JOURNAL OF TECHNIQUES

Journal homepage: http://journal.mtu.edu.iq

RESEARCH ARTICLE - MEDICAL TECHNIQUES

Studying Some Factors That Increase the Risk of Thalassemia in The City of Baquba

Sara Fawzi Sahm[1*]

[1] Continuous education center, Middle Technical University, Baghdad, Iraq

[*] Corresponding author E-mail: sara.fawzi.s@mtu.edu.iq

Article Info.

Article history:

Received
14 September 2022

Accepted
07 October 2022

Publishing
15 November 2022

Abstract

In the current study, 200 cases with thalassemia (111 males and 89 females) were used. Case samples were obtained from persons listed for Baquba Teaching Hospital / Hematology Center from November 1st, 2019 to January 31, 2020. Data were taken from patients' get-in hospital clinical reports through a form prepared for collecting data. The study aims to measure the impact of some factors on Thalassemia. The results showed that there is an effect of the relationship between chronological age and getting the disease, as it was found that the highest percentage was within the age group (1-10) years. It was also found that the number of males carry the disorder is higher than the number of infected females. Also, the effect of the type of Thalassemia factor on the infection of the disease shows that those who carry thalassemia major are more affected than those who carry thalassemia intermedia. The effect of the blood type factor on the infection rate shows that the highest number of infected people was those with blood type O. The effect of the disease history factor on the infection of the disease shows that the category (less than one year old) is more likely to get the disease. The effect of the factor of the number of infections in the family shows that the category (there are no previous infections in the family) are more numerous.

Publisher : Middle Technical University

Keywords: Thalassemia; Hemoglobin; Hemolytic anemia; Genes.

1. Introduction

Thalassemia is one of the diseases known since a long time ago. This disease was discovered by Dr. Cooley in 1925. It is a genetic disease that affects red blood cells, making them unable to mature and grow properly. This disease is caused by a defect in the genes. This defect occurs in the manufacture of hemoglobin inside a red blood cell, so the hemoglobin molecule becomes unable to perform its function properly. The spleen breaks it and exits the blood circulation, which results in hereditary and chronic anemias affecting children in the early stage, since they receive two defective genes, one gene from each parent [1]. There is also an effect upon the red blood cells in terms of number, morphological and volumetric characteristics during the stages of their formation within the bone marrow, as they tend as a result to be small in size. This could lead to making it vulnerable to destruction by macrophages, which are abundant in bone marrow tissue, where they can identify and distinguish abnormal cells and eat them. This leads to destructing a large group of red blood cells during the stages of development [1].

Thalassemia is an inherited disease with autosomal recessive gene disorders due to impaired synthesis of one or more globin chains. Such impairment changes hemoglobin production [2]. Thalassemia can be defined as a complex of different hereditary hemoglobin disorder combination showing inadequate production of at least a single globin chain causing imbalanced globin chain productions and damaged hemoglobin molecule and finally leading to anemia [3]. The metalloprotein, hemoglobin (Hb), is present inside red blood cells (RBCs) of all vertebrates (except the fish family Channichthyid) and certain invertebrates. Hemoglobin acts as a vehicle for oxygen transportation [4].

Thalassemia's are a group of disorders where there is an impairment in the balance between alpha and beta globins, and such imbalance results from a reduction in one or more globin gene production. The decreased globin chain results in un-pairing of the remaining globins, that are precipitated in the RBC precursor (ineffective erythropoiesis) or destroyed eventually in blood circulation (haemolysis) [5]. Consequently, patients will develop different anemia degrees with extramedullary haematopoiesis. The beta thalassemias usually occur due to point mutation, while alpha thalassemias often occur due to deletional mutations [6, 7].

The term ''thalassemia'' is from the Greek word "thalass" meaning (sea), and ''emia'' from the Latin meaning ''blood'', and the thalassemia syndrome is named in accordance with the affected globin chain or the abnormal hemoglobin engaged. The defect of β-globin gene gives rise to β-thalassemia, while the α-globin gene mutations cause α-thalassemia.

The thalassemia was first detected among the Mediterranean people and referred to as Mediterranean anemia. The most common hemoglobinopathy is thalassemia. It is estimated that about 5% of the world population carry at least a single variant allele of thalassemia. Some thalassemia types have rare dominant inheritance patterns, although they usually exhibit autosomal recessive inheritance patterns [8].

How is thalassemia diagnosed?

Symptoms of hemolytic anemias such as pallor and hepatosplenomegaly directly from birth in α-thalassemia, or from many months following birth in β-thalassemia, suggest severe form of thalassemia, particularly when there is a microcytic anemia, thus, the second line testing must be done immediately. The major feature of minor, intermediate or major thalassemia is microcytic, hypochromic erythrocytes (MCV= 4%) [9].

Nomenclature			
Hb	Hemoglobin	α	Alpha
RBCs	Red Blood Cells	β	Beta

Thalassemia prevention depends on public awareness of the disease, carrier detection, genetic consultation and prenatal examination [10, 11] In many countries, such as the UK, the neonatal and antenatal haemoglobinopathy screening programmer facilitates early identification of risky couples and easily diagnosis of children with serious forms of major α and β-thalassemia (www.sct.screening.nhs.uk). To find out the hydrops fetalis risk or diagnose children with hemoglobin H disease, the α-thalassemia carriers with deletions on the same chromosome must be offered to genetic counselling [10]. Counselling and testing must also be offered to the first-degree relatives of thalassemia patients.
Evaluation of iron level and functions in the liver and heart is very important. Disparity between heart and liver iron load may be present due to the variation in the kinetics and mechanisms of iron clearance and uptake between the body organs. Although many observational studies revealed a relation between high serum ferritin levels and early deaths, the prediction of iron load (particularly cardiac iron) from serum ferritins may be undependable [12]. In an intervention study, patients who had ferritin levels lower than 2500 μg/L for more than 10 years, showed a significant decreased risk of heart diseases and deaths, in comparison with those who showed ferritin levels grater than 2500 μg/L [13].
According to the defect in genes responsible for producing alpha or beta protein chains, thalassemia is divided into beta thalassemia and alpha thalassemia. The disease may be caused by a number of genetic mutations and the disease may appear in the event of the confluence of two defective beta genes. There are four genes involved in producing the alpha chain. When there is a defect in three genes or a defect in all four genes, all of them could lead to the emergence of symptoms of the disease [14].
Thalassemia is genetically transmitted from parents to children. When one of the parents carries the disease or infected with, it may be transmitted to certain children in the simple form (children become disease carriers). When both parents are carriers or affected by the thalassemia, there will be a 25% chance that a baby will be born with severe disease. As a result, thalassemia is divided into major thalassemia, thalassemia intermedia, and thalassemia minor [15].

1.1. Research objectives

The aim of this research lies in answering the following questions:
1. Does gender affect the increase in thalassemia disease?
2. Does the age of the individual have an effect on thalassemia?
3. Does the blood type factor have an effect on thalassemia?
4. Are the medical history and the number of infections in the family influencing factors in increasing the infection of this disease?

2. Research Materials and Methods

This study was conducted in Baquba district for the period from November 1st, 2019 to January 31, 2020. Case samples were obtained from people who visited Baquba Teaching Hospital / Hematology Center from the files of people infected with thalassemia. The disease is diagnosed by the competent physician through the required information form. The total number of samples was (200) samples (111 males and 89 females). The study of the sample relied on some factors to measure the extent of their impact on thalassemia. A special information form was also prepared to include (patient's age, gender, blood type, type of thalassemia, pathological history, number of infections in the family) according to the form shown in Table 1.

Table 1 shows the information form

No	Age	Gender	Blood type	Type of Thalassemia	Pathological history	No. of family infected cases
1						
2						
3						

3. Results and discussions

3.1. The effect of age factor upon getting the disease

As noticed in Table 2 the effect of the relationship between chronological age and the infection of the disease, as it was found that the highest infection of disease was within the age group (1-10) years. We find that the patient's chronological age has an effect on the infection, so the number of infected people was (111) out of the total number, as the infection rate was 55.5%. In the second level, the age group was (11-20) years, in which the number of infected people was (55) out of the total, and the infection rate was 27.5%. Hence, it is clear that the infection of the disease decreases with age, as we note that the lowest rate of infection was for the age group (41-50) years, which was 2% of infection. This result is in agreement with the study [16].

3.2. The effect of gender type on carrying the disease

It is noticed from Table 2 that the number of males infected with thalassemia is higher than the number of infected females, as the number of males was (111) persons, at a rate of 55.5%. As for females, they were (89), or 44.5%. This result is in agreement with the study by [17].
Increasing the number of infected males over females may not mean that there is a genetic link to gender, but it may reflect families' interest in males more than females, in addition to the families' reluctance to visit hospitals when the disease appears in females, but we cannot neglect the possibility of gender cause [16].

Table 2 shows the effect of age and gender factors on thalassemia

Factor		No	Infection rate
Age	10-1	111	% 55.5
	20-11	55	% 27.5
	30-21	26	% 13
	40-31	4	% 2
	50-41	4	% 2
	Total	200	% 100
Gender	Males	111	% 55.5
	Females	89	% 44.5
	Total	200	% 100

3.3. The effect of the type of thalassemia factor on the infection of the disease

Table 3 shows the effect of the type of thalassemia factor on the infection of the disease. It was found that people with thalassemia major are more numerous than people with thalassemia intermedia, as they were 157 and their percentage was 78.5%. The number of people with thalassemia intermedia was (43) infected persons, or 21.5%. This result is in agreement with the study by [16]. The type of thalassemia major needs to give the patient medication for the disease, as well as giving him blood periodically. This costs the family a large financial burden, in addition to the psychological problems that the patient suffers from.

3.4. The effect of the blood type factor on the infection of the disease

Table 3 shows the effect of the blood type factor on the infection rate, as the table shows that the highest number of infected people was of type (O). Their number was (76) infected with a percentage of 38%, and the number of those infected with type (A) was (64) with a percentage of 32%. The number of infected (B) group was (45), with a rate of 22.5%. The others are from the blood type (AB), their number was (15) and their percentage was 7.5%. This result agrees with the study of [17] and disagree with the study of [18]. The reason may be due to the fact that there is a relationship between the transmission of the gene responsible for producing globulin on chromosome 16 and the other gene responsible for the type of antigen on the cell membrane of erythrocytes and responsible for the type of blood [7].

Table 3 shows the effect of thalassemia type and blood type factors on thalassemia disease

Factor		Number	Infection rate
Kind of thalassemia	Beta (major)	157	% 78.5
	Alpha (intermedia)	43	% 21.5
	Total	200	% 100
Blood type	O	76	% 38
	A	64	% 32
	B	45	% 22.5
	AB	15	% 7.5
	Total	200	100 %

3.5. The effect of the pathological history factor on getting Thalassemia

Table 4 shows the effect of pathological history on getting the Thalassemia, where the infected persons were divided into three categories (less than a year, 1-3 years, and 4-7 years). It was found that people who have a pathological history (less than a year) are the most common carrier of the disease. They were (91) infected persons, and their percentage was 45.5%. As for the number of people with a pathological history (1-3) years, it was (79) infected, and their percentage was 39.5%. As for people with a history of illness (4-7) years, their number was (30) infected, and their percentage was 15%.

3.6. Effect of the factor of the number of individuals in the family carrying the disease on getting Thalassemia

Table 3 shows the effect of the number of infections in the family on the infection rate, as the infected ones were divided into five categories (no infection, one infection, two infections, three infections, and four infections). The table shows that the highest number of infected persons was from the category of those who had no previous infection in the family, as they were (131) infected persons, with a rate of 65.5%. The infected persons from the category of those who had one previous infection in the family were (49) infections, with a rate of 24.5%. The infected persons were from the category of those who had two previous infections in the family, where they were (11) infected persons, with a rate of 5.5%. The infected ones were from the category of those who had three previous infections in the family, their number was (6) infected persons, with a rate of 3%. As for the other (3) people, they were from the category of those who have four previous infections, and their percentage is 1.5%. This result is in agreement with the study by [19].

This proves the lack of awareness of these families about the extent of the seriousness of the disease, which compels them to stop having children or reduce childbearing when they notice the recurrence of infection in their children, especially parents who are infected with the disease or carriers of the disease.

Table (4) shows the effect of pathological history factors and the number of infections in the family on the infection of thalassemia.

Factor		Number	Infection rate
Pathological history	Less than a year	91	% 45.5
	1 3-years	79	% 39.5
	4 7-years	30	% 15
	Total	200	% 100
Number of infected case in the family	No infection	131	% 65.5
	1	49	% 24.5
	2	11	% 5.5
	3	6	% 3
	4	3	% 1.5
	Total	200	% 100

4. Conclusion

1. The heritage factor is the main cause of thalassemia.
2. Thalassemia is genetically transmitted from parents to children.
3. The disease of thalassemia is so common at the last years.
4. The disease may be caused by a genetic mutation.

Acknowledgement

I would like to send my special thanks and gratitude to middle technical university and Baquba Teaching Hospital / Hematology Center who provided me with this precious opportunity to perform this research and helped me to do a lot of researches and enabled me to know so many related topics.

References

[1] Dedousis GVZ, Mandilara GD, Boussin M. Loutradis.(2000). Hb production beta thalassemia. Wiley-liss Inc. 2000;646:151–5.
[2] Rich, S.A., Ziegler, F.D., & Grimley, P.M. (2002). Prevention of homozygous beta thalassemia by carrier screening in pregnancy. Haema, 5 (3), p.p. 242-245.
[3] CAPPELLINI, M.D., COHEN, A., PORTER, J., TAHER, A. and VIPRAKASIT, V., 2014. Guidelines for the management of transfusion dependent thalassaemia (TDT). Nicosia, Cyprus: Thalassaemia International Federation, pp. 148-149.
[4] BURMESTER, T. and HANKELN, T., 2014. Function and evolution of vertebrate globins. Acta Physiologica (Oxford, England), vol. 211, no. 3, pp. 501-514. http://dx.doi.org/10.1111/apha.12312. PMid:24811692.
[5] Sankaran VG, Nathan DG, Orkin SH. Thalassemias. In: Orkin SH, Nathan DG, Ginsburg D, Look AT, Fisher DE, Lux S (eds) Nathan and Oski's Hematology and Oncology of Infancy and Childhood. Eight Edition. Elsevier. PA, USA. 2015;715-769.
[6] Ünal Ş, Gümrük F. The Hematological and molecular spectrum of α-thalassemias in Turkey: The Hacettepe Experience. Turk J Haematol. 2015;32(2):136-43. https://doi.org/10.4274/tjh.2014.0200
[7] Aydınok Y, Oymak Y, Atabay B, et al. A national registry of thalassemia in Turkey: Demographic and disease characteristics of patients, achievements, and challenges in prevention. Turk J Haematol. 2018;35(1):12-8.
[8] Guidelines for the Management of Transfusion Dependent Thalassaemia (TDT). Cappellini MD, Cohen A, Porter J, Taher A, Viprakasit V. 3rd ed. Thalassaemia International Federation TIF Publication No. 20.
[9] Ryan K, Bain BJ, Worthington D, James J, Plews D, Mason A, et al. Significant haemoglobinopathies: guidelines for screening and diagnosis. Br J Haematol 2010;149:35-49
[10] Vichinsky EP. Alpha thalassemia major—new mutations, intrauterine management, and outcomes. Hematology Am Soc Hematol Educ Program 2009;1:35-41.
[11] Cao A. Carrier screening and genetic counselling in beta-thalassemia. Int J Hematol 2002;76(Suppl 2):105-13.
[12] Anderson LJ, Holden S, Davis B, Prescott E, Charrier CC, Bunce NH, et al. Cardiovascular T2-star (T2*) magnetic resonance for the early diagnosis of myocardial iron overload. Eur Heart J 2001;22:2171-9.
[13] Olivieri NF, Brittenham GM, Matsui D, Berkovitch M, Blendis LM, Cameron RG, et al. Iron-chelation therapy with oral deferiprone in patients with thalassemia major. N Engl J Med 1995;332:918-22.
[14] Muncie Jr HL, Campbell JS. Alpha and beta thalassemia. Am Fam Physician. 2009;80(4):339–44.
[15] Singer ST. Variable clinical phenotypes of α-thalassemia syndromes. ScientificWorldJournal. 2009;9:615–25.
[16] Mikael NA, Al-Allawi NAS. Factors affecting quality of life in children and adolescents with thalassemia in Iraqi Kurdistan. Saudi Med J. 2018;39(8):799.
[17] Hassan AN. Molecular and Some Hematological Investigations of β-thalassemic Children in Erbil Governorate. PhD, Salahaddin Univ Erbil. 2016;
[18] Abid QH, Ereiby AM. Study of relationship between thalassemia disease and blood groups, weight and some of blood parameters. Drug Invent Today. 2019;11(11).
[19] Nebeker JR, Hoffman JM, Weir CR, Bennett CL, Hurdle JF. High rates of adverse drug events in a highly computerized hospital. Arch Intern Med. 2005;165(10):1111–6.