ISSN: 2457-0400

Impact Factor: 7.675

WORLD JOURNAL OF ADVANCE HEALTHCARE RESEARCH

www.wjahr.com

Volume: 10, Issue: 8

Page N. 112-117

Year: 2026

Original Article

Coden USA: WJAMA3

PREVALENCE OF THALASSEMIA AND CLINICODEMOGRAPHIC PROFILE OF THALASSEMIC PATIENTS IN NINEVEH PROVINCE

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Article Received: 24 June 2026 **Article Revised: 14 July 2026** **Article Published: 01 August 2026**

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DOI: https://doi.org/10.5281/zenodo.21702551

How to cite this Article: *Heba Qasem Fathi Ali, Dr. Ammar H. Yahia (2026). Prevalence Of Thalassemia And Clinicodemographic Profile Of Thalassemic Patients In Nineveh Province. World Journal of Advance Healthcare Research, 10(8), 112–117.

ABSTRACT

Background: Thalassemia is a common inherited hemoglobin disorder caused by mutations in the α- or β-globin genes, resulting in impaired globin chain synthesis and ineffective erythropoiesis. **Aim:** This study aimed to determine the epidemiological and clinical characteristics of patients with thalassemia in Nineveh Province, Iraq. A total of 604 patients with diagnosed thalassemia attending the Thalassemia Center were included. **Methods:** A descriptive cross-sectional study included 604 confirmed thalassemia patients (January–June 2025). Data were collected through patient interviews and hospital registry records to assess demographic and clinical characteristics and estimate prevalence and incidence rates. **Results:** The prevalence was 23 per 100,000 population, and the six-month incidence rate was 0.72 per 100,000 population. Females accounted for 51.32% of the study population, and 44.87% of patients were aged 10–20 years. β-thalassemia major was the predominant subtype (69.7%). Parental consanguinity was reported in 77.32% of cases. Iron chelation therapy was received by 88.41% of patients. The most common complications were splenomegaly and growth retardation. **Conclusions**: The prevalence of thalassemia was 23 per 100,000 population, with ongoing new cases. Thalassemia major was the predominant subtype, affecting mainly adolescents, with most patients requiring iron chelation therapy. High parental consanguinity and common complications, including splenomegaly and growth retardation, highlight the need for strengthened prevention and comprehensive disease management.

KEYWORDS: Thalassemia; β-thalassemia major; Epidemiology; Prevalence; Cross-sectional study; Consanguinity; Iron chelation therapy.

INTRODUCTION

Thalassemia is one of the most common inherited hemoglobin disorders worldwide. It results from mutations affecting α- or β-globin synthesis, leading to ineffective erythropoiesis, chronic hemolytic anemia, iron overload, and progressive organ damage.[1] Although traditionally concentrated in the Mediterranean region, Middle East, South Asia, and Africa, population migration has expanded its global distribution.[2] In Iraq, β-thalassemia major remains the predominant subtype, with increasing prevalence over recent years.[3] Thalassemia is classified into α-thalassemia and β-thalassemia. β-thalassemia includes major, intermedia, and minor forms and may also be classified as transfusion-dependent or non-transfusion-dependent. α-thalassemia ranges from silent carrier status to hemoglobin Bart's hydrops fetalis. Other less common forms include sickle-thalassemia and several rare globin gene variants.[4–10] Thalassemia remains a major public health concern in Iraq, with 13,390 registered cases.[11]

AIM

To determine the epidemiological and clinical characteristics of patients with thalassemia in Nineveh Province, Iraq.

PATIENTS AND METHODS

Ethical and Administrative Considerations

The Medical Ethical Committee of the Arab Board of Health Specializations approved this study. An official

agreement was obtained from the Ministry of Health and Nineveh Health Directorate in Mosul city before conducting the present study. Verbal consent was taken from all patients or their caregivers who were informed about the objectives of the study before starting it.

Study Setting
This study was conducted at the Thalassemia Center of Al-Hadbaa Specialized Hospital for Hematological Disorders and Bone Marrow Transplantation in Mosul, Iraq. Established in 2023, The Thalassemia Center is the sole referral center for thalassemia in Nineveh Province and provides comprehensive healthcare services to patients throughout the province.

Study Period
The study was conducted over six months, from January 2,2025, to July 1, 2025.

Study Design
A descriptive cross-sectional study design was adopted in order to achieve the objectives of this study.

Study Sample
The study employed a non-probability convenience sampling technique. The sample included both newly and previously diagnosed patients with thalassemia who attended the Thalassemia Center at Al- Hadbaa Hospital during the study period for management and follow-up.

Case Definition
Cases were identified from patients attending the hospital's Thalassemia Center. All patients had been diagnosed by a specialist based on clinical evaluation and confirmed through hemoglobin electrophoresis.

Inclusion Criteria
Patients of any age and sex with a confirmed diagnosis of thalassemia who attended the center during data collection.

Exclusion Criteria
Patients with unconfirmed diagnoses, incomplete data, who refused to provide consent, diagnosis other than thalassemia, or who were from another province were excluded from the study.

Data Collection Tools
Data were collected through direct interviews using a structured questionnaire, following verbal consent and with full respect for patient privacy and confidentiality. No clinical examinations or laboratory data were collected as part of this study. In addition, certain information, such as thalassemia subtype, was obtained from the center's statistical records.

Statistical Analysis
Data were coded and analyzed using Microsoft Excel (2021). Descriptive statistics were used to summarize the data. The prevalence rate was calculated per 100,000 population using the total number of registered confirmed thalassemia cases in the Thalassemia Center database (n = 1,025) and the official population estimate of Nineveh Province as the denominator (4,456,522). The six-month incidence rate was calculated per 100,000 population using the number of newly diagnosed cases recorded during the study period (n = 32) and the same population denominator.

Other statistical analyses were performed using the study sample (n = 604). Continuous variables were presented as mean ± SD, while categorical variables were expressed as frequencies and percentages. The mean age was estimated using the midpoints of the age categories. Because many patients did not provide medical records or laboratory results during follow-up, thalassemia classification and complication data were obtained from the Thalassemia Center's statistical records, which were manually reviewed to include only eligible study participants. For the analysis of complications, minor thalassemia cases were excluded.

RESULTS

Descriptive data analysis
The estimated prevalence of thalassemia was 23 per 100,000 population, while the six-month incidence rate was 0.72 per 100,000 population. A total of 604 patients were included in the study. Females accounted for 51.32% of the study population, and the largest age group was 10–20 years (44.87%). The estimated mean age was 17.0 ± 9.43 years.

Table 1: Distribution of age and sex among study sample.

Age Group(years)	Females No. (%)	Males No. (%)	Total No. (%)
<10.	63 (10.43)	71 (11.76)	134 (22.19)
10-20.	148 (24.5)	123 (20.36)	271 (44.87)
21-30.	73 (12.09)	75 (12.42)	148 (24.5)
31-40.	22 (3.64)	18 (2.98)	40 (6.62)
>40.	4 (0.66)	7 (1.16)	11 (1.82)
Total	310 (51.32)	294 (48.68)	604 (100)

Clinical characteristics data analysis

β-thalassemia major was the predominant subtype (69.7%), followed by β-thalassemia intermedia (22.52%).

Table 2: Distribution of thalassemia type among the study sample.

Thalassemia type		Females No. (%)	Males No. (%)	Total No. (%)
Thalassemia major		204 (33.77)	217 (35.93)	421 (69.7)
Thalassemia intermedia	β	80 (13.25)	56(9.27)	136 (22.52)
	α	6 (0.99)	7(1.16)	13 (2.15)
Sickle thalassemia		13 (2.15)	7 (1.16)	20 (3.31)
Thalassemia minor		7 (1.16)	7 (1.16)	14 (2.32)
Total		310 (51.32)	294 (48.68)	604 (100)

Among adult patients, 21.71% were married and 16.28% had children.

Table 3: Distribution of marital status and parenthood according to thalassemia type.

Thalassemia type		Married No. (%)		Parenthood No. (%)	
		Females No. (%)	Males No. (%)	Females No. (%)	Males No. (%)
Thalassemia major		6 (2.33)	14 (5.43)	2 (0.78)	8 (3.1)
Thalassemia Intermedia	β	14 (5.43)	10 (3.88)	12 (4.65)	8 (3.1)
	α	3 (1.16)	1 (0.39)	3 (1.16)	1 (0.39)
Thalassemia minor		4 (1.55)	4 (1.55)	4 (1.55)	4 (1.55)
Sickle Thalassemia		--	--	--	--
Total		27 (10.47)	29 (11.24)	21 (8.14)	21 (8.14)
		56 (21.71)		42 (16.28)	

Family history and genetic data analysis

Figure 1 demonstrates that parental consanguinity was reported in 77.32% of patients.

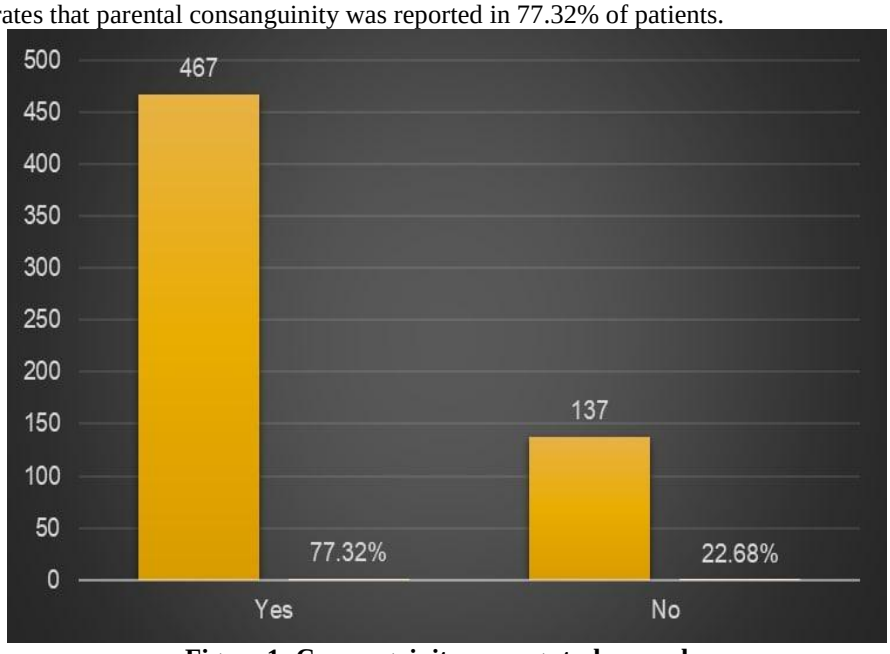

Figure 1: Consanguinity among study sample.

Medical history and management data analysis

Iron chelation therapy was received by 88.4% of patients.

Table 4: Distribution of study sample according to iron chelation therapy (ICT).

ICT	Females No. (%)	Males No. (%)	Total
Received ICT	274 (45.36)	260 (43.05)	534 (88.41)
Not received ICT	36 (5.96)	34 (5.63)	70 (11.59)

Regarding surgical history, 23.2% underwent splenectomy, whereas 67.1% had no history of splenectomy or cholecystectomy.

Table 5: Distribution of study sample according to surgical history.

Operation	Females No. (%)	Males No. (%)	Total
Splenectomy	56 (9.27)	84 (13.91)	140 (23.2)
Cholecystectomy	12 (1.99)	2 (0.33)	14 (2.32)
Splenectomy and Cholecystectomy	30 (4.97)	15 (2.48)	45 (7.45)
No Splenectomy and/or Cholecystectomy	212 (35.1)	193 (31.95)	405 (67.1)
Total	310 (51.32)	294 (48.68)	604 (100)

The most common complications were splenomegaly, growth retardation, and osteoporosis.

Table 6: Distribution of common complications among patients with clinically significant thalassemia.

Complications	Thalassemia types				Total
	Major No. (%)	Intermedia No. (%)		Sickle thalassemia No. (%)	
		β	α		
Splenomegaly	166 (28.14)	32 (5.42)	3 (0.51)	3 (0.51)	204 (34.57)
Delayed growth	155 (26.27)	24 (4.07)	1 (0.17)	6 (1.02)	186 (31.53)
Osteoporosis	7 (1.19)	20 (3.39)	2 (0.34)	5 (0.85)	34 (5.76)
Cardiac complication	13 (2.2)	2 (0.34)	0	1 (0.17)	16 (2.71)
Diabetes mellitus	12 (2.03)	2 (0.34)	0	0	14 (2.37)
Thyroid disorders	9 (1.53)	0	0	0	9 (1.53)
Viral hepatitis	13 (2.2)	0	0	0	13 (2.2)

Percentages were calculated after excluding patients with thalassemia minor (n = 590).

DISCUSSION

The present study provides updated epidemiological and clinical data on thalassemia in Nineveh. The estimated prevalence (23 per 100,000 population) and six-month incidence rate (0.72 per 100,000 population) were lower than those reported from Erbil and the national Iraqi estimates, possibly reflecting regional differences in healthcare access, case ascertainment, and diagnostic practices.[3,12]

The study population showed an almost equal sex distribution, consistent with the autosomal recessive inheritance of thalassemia. Most patients were aged 10–20 years, similar to regional findings and the Turkish National Thalassemia Registry.[12,13,14] β-thalassemia major was the predominant subtype, in agreement with previous reports from Iraq and Syria.[15,16]

Marriage and parenthood were more common among patients with β-thalassemia intermedia than those with thalassemia major, consistent with findings from Wasit and likely reflecting the milder clinical course and better survival of patients with β-thalassemia intermedia.[17]

Parental consanguinity was reported in 77.32% of patients, closely matching findings from Nineveh and Babylon, highlighting the continuing contribution of consanguineous marriage to the persistence of thalassemia in Iraq.[18,19]

Iron chelation therapy was administered to 88.4% of patients, reflecting good treatment coverage and adherence to current treatment protocols. This rate was comparable to that reported in Syria and higher than that observed in Wasit, where a proportion of patients were not receiving chelation therapy.[16,20]

Splenectomy was the most common surgical intervention (23.2%), with a frequency comparable to that reported from Babylon.[18]

The most common complications were splenomegaly (34.57%) and growth retardation (31.53%),

predominantly among patients with β-thalassemia major. Although these complications were less frequent than those reported from Wasit, they remain clinically important and support the need for comprehensive long-term multidisciplinary care.[20]

Limitations

The study was limited by incomplete patient follow-up, restricted data collection periods, and the lack of available records for thalassemia minor cases. In addition, missing laboratory results and incomplete medical records required reliance on registry data for some classifications and complications, which may have led to underestimation of certain findings. Furthermore, undiagnosed or unregistered cases in the community may have resulted in an underestimation of the true prevalence of thalassemia, and the single-center design may limit the generalizability of the findings.

CONCLUSIONS

Thalassemia remains a significant public health problem in Nineveh Province, with β-thalassemia major as the predominant subtype. High parental consanguinity and common complications, including splenomegaly, growth retardation, and osteoporosis, highlight the need for strengthened prevention and comprehensive disease management.

Recommendations

Strengthen premarital screening and genetic counseling programs.

Improve comprehensive multidisciplinary care and long-term follow-up for patients with thalassemia.

Conduct multicenter studies to monitor disease trends and evaluate preventive strategies.

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