoglobin (MCH) 18.8 pg, mean cell haemoglobin concentration (MCHC) 31.7 g/dL, red cell distribution width (RDW) 18.8)this results agreed with several studies in literature Eg. Galanello, et al. Tahir, et al. Idit, et al. [11-13] specially with Dr. Sana [14] literature. Also this study agreed with the data mentioned in thalassemia international federation (TIF). Hb electrophoresis was measured by capillary electrophoresis results shows mean of HbF is 2.3% which was high, and HbA2 is 6.5% with the min value of 3.6% and max of 12.2% also shows significant increase which agreed with several studies eg. Sana, [14,15]. The mean of serum iron was 82.75ug/dL, 7 patients showed low level, 35 was normal level and 19 high levels those are mainly due to blood transfusion, unfortunately data of blood transfusion was missing in this study, Many studies shows similar result unless in recurrent transfused thalassemic patients levels are much higher. Comparison of hematological analysis (HbA2) in thalassemic patients coexisted with Iron deficiency and without result was insignificant difference (p = 0.645) this result disagree with many references that say Iron deficiency masking HbA2. Nevertheless the association between HbA2 and HbF revealed a statistically significant difference (p<.013), that means there is positive correlation between HbA2 and Hb F. and the correlation between HbA2 with Hb was insignificant (p =.260) [16-18].

Conclusion

This study was detected the hematological characterization of beta thalassemia the results obtained for Hb, RBCs indices, and Hb electrophoresis were agreed with several studies, In areas where modern equipment's for diagnosis are not available, the red cell indices and Hemoglobin electrophoresis gives more reliable diagnosis

for beta thalassemia and molecular study provide definitive diagnosis. Since there are no symptoms for beta thalassemia trait it's very important to discover at early as possible from routine hematological test to prevent beta thalassemia major offspring's.

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