Assessment of Some Health and Social Parameters in Thalassemia Major Patients Registered at the Inherited Blood Disorders Center in Al-Karamah Hospital in Baghdad

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ABSTRACT

Background: Beta thalassemia major is a common disease in the Mediterranean basin and it led to early death of the patient if not treated and it is a public health problem in many countries.

Objectives: To evaluate some health and social parameters in beta-thalassemia major patients in Baghdad.

Methods: This is a cross sectional study conducted in the period between April and September 2022 in the Inherited Blood Disorders Center (Thalassemia center) in Al-Karamah hospital in Baghdad. The patients included in the study were diagnosed and registered in the thalassemia center as beta-thalassemia major. The information were taken from the files of the patients.

Results: The study included 82 patients; their mean of age was 17.2 years and the range was 1-39 years age. Forty-seven (57.4%) patients were males and 35 (42.6%) patients were females. Most of the patients (74.4%) presented and diagnosed as thalassemia major in the first year of life. The results of screening for viral hepatitis showed that one (1.2%) patient was infected with hepatitis B virus (HBV) and 22 (26.8%) patients were infected with hepatitis C virus (HCV). All the studied patients screened for human-immunodeficiency virus (HIV) antibody and the results were negative. Fifteen (18.3%) patients underwent splenectomy and four (4.9%) patients underwent cholecystectomy. The study showed that 47 (57.3%) families had one affected sibling, 23 (28 %) families had two affected siblings and 12 (14.6%) families had three and more affected siblings. Fifty-nine (72%) families showed a degree of consanguinity between the parents while 23 (28%) families had no consanguinity.

Conclusions: Most of the patients had severe type of thalassemia as they presented in the first year of life. Infection with HCV was much more than the infection with HBV and HIV. There was weakness in the preventive measures as many families had more than one affected sibling. In most of the families, there was consanguineous relation between the parents, which increased the incidence of beta thalassemia major.

Keywords: Thalassemia major, Al-Karamah hospital, HCV, Consanguinity.

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Thalassemias are a heterogeneous group of genetic disorders that result from a reduced rate of synthesis of α- or β-globin chains. Beta-Thalassemia is more common in the Mediterranean region, while α-thalassemia is more common in South-East Asia[(1)].

The clinical spectrum of β-thalassemia reflects the heterogeneity of the genetic molecular lesions causing the disease. β-Thalassemia minor is usually caused by heterozygous β-thalassemia. These patients may have mild anemia and misdiagnosed as having iron deficiency anemia. Iron studies show normal to increased iron with normal iron saturation. Documentation of a compensatory increase in HbA2 and HbF on hemoglobin electrophoresis confirms the diagnosis.

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Patients with β-thalassemia intermedia have two β-thalassemia alleles, but at least one of them is a mild β^+ mutation. These patients have severe chronic hemolytic anemia but do not require transfusions. Because of ineffective erythropoiesis, the patients chronically hyperabsorb iron and may develop iron overload in the absence of transfusions[2,3].

β-Thalassemia major results from homozygous (or compound heterozygous) β^0-thalassemia, leading to severe hemolytic anemia; such patients are diagnosed in infancy and become transfusion dependent after the first 4-6 months of life. This condition characterized by ineffective erythropoiesis, shortened red cell life, expanded intramedullary hemopoiesis and hemopoiesis at extra medullary sites such as liver and spleen. Sufferers are dependent on blood transfusion for survival. Diagnosis of beta thalassemia homozygosity and compound heterozygosity is by demonstration that HbA is absent or greatly reduced with HbF being the dominant hemoglobin[4].

Clinical management of thalassemia major consists in regular long life red blood cell transfusion and iron chelation therapy to remove iron introduced in excess with transfusion. At present, the only definitive cure is bone marrow transplantation. Therapies under investigation are the induction of fetal hemoglobin with pharmacological compound and stem cell gene therapy[5].

The aim of this study is to evaluate some social parameters and health complications in beta-thalassemia major patients in Baghdad.

Methods

This is a cross-sectional study conducted in the period between April and September 2022 in the Inherited Blood Disorders Center (Thalassemia center) in Al-Karamah hospital-Baghdad. The study included 82 patients diagnosed and registered in the thalassemia center as beta-thalassemia major. The information were taken from the files of the patients. They included file number, date of registration in the center, age of the patient, gender, age at diagnosis, the results of the viral screen for HBV, HCV and HIV and some complications, consanguinity between the parents, the number of the affected siblings in the family.

Data analysis made by the use of the statistical package (Epi-info version 6).

The data presented by frequency distribution and means and standard deviation (SD) and median made for selected variables.

Results

The mean of age for the patients included in the study was 17.2 ±11 years with male to female ratio of 1.3:1.

The distribution of these patients according to the decades of age is shown in table 1; the peak was in the second decade of life.

Regarding the age at diagnosis, most of the patients presented and diagnosed as thalassemia major in the first year of life; 61(74.4%) patients diagnosed during the first year of life, 9 (11%) patients in the second year of life, 12 (14.6%) patients after the 2nd years of life, (Table 2).

The results of screening for viral hepatitis in the studied patients showed that one (1.2%) patient were infected with hepatitis B virus (HBV) and 22 (26.8%) patients infected with hepatitis C virus (HCV) and no HIV positive, (Table 3).

Fifteen (18.3%) patients underwent splenectomy and four (4.9%) patients underwent cholecystectomy.

Thirteen (15.8%) patients suffered from osteoporosis and four (4.9%) patients suffered from cardiomyopathy, (Table 4).

The study showed that 47 (57.3%) families had one affected sibling, 23 (28 %) families had two affected siblings and 12(14.6%) family had three and more affected siblings, (Table 5).

Regarding consanguinity between the parents, 59 (72%) families showed some

degree of consanguinity between the parents as following: 24 (29.3%) families showed 1st degree of consanguinity, 30 (36.6%) families showed 2nd degree, 5(6.1%) families showed 3rd degree while 23(28%) families had no consanguinity, (Table 6).

Table 1: Age distribution of thalassemia.

Decade	No. of patients	Percentage
First decade	21	25.6
Second decade	31	37.8
Third decade	21	25.6
Fourth decade	9	11.0
Total	82	100

Table 2: The distribution of patients according to the age at diagnosis.

Age at diagnosis	No. of patients	Percentage
1st year of life	61	74.4
2nd year of life	9	11
After 2nd year	12	14.6
Total	82	100

Table 3: The distribution of viral screen.

Viral screen	No. of patients with +ve results	Percentage
Hepatitis B virus	1	1.2
Hepatitis C virus	22	26.8
Human immunodeficiency virus	Zero	Zero

Table 4: The complications of thalassemia.

Complications	No. of patients	Percentage
Splenectomy	15	18.3
Osteoporosis	13	15.8
Cholecystectomy	4	4.9
Cardiomyopathy	4	4.9

Table 5: The distribution of number of affected siblings in the families.

No. of affected siblings in the family	No. of families	Percentage
one	47	57.4
two	23	28
Three and more	12	14.6
total	82	100

Table 6: The distribution of consanguinity in the families.

The consanguinity between the parents	No. of families	Percentage
1st degree	24	29.3
2nd degree	30	36.6
3rd degree	5	6.1
No consanguinity	23	28
Total	82	100

Discussion

Thalassemia major is a medical, social and economic problem; medically the thalassemic patients suffer chronic anemia and blood transfusion complications especially iron overload[6]. Socially, they suffer physical weakness that prevent the patient from full social engagement and economically the treatment is costly[7].

The present study showed that 25 % and 11% of patients were in the 3rd and 4th decade of life and this reflect the efficacy of treatment because when treatment is not sufficient, the patients die in the first decade of life[6]. In the seventies of the previous century, the mean of age for these patients in the United States was 11 years, but it reached 25 years due to treatment evolution[7].

The present study showed that most of the patients presented and diagnosed as thalassemia major in the first year of life, this situation might indicate a severe type of beta thalassemia mutations, because the less severe types presented in the second year of life or later[8].

This study showed that the results of viral screen was zero for the HIV and only 1% for HBV and such results point to the efficacy of the screening of the transfused blood for these viruses.

The finding of HCV was high figure (26%). It is higher than that in Ninawa, Iraq (7%)[9], and the study of Al-Sweedan and Jaradat et al 2011 in Jordan, which showed 16%[10]. This difference may be due to the source of blood for transfusion and the administration of intra muscular and intravenous drugs which are under control in Ninawa more than in Baghdad.

The finding of HCV was similar to study of Esfandiary and Sadigh et al at 2019 which shows prevalence of 29% in Iranian thalassemia major patients[11] and also similar to the study of Waheed and Saba et al 2021 which showed prevalence of 29% among beta thalassemia major patients in Pakistan[12].

The present study shows that 18% of the studied patients underwent splenectomy and this figure is lower than that reported in the study of Al-Hawsawi and Hummaida et al 2001, which was 32%[13].

The present study showed that 28% of families have two affected siblings and 14% have three and more affected siblings and this high figures may be due to the weakness of the preventive measures, although there is pre-marital testing, but this is not enough. Initiation of active preventive program play important role in decreasing the incidence of thalassemia major. Pakistan was one of the Islamic countries that initiate a preventive program for thalassemia and succeeded in the management of the disease[14-16].

The present study showed that in most of the families (72%) there was a degree of consanguinity between the parents and such state increase the probability of siblings with thalassemia major. This result was similar to the study of Omer and Jamaleldin 2011 in Ninawa governorate[9] and similar to studies carried out in Turkey[17,18] and in Syria[19].

In conclusions; most of the patients had severe type of thalassemia as they presented in the first year of life. Infection with HCV was much more than the infection with HBV and HIV. There was weakness in the preventive measures as many families had more than one affected sibling. In most of the families, there was consanguineous relation between the parents, which increased the incidence of beta thalassemia major.

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