



## PREVALENCE AND GENDER DISTRIBUTION OF THALASSEMIA TRAITS IN COUPLES UNDERGOING PREMARITAL SCREENING AT AL-MADAEN GENERAL HOSPITAL IN BAGHDAD-IRAQ

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Article Received: 02 February 2026

Article Revised: 23 February 2026

Article Published: 01 March 2026



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DOI: <https://doi.org/10.5281/zenodo.18815410>

**How to cite this Article:** <sup>1</sup>Dr. Bahaa Dawood Abdulkareem (M.B.Ch.B., C.A.B.H.S. /FM), <sup>2</sup>Dr. Hiba Khalid Ali (M.B.Ch.B., F.I.C.M.S./Pathology) (2026). Prevalence And Gender Distribution Of Thalassemia Traits In Couples Undergoing Premarital Screening At Al-Madaen General Hospital In Baghdad-Iraq. World Journal of Advance Healthcare Research, 10(3), 134–138.



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### ABSTRACT

**Background:** Thalassemia is a heterogeneous group of inherited hemoglobin disorders caused by quantitative defects in globin chain synthesis, resulting in chronic anemia of different severity. Thalassemia is a significant public health challenge in Iraq, preventive methods, particularly premarital screening programs, have been implemented to detect carriers early on and provide appropriate genetic counseling. **Objectives:** To determine the prevalence and gender distribution of thalassemia trait among couples undergoing premarital screening in Baghdad, Iraq. **Methods:** This is a descriptive cross-sectional study carried out during the period from 2<sup>nd</sup> of January 2024 till the 27<sup>th</sup> of December 2024 at Al-Madaen General hospital. All individuals with microcytic anemia who completed the premarital screening process and had complete hematological data were included in the study. Individuals with already known hemoglobinopathies, those with inadequate laboratory data, and those who declined to participate were excluded. **Results:** Among 998 screened individual (499) couples, 149 individuals were identified as having thalassemia trait, corresponding to an overall prevalence of 14.9%. The mean age  $\pm$  standard deviation of the study participants is  $25.87 \pm 4.75$  years. Within these couples, only male carrier was prevalent among 71 (14.23%), while female carrier was prevalent among 38 (7.26%) and both partner (high risk) among 20 (4%). **Conclusion:** Continuous use of specific screening approaches (such as microcytosis-based selection) in conjunction with traditional premarital screening. The study highlights the importance of improved hematological screening for identifying silent carriers and expanding genetic counseling reach.

**KEYWORDS:** Beta thalassemia, Counseling, Mariage, Screening.

### 1. INTRODUCTION

Thalassemia is a heterogeneous group of inherited hemoglobin disorders caused by quantitative defects in globin chain synthesis, resulting in chronic anemia of different severity.<sup>[1]</sup> Thalassemia is widely spread over the Mediterranean region, the Middle East, South Asia, and portions of Africa, where carrier rates remain high due to genetic, cultural, and consanguinity reasons.<sup>[2-3]</sup> Although people with thalassemia trait are frequently asymptomatic, they can cause disease transmission and they are at risk of bearing children with severe

transfusion-dependent thalassemia if both parents are carriers.<sup>[4-5]</sup>

Thalassemia is a significant public health challenge in Iraq, putting additional strain on healthcare resources for long-term management such as blood transfusions, iron chelation therapy, and complication management.<sup>[6]</sup> Preventive methods, particularly premarital screening programs, have been implemented to detect carriers early on and provide appropriate genetic counseling. These programs seek to lower the prevalence of thalassemia

major by facilitating educated marital and reproductive decisions.<sup>[7]</sup>

Couple screening prior to marriage is particularly important since it enables for the detection of carrier states in both partners and facilitates counseling on potential genetic concerns.<sup>[8]</sup> Despite the establishment of premarital screening services in Baghdad, thorough local data on the prevalence and demographic distribution of thalassemia characteristics among screened couples is still limited. Detailed epidemiological data are required to evaluate the efficacy of current screening programs, as well as to guide public health policy and future preventative efforts.<sup>[9]</sup> As a result, the current study was done to determine the prevalence of thalassemia trait among couples undergoing premarital screening in Baghdad, Iraq, as well as to examine the gender distribution of detected carriers within this population.

## 2. PATIENTS AND METHODS

This is a descriptive cross-sectional study carried out during the period from 2<sup>nd</sup> of January 2024 till the 27<sup>th</sup> of December 2024 at Al-Madaen General hospital. The study population consisted of couples referred for standard premarital screening as part of the national health program who accidentally discovered to have microcytic anemia in one or both partners. Participants were adults of marriageable age and were apparently healthy at the time of screening.

All individuals with microcytic anemia who completed the premarital screening process and had complete hematological data were included in the study. Individuals with already known hemoglobinopathies,

those with inadequate laboratory data, and those who declined to participate were excluded.

Demographic information, including gender, was collected using standardized screening forms. Each participant underwent routine hematological evaluation as part of the premarital screening protocol. The screening for partial thalassemia was based on recognized hematological criteria, which included full blood count parameters such mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), and red blood cell count. Participants with suggestive indices underwent confirmatory diagnostic tests in accordance with local guidelines. Individuals who met the diagnostic criteria were considered as having partial thalassemia.

The data were analyzed using descriptive statistical methods. Continuous variables were summarized using means and standard deviations where applicable, while categorical variables were reported as frequencies and percentages. Partial thalassemia prevalence was computed as the proportion of detected carriers in the overall screened population. The gender distribution was evaluated descriptively.

## 3. RESULTS

A total of 499 couples underwent premarital screening during the study period and discovered to have microcytic anemia in one or both partners. Among the screened population, 149 individuals were identified as having thalassemia trait, corresponding to an overall prevalence of 14.9% (figure 1). The mean age  $\pm$  standard deviation of the study participants is  $25.87 \pm 4.75$  years.

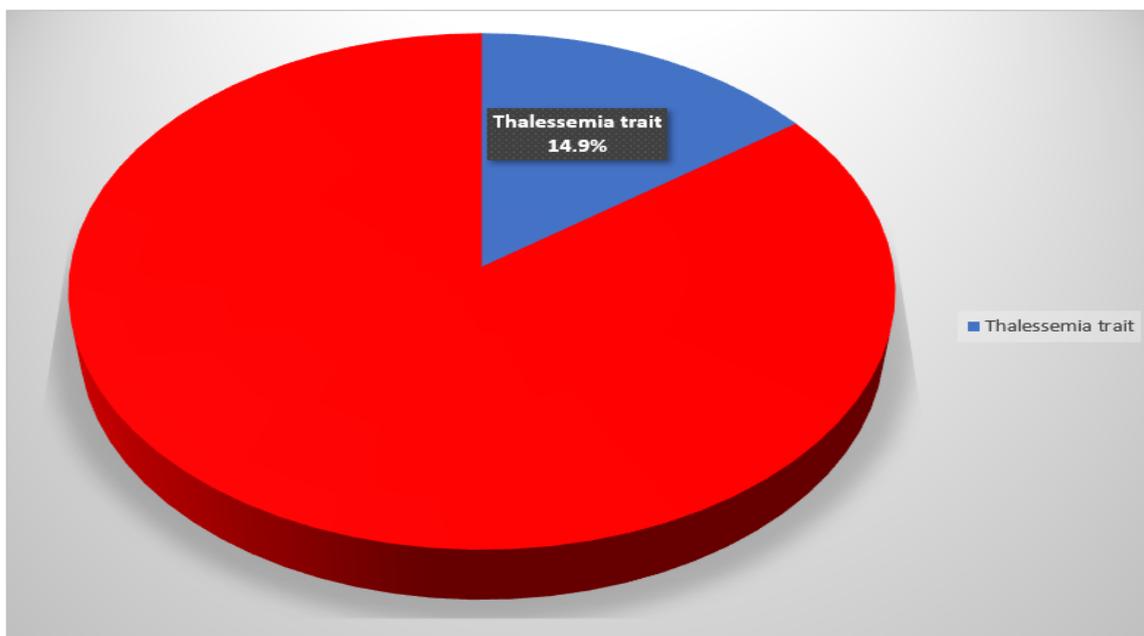
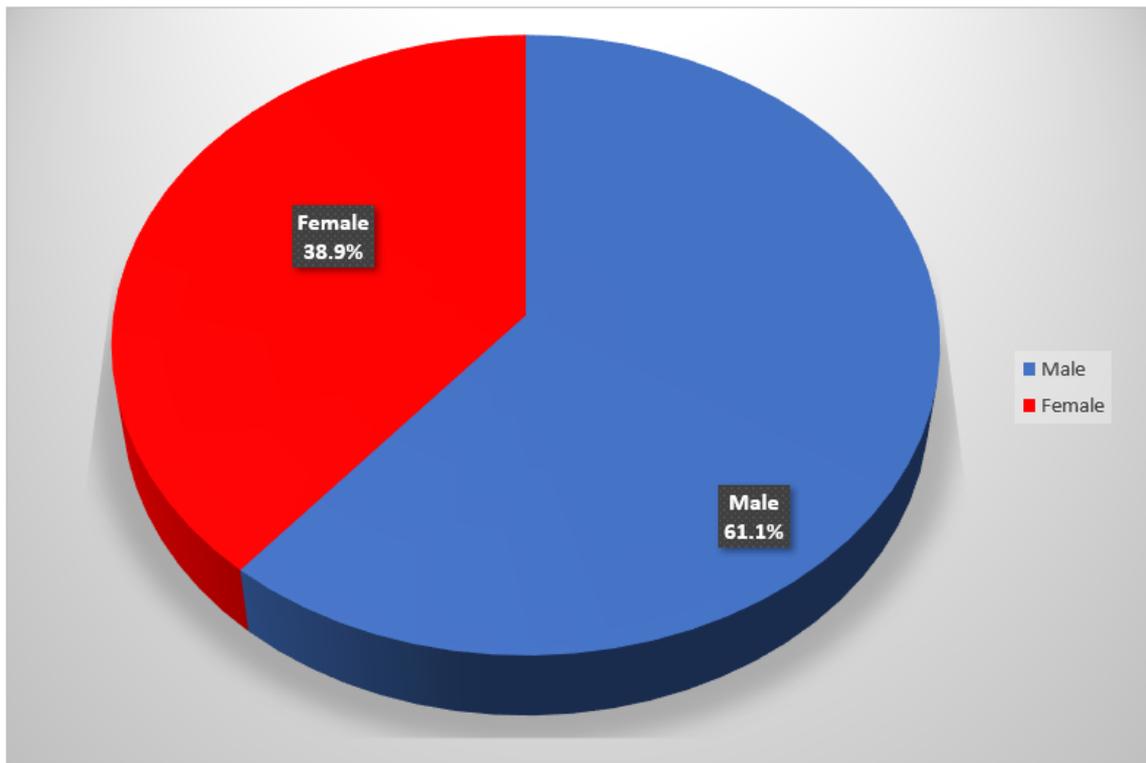


Figure 1: Distribution of the study participants according to their screening results.

Analysis of gender distribution revealed that 91 (61.1%) of the carriers were males, whereas 58 (38.9%) were females. As shown in figure 2.



**Figure 2: Distribution of the study participants according to their gender.**

Table 1 shows distribution of the study participants according to their thalassemia carrier status. Only male carrier was prevalent among 71 (14.23%), while female

carrier was prevalent among 38 (7.26%) and both partner (high risk) among 20 (4%).

**Table 1: Distribution of the study participants according to their thalassemia carrier status.**

Variable	Number	Percentage
Both partners non-carriers	370	74.15%
Only male carrier	71	14.23%
Only female carrier	38	7.62%
Both partners carriers	20	4.0%

Table 2 shows distribution of the study participants according to consanguinity relationships. It's evident that among 155 (31.06%) out of 499 couples having positive consanguinity. Male carrier was found in 21 (4.22%)

couple, while female carrier was prevalent in 11 (2.20%) couple and both partner carriers was prevalent in 12 (2.40%) couples.

**Table 2: Distribution of the study participants according to their consanguinity relationships.**

Variable	Number	Percentage
Both partners non-carriers	111	22.24%
Only male carrier	21	4.22%
Only female carrier	11	2.20%
Both partners carriers	12	2.40%
Total consanguinity couple	155	31.06%

Table 3 shows hemoglobin levels by carrier status and gender. Female carrier showed lower hemoglobin mean level ( $11.5 \pm 1$  g/ dL), followed by both gender carrier showed hemoglobin mean level of ( $11.8 \pm 1$  g/ dL).

**Table 3: Hemoglobin level by carrier status and gender.**

Variable	Number	Mean $\pm$ standard deviation (g/dL)
Male carrier	91	12.2 $\pm$ 1.1
Female carrier	58	11.5 $\pm$ 1
Male non carrier	408	14.3 $\pm$ 0.9
Female non carrier	441	13.5 $\pm$ 0.8
Both genders carrier	40	11.8 $\pm$ 1
Both genders non carrier	458	14.1 $\pm$ 0.9

#### 4. DISCUSSION

The current study assessed thalassemia carrier status in 499 premarital couples (998 individuals) with microcytic anemia following regular premarital screening in Baghdad. By restricting the study population to people with microcytosis, the screening technique specifically targeted a high-risk hematological category where thalassemia traits are more likely to be discovered. This approach explains the relatively high overall carrier prevalence of 14.9%, which is higher than rates reported in unselected premarital groups and reflects the diagnostic yield of targeted screening among people with suspicious red blood cell indices. Several studies using targeted screening based on microcytosis have produced comparable results, supporting the current study's conclusions. In Iran, Miri-Moghaddam *et al* found that screening individuals with low mean corpuscular volume and mean corpuscular hemoglobin increased the detection rate of  $\beta$ -thalassemia carriers by 15-20%, compared to lower rates in unselected premarital populations.<sup>[10]</sup> Similarly, data from the Saudi premarital screening program revealed that the majority of hemoglobinopathy carriers were found among people with microcytic indices, with carrier frequencies exceeding 10% in this category.<sup>[11]</sup> Turkish studies have also proven that unexplained microcytosis is a major predictor of thalassemia phenotype, and that hematological preselection significantly enhances screening efficiency.<sup>[12]</sup> In Egypt, El-Beshlawy *et al.* found that screening patients with microcytic anemia yielded higher diagnostic results for both  $\alpha$ - and  $\beta$ -thalassemia compared to the general population.<sup>[13]</sup> The World Health Organization's international recommendations support this approach by emphasizing that targeted screening of high-risk groups particularly those with microcytosis or hypochromia, is more cost-effective and diagnostically efficient than universal screening in regions with moderate to high thalassemia prevalence.<sup>[14]</sup>

Although thalassemia is inherited in an autosomal recessive pattern, this study found a larger proportion of male carriers (61.1%) than female carriers. This finding may be influenced by the microcytic selection criteria as well as disparities in iron deficiency prevalence among females, which can make thalassemia detection difficult using only hematologic indicators. Which is consistent to studies from the Middle East and South Asia have revealed that screening programs concentrating mostly on hematologic indices (such as microcytosis and

hypochromia) tend to detect a higher proportion of male carriers.<sup>[15-16]</sup>

Couple-level analysis found that 25.9% of couples had at least one thalassemia carrier, and 4.0% were high-risk couples, meaning both partners carried the trait. This figure is directly relevant to genetic counseling since high-risk couples face a significant risk (about 25% every pregnancy) of having children with transfusion-dependent thalassemia major. Recent Iraqi study indicates that structured premarital screening and counseling programs, when implemented, have been critical in lowering impacted births, notably in the Kurdistan Region, where premarital programs developed after 2008 have demonstrated declines in affected births over time.<sup>[17]</sup>

Consanguinity continues to be a key modulator of inherited disease risk in this region. It was estimated consanguinity was higher among high-risk carrier couples, confirming the previously established link between cousin marriage and an increased possibility of sharing pathogenic alleles. This pattern is confirmed by regional epidemiological surveys, which show that consanguineous marriage is frequent in Iraq and associated with higher rates of hemoglobinopathies.<sup>[18]</sup>

Hemoglobin levels were lower in carriers than in non-carriers, which is consistent with established trait hematology and supports evidence from recent study conducted in Jordan demonstrating that thalassemia trait contributes to microcytic anemia patterns even in asymptomatic carriers.<sup>[19]</sup>

The main limitation of this study, is that it was restricted to people with microcytic anemia, which improves detection but may overstate prevalence in comparison to larger premarital populations. Furthermore, consanguinity was estimated rather than verified. Future studies should incorporate direct molecular diagnostics and family history data to improve risk stratification and validate particular mutation prevalence in Iraqi communities.

#### 5. CONCLUSIONS AND RECOMMENDATIONS

Taken together, these findings support the continued use of specific screening approaches (such as microcytosis-based selection) in conjunction with traditional premarital screening. The relatively high carrier prevalence shown here highlights the importance of

improved hematological screening for identifying silent carriers and expanding genetic counseling reach.

#### Conflict of interest

The authors of this study report no conflicts of interest.

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