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RESEARCH ARTICLE

THE CLINICAL PROFILES AND THE HEMATOLOGIC CHARACTERISTICS OF THALASSEMIA AMONG PATIENTS AT IBN-ALATHEER HOSPITAL IN MOSUL CITY

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ABSTRACT

Background: Thalassemia is the name of a group of genetic blood disorders. To understand how thalassemia affects the human body, you must first understand a little about how blood is made. Hemoglobin is the oxygen-carrying component of the red blood cells. It consists of two different proteins, an alpha and a beta. If the body doesn't produce enough of either of these two proteins, the red blood cells do not form properly and cannot carry sufficient oxygen. The result is anemia that begins in early childhood and lasts throughout life. Since thalassemia is not a single disorder but a group of related disorders that affect the human body in similar ways, it is important to understand the differences between the various types of thalassemia. **Aim:** The aim of the research was to determine the prevalence of thalassemia and its characteristics among patients of a thalassemia center at Ibn-Alatheer hospital, Mosul /Iraq. **Methods:** This study was conducted on 86 cases over a period of one month from November 10,2017 until December 10, 2017.A specially designed questionnaire form has been prepared; this questionnaire was filled by direct interviews with thalassemic patients taking clinical and familial history then comparing them with their clinical profiles. **Results:** During the study period of 1 month, about 86 cases were studied. All patients were thalassemia beta major .The first degree consanguinity were 47(55%) from 86 patients. While second degree 31 (36%) from 86 patients. The last percentage were non consanguinity 8(9%) from 86 patients. The age of the patients ranged between 1 – 29 years old. The mean age was found to be 10 years old. All cases displayed abnormal Hb variability. Thalassemic females were 48(56%) while males were 38(44%). **Conclusion:** In view of the high prevalence of hemoglobinopathy of thalassemia in this region, a routine premarital screening program is needed for the identification and prevention of high-risk marriages and thus, prevention of the psychosocial trauma of bearing a transfusion-dependent child for life.

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INTRODUCTION

Thalassemia is inherited blood disorder. "Inherited" means that the disorder is passed from parents to children through genes. Thalassemia causes the body to make fewer healthy red blood cells and less hemoglobin than normal. Hemoglobin is an iron-rich protein in red blood cells. It carries oxygen to all parts of the body. Hemoglobin also carries carbon dioxide (a waste gas) from the body to the lungs, where it's exhaled. People who have thalassemia can have mild or severe anemia. Anemia is caused by a lower than normal number of red blood cells or not enough hemoglobin in the red blood cells (NHLBI, 2012). The thalassemia syndromes are a group of congenital hemolytic anemias characterized by the reduced or absent synthesis of one or more globin chains of hemoglobin. (Weatherall, 2001) Hemoglobin, which carries oxygen to all cells in the body, is made of two different parts, called alpha and beta.

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When thalassemia is called "alpha" or "beta," this refers to the part of hemoglobin that isn't being made. If either the alpha or beta part is not made, there aren't enough building blocks to make normal amounts of hemoglobin. Low alpha is called alpha thalassemia. Low beta is called beta thalassemia.

Alpha Thalassemia: In alpha thalassemia, the hemoglobin does not produce enough alpha protein. To make alpha globin protein chains we need four genes, two on each chromosome 16. We get two from each parent. If one or more of these genes is missing, alpha thalassemia will result.The severity of thalassemia depends on how many genes are faulty, or mutated. Alpha thalassemia is common in southern China, Southeast Asia, India, the Middle East, and Africa (https:// www.medicalnewstoday.com/articles/263489.php).

Beta Thalassemia: We need two globin genes to make beta globin chains, one from each parent. If one or both genes are faulty, beta thalassemia will occur. Severity depends on how many genes are mutated. Beta thalassemia is more common

among people of Mediterranean ancestry. Prevalence is higher in North Africa, West Asia, and the Maldives Islands (https://www.medicalnewstoday.com/articles/263489.php). Carriers of thalassemia, are apparently healthy and normal, but may have slight anemia. In alpha thalassemia, severe anemia begins even before birth and survival past the first few hours of life is rare (Galanello *et al*., 2004). In children with beta thalassemia, symptoms appear in the first two years of life and include paleness, headache, fatigue, irritability, failure to grow, shortness of breath (Yoshida *et al*., 1990) People with thalassemia can get an overload of iron in their bodies, either from the disease itself or from frequent blood transfusions (Cianciulli, 2008), and they have an increased risk of infection, this is especially true if the spleen has been removed. (https://web.archive.org/web/20161120112707/http://www.mayoclinic.org/about-this-site/welcome), thalassemia can make the bone marrow expand, which causes bones to widen. This can result in abnormal bone structure, especially in the face and skull. Bone marrow expansion also makes bones thin and brittle, increasing the risk of broken bones (Vogiatzi *et al*., 2009). Thalassemia is often accompanied by the destruction of a large number of red blood cells and the task of removing these cells causes the spleen to enlarge. Splenomegaly can make anemia worse, and it can reduce the life of transfused red blood cells. Severe enlargement of the spleen may necessitate its removal (http://www.mayoclinic.org/diseases-conditions/enlarged-spleen/symptoms-causes/dxc-2021472219). Anemia can cause a child's growth to slow. Puberty also may be delayed in children with thalassemia (Soliman *et al*., 2014). Others diseases, such as congestive heart failure and abnormal heart rhythms, may be associated with severe thalassemia. (http://www.mayoclinic.com/health/thalassemia/DS00905/DSECTION=complications/3).

Treatment options for Thalassemia

Blood transfusions: This is done to replenish hemoglobin and red blood cell levels. Patients with moderate to severe thalassemia will have repeat transfusions every 4 months, while those with more severe disease may require transfusions every two to four weeks. Patients with mild symptoms may require occasional transfusions when they are ill or have an infection.

Iron chelation: This involves removing excess iron from the bloodstream. Sometimes blood transfusions can cause iron overload. Iron overload is bad for the heart and some other organs. Patients may be prescribed subcutaneous (injected under the skin) deferoxamine or oral (taken by mouth) deferasirox (Atheer ahmed, 2017).

Objective and Aims

- The aim was to determine the prevalence of thalassemia and its characteristics among patients in thalassemia center at Ibn-Alatheer hospital, Mosul /Iraq.
- Evaluate the clinical profile of thalassemic patients.
- Describe the appearance of a peripheral blood smear in thalassemia.
- Represent the link between thalassemia and consanguinity.

MATERIALS AND METHODS

Place of the study: Thalassemia Center at Ibn-Alatheer Hospital in Mosul city.

Time of the study: One month from November 10, 2017 until December 10, 2017.

Tools and data collection of the study: A specially designed questionnaire form has been prepared; this questionnaire was filled by direct interviews with thalassemic patients taking clinical history and familial history then comparing them with their clinical profiles**.** The questionnaire form contains information regarding name, age, residence, gender, weight, height, thalassemia type, blood film, Hb variant, first time of diagnosis, symptoms, Hb count in blood, degrees of consanguinity, number of brothers and sisters carriers or dead, complications.

Sample size: total no. 86 thalassemic patients.

Statistical analysis: The questionnaire made by Google form. The MS Excel 2007 and Numbers app on macOS and spss program were used for data entry and analysis.

RESULTS

During the study period of 1 month, about 86 cases were studied. All patients were thalassemia beta major. The first degree consanguinity were 47(55%) from 86 patients. While second degree 31 (36%) from 86 patients. The last percentage were non consanguinity 8(9%) from 86 patients. The age of the patients ranged between 1 – 29 years old. The mean age was found to be 10 years old. All cases displayed abnormal Hb variability. Thalassemic females were 48(56%) while males were 38(44%).

Table 1. Gender of the patient

Male	38	44%
Female	48	56%

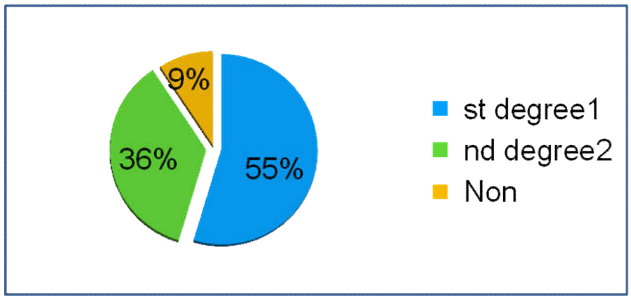

Fig 1. Parents Consanguinity of Thalassemia patients

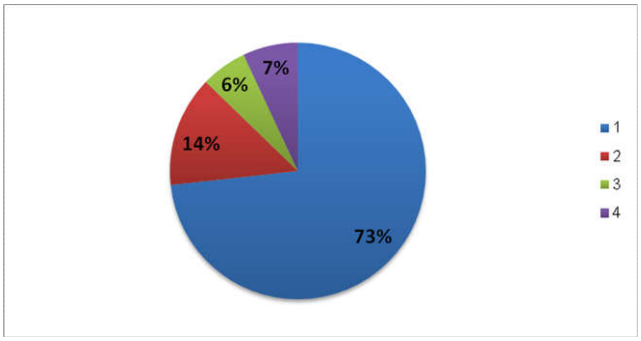

Fig 2. Blood film analysis of thalassemic patients

No.1 -represents microcytic cells
No.2- represents macrocytic cells
No.3-noromochromic normocytic cells
No.4-represents non mentioned blood films in clinical profiles

Table 2. Comparison between Hb variant types

Hb Variant Table	Hb A	Hb A2	Hb F
Average	0.153	0.023	1.83
max	78	9	98.2
min	0	0	0.6

DISCUSSION

In the research about thalassemia in Mosul city by using data from (Ibn_Al_Atheer Hospital /Thalassemia center). The study cover 86 patients with Thalassemia Major of these about 77% below age of 10 years old, generally as a result of deficiency in health care and especially for thalassemic patients during the last 3 years with embargo and war, deficiency of blood, drugs and investigations played a major role in reducing the average age of survivals. As a hereditary syndrome 91% were parents consanguinity, 9% of parents were not relative, founded in this research. All patients presented with a variable clinical picture conditions indistinguishable of beta thalassemia major requiring blood transfusions from infancy showing asymptomatic anemia. The most common Hb abnormality detected in this study was that of β thalassemia trait .Colah R *et al*. reported that nearly 1.5% of the world's population was carriers of β thalassemia (Colah *et al*., 2010). Several studies reveal that in most parts of India, β thalassemia trait was the commonest Hb disorder. A study conducted by Madan N and coinvestigators showed the overall gene frequency of β thalassemia trait reported in northern and western India was 4.05% (Madan *et al*., 2010). The prevalence of β thalassemia trait has been reported to be as high as 10.38% in the rural parts of West Bengal and in central India, it was found to be 9·59%. (Dolai *et al*., 2012; Chatterjee *et al*., 2010). The 643,580 research subjects in China included in the 16 studies examined can generally be divided into neonates, children and adults. Although the age ranges of the different studies varied widely, subgroup analysis based on age was not performed because thalassemia is an inherited disease, and the carrying rates of different age groups are consistent in the same area. Five included studies (Pan *et al*., 2007; Zeng and Huang, 1987; Zhang *et al*., 2010; Chen *et al*., 2004; Cai *et al*., 2002). In which the reported cases were only neonates attempted to determine the prevalence of α-thalassemia. The neonates were randomly selected such that the rates of thalassemia were representative and could be compared with the results for children or adults. Surveys on thalassemia in China began in the 1980s. In 1987, Zeng calculated that the nationwide incidence of α-thalassemia and β-thalassemia was 2.64% and 0.66%, respectively (Zeng and Huang, 1987).

In Egypt there are 10,000 registered thalassemia cases and more than 20,000 non-registered cases. 95% are beta thalassemia major; 5% are thalassemia intermedia or hemoglobin H disease. We do not have any cases of e beta thalassemia. Trait carrier rate ranges between 5.5%to >9.5% based on a study carried out on 5000 normal candidates in 5 governorates in Egypt. (Dr.mohamed, 2009). A study on 7,837 consecutive cord blood samples from April 2005 to March 2007 in Oman showed that 429 neonates were carriers for HbS (5.48 %) (Alkindi *et al*., 2011). The samples also showed the presence of Hb variants indicating the presence of α-thalassemia genes in 48.5 % of the neonates though no case of HbH was detected (Alkindi *et al*., 2010). In Saudi Arabia, a national PMS for the most prevalent disorders (thalassemia and sickle cell anemia) was initiated in 2004 on a voluntary basis in all regions of Saudi Arabia and became compulsory in 2005 where all couples contemplating marriage have to be screened for carrier status before issuing their marriage certificate (Al-Sulaiman *et al*., 2010). B-thalassemia is a common inherited hematological disorder in Iraq, with an average prevalence of carriers of about 4 % and an estimated 15,000 registered thalassemia major/intermedia patients throughout the country. The huge burden on the already precarious resources by this huge number of patients has led the health authorities to search for a prevention strategy. In the Kurdistan region in the north of the country where around 20 % of the population of Iraq reside, pilot studies were initiated in 2006 to determine the service indicators for β-thalassaemia prevention and the molecular basis of this condition in the region (Al-Allawi *et al*., 2006; Al-Allawi and Al-Dousky, 2010; Jalal *et al*., 2010; Jalal *et al*., 2008).

These studies showed that a PMS was feasible and after consultation with the religious scholars in the region, the local government took a decision to make premarital screening for hemoglobinopathies mandatory by law (Al-Allawi, 2008). The program of β-thalassaemia premarital screening for carrier detection was implemented in 2008 by all health authorities in the Kurdistan region, based on the principles of premarital screening, counseling and PND. Preliminary results of the first 3 years (2008–2010) revealed that more than 115,000 individuals were screened and 3.7/1,000 of the couples were identified to be at risk of having children with a major hemoglobinopathy. Of couples at risk, 91 % proceeded with their marriage as scheduled. Of the latter 38 % sought PND in early pregnancy by chorionic villus sampling. The remaining couples either did not seek PND or came late in second trimester, when it is not permissible to perform selective abortion. All couples who underwent PND and had an affected fetus chose to terminate pregnancy (Al-Allawi, 2011). The main problems facing the program included the limited awareness of the population on inherited disorders, the high rate of consanguineous marriage (24–27 %), the cost of PND (which is not fully covered by the local authorities) and the limited time between mandatory testing and the actual marriage limiting annulations of marriage plans due to social reasons. Moreover, some couples were not convinced by the results of the screening test given to them. On the other hand, religious beliefs had a limited impact on decisions related to the selective termination of pregnancies with affected fetus.

Conclusion and recommendation

In view of the high prevalence of hemoglobinopathy of thalassemia in this region, a routine premarital screening program is needed for the identification and prevention of high-risk marriages and thus, prevention of the psychosocial trauma of bearing a transfusion-dependent child for life.. Consanguineous marriage in rural areas is common in Mosul city, also the availability of investigations and diagnosing carriers is less, so affected patients from rural areas are more in the research. Recently medical investigations including thalassemia prior marriage were obligatory this will lead to decrease numbers of thalassemic patients in the future. We hope to add Healthy People 2020 Program Includes Thalassemia Objectives to Mosul city (http://www.thalassemia.org/healthy-people-2020-program-includes-thalassemia-objectives-2/). This program carries a lot of health changes and it will limit the prevalence of thalassemia**.**

REFERENCES

"What Are Thalassemias?". NHLBI. 3 July 2012. /articles/ 263489.php. By Christian Nordqvist, Mon 15 May 2017.

Al-Allawi, N. 2008. The preventive program for hemoglobinopathies in Duhok: an option or a necessity. *Duhok Med J.,* 2(1).

Al-Allawi, N. 2011. Molecular characterization of inherited haematological disorders in Iraqis: trying to bridge the gap. *HUGO.,* 5:124.

Al-Allawi, NA. and Al-Dousky, AA. 2010. Frequency of haemoglobinopathies at premarital health screening in Dohuk, Iraq: implications for a regional prevention programme. *East Mediterr Health J.,* 16(4):381-5.

Al-Allawi, NA., Jubrael, JM. and Hughson, M. 2006. Hemoglobin, Molecular characterization of beta-thalassemia in the Dohuk region of Iraq; 30(4):479-86.

Alkindi, S., Al-Zadjali, S., Al-Madhani, A., Daar, S., Al-Haddabi, H., Al-Abri, Q., Gravell, D., Berbar, T., Pravin, S., Pathare, A. and Krishnamoorthy, R. 2010. Forecasting hemoglobinopathy burden through neonatal screening in Omani neonates. Hemoglobin. 34:135–144. doi: 10.3109/ 03630261003677213.

Alkindi, S., Pathare, A., Al-Madhani, A., Al-Zadjali, S., Al-Haddabi, H., Al-Abri, Q., Gravell, D., Mathew, M. and Krishnamoorthy, R. 2011. Neonatal Screening: mean haemoglobin and red cell indices in cord blood from Omani neonates. *Sultan Qaboos Univ Med J.,* 11:462–469.

Al-Sulaiman, A., Saeedi, M., Al-Suliman, A. and Owaidah, T. 2010. Postmarital follow-up survey on high risk patients subjected to premarital screening program in Saudi Arabia. *Prenat Diagn.,* 30:478–481.

Cai, R., *et al*., 2002. Prevalence survey and molecular characterization of α and β thalassemia in Liuzhou city of Guangxi. *Chin J Epidemiol*., 23:281–285.

Center for disease control and prevention May 24, 2016 Article online on:https://www.medicalnewstoday.com

Chatterjee, N., Mishra, A., Soni, R., Kulkarni, H., Mamtani, M. and Shrivasatava, M. 2010. Bayesian estimates of the prevalence of β-thalassemia trait in voluntary blood donors of central India: A survey. *Hemoglobin*, 34:548–60.

Chen, PY., Chen, YJ., Xia, YY. and Qin, CR. 2004. Molecular epidemiological study of α-thalassemia in Shaoguan city of Guangdong. *Chin J Prim Med Pharm.,* 11:518–520.

Cianciulli, P. October 2008. "Treatment of iron overload in thalassemia". Pediatr Endocrinol Rev. 6 (Suppl 1): 208–13. PMID 19337180.

Colah, R., Gorakshakar, A. and Nadkarni, A. 2010. Global burden, distribution and prevention of β-thalassemias and hemoglobin E disorders. *Expert Rev Hematol.,* 3:103–17.

Dolai, TK., Dutta, S., Bhattacharyya, M. and Ghosh, MK. 2012. Prevalence of hemoglobinopathies in rural Bengal, India. *Hemoglobin,* 36:57–63.

Dr.mohamed, 9th Cooley'sAnemiaSymposium, representatives from CAF, Cairo University in Egypt, November 10, 2009.

Galanello, R., Perseu, L., Perra, C., Maccioni, L., Barella, S., Longinotti, M., Cao A. and cazzola, M. 2004. Somatic deletion of the normal beta-globin gene leading to thalassemia intermedia in heterozygous beta-thalassemia patients. *Br. J. Hematol*., 127: 604-606.

Healthy People 2020 Program Includes Thalassemia Objectives, June 13, 2011. online on: http://www.thalassemia.org/healthy-people-2020-program-includes-thalassemia-objectives-2/.

http://www.mayoclinic.org/diseases-conditions/enlarged-spleen/symptoms-causes/dxc-2021472219 November 2016.

Jalal, SD., Al-Alawi, NA., Faraj, AH. and Ahmed, NH. 2008. Prevalence of hemoglobinopathies in Sulaimani-Iraq. *Dohuk Med J.,* 2(1):71–

Jalal, SD., Al-Allawi, NA,. Bayat, N., Imania,n H., Najmabadi, H. and Faraj, A. 2010. β-Thalassemia mutations in the Kurdish population of northeastern Iraq. Hemoglobin. 34(5):469-76.

Madan, N., Sharma, S., Sood, SK., Colah, R. and Bhatia, LH. 2010. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. *Indian J Hum Genet.,* 16:16–25.

Mayoclinic online on: https://web.archive.org/web/20161120112707/http://www.mayoclinic.org/about-this-site/welcome 20 November 2016

Pan, HF., *et al*., 2007. Current status of thalassemia in minority populations in Guangxi, China. *Clin Genet.,* 71:419–426. doi: 10.1111/j.1399-0004.2007.00791.x.

Soliman, Ashraf, T., Kalra and Sanjay, 2014. De Sanctis, Vincenzo."Anemia and growth". *Indian Journal of Endocrinology and Metabolism*.

Thalassemia complications online on : http://www.mayoclinic.com/health/thalassemia/DS00905/DSECTION=complications/3 October 2011.

The university of Georgia Atheer ahmed 1330129 Hematology THALASSEMIA Feb 21, 2017.

Vogiatzi, Maria, G., Macklin, Eric, A., Fung, Ellen B., Cheung, Angela, M., Vichinsky, Elliot, Olivieri, Nancy, Kirby, Melanie, Kwiatkowski, Janet, L., Cunningham, Melody, Holm, Ingrid, A., Lane, Joseph, Schneider, Robert; Fleisher, Martin, Grady, Robert, W., Peterson, Charles, C. and Giardina, (March 2009). *Patricia J.,* "Bone Disease in Thalassemia: A Frequent and Still Unresolved Problem.

Weatherall, DJ. 2001. Phenotype-genotype relationships in monogenic disease: lessons from the thalassaemias. Nat Rev Genet., 2(4):245-55.

Yoshida, N., Horikoshi, A., Kanemaru, M., Shimada, H., Takeuchi, J., Ohshima, T., Horie, T., Tsuchiya, T., Kamei, K., Ishikawa, S., *et al*., 1990. An erythremia with acquired HbH disease and chromosomal abnormality. Rinsho Ketsueki. 31: 963-968.

Zeng, YT. and Huang, SZ. 1987. Disorders of haemoglobin in *China. JMedGenet.,* 24:578–583.doi:10.1136/jmg.24. 10.578.

Zhang, CM, *et al*., 2010. Molecular epidemiology survey of α-thalassemia in Zhongshan city. *Chinese Journal of Child Health Care.,* 18:361–364.

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