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PROFESSIONAL PAPER

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# The Incidence of Minor $\beta$ -thalassemia Among Individuals Participated in Premarital Screening Program in Ardabil Province: North-west of Iran

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## ABSTRACT

**Introduction:** The most effective way to prevent thalassemia is to screen for the disease at the population level and then to evaluate the molecularly of individuals. Considering the importance of minor  $\beta$ -thalassemia and its high prevalence in societies. **Aim:** to investigate the incidence of minor  $\beta$ -thalassemia among Individuals participated in Premarital Screening Program in Ardabil province: North-west of Iran. **Methods:** This descriptive cross-sectional study was conducted on 39620 individuals participated in the national screening program for Minor  $\beta$ -Thalassemia. Of them 1925 cases had mean corpuscular volume (MCV) < 80 in one person or couple which referred to HbA2 concentration check. **Results:** Of all 1925 cases, 95 cases (4.93%) had HbA2>3.5 and defined as Minor  $\beta$ -Thalassemia. The total incidence of minor  $\beta$ -thalassemia among all participated people was 2.4%. Of thalassemia cases 48.4% were women and 51.6% were men. Of all  $\beta$ -thalassemia cases, 49.5% live in Ardabil city and rest of them live in other cities. **Conclusion:** Results showed that the prevalence of minor  $\beta$ -thalassemia in Ardabil province was lower than country average rate and a study should be done in future for exact estimation of the disease.

**Keywords:** Minor  $\beta$ -Thalassemia, Screening, MCH, MCV, Ardabil.

## 1. INTRODUCTION

Thalassemia is one of the most common genetic diseases in Iran and due to it, the patient's blood loses a large number of red blood cells and unable to carry oxygen. Hemoglobin consists of two different chains called alpha and beta.

If the body doesn't have the ability to produce enough protein of any kind, the blood cells are not completely formed and don't have the ability to transfer sufficient oxygen and the result is an anemia that begins in childhood and lasts up to the end of life.

Thalassemia is found in people of Mediterranean such as Greece and Italy, Iran, Africa, South Asia and South China (1-3). Disruption of the alpha chain genes causes alpha thalassemia. The other chain of hemoglobin is the beta chain and disruption of the beta gene causes major beta-thalassemia and because there are only two beta genes in comparison to the alpha gene, the probability of defective both versions is high when the parents are carriers of thalassemia.

For this reason the prevalence of beta thalassemia is much higher than that of alpha (4-6). In beta-thalassemia if a gene is defective it is known as minor thalassemia and there is no particular problem in these people. If both beta genes are defective, they cause a dangerous thalassemia type called major thalassemia. Major thalassemia occurs only when the parents of the affected person have both minor thalassemia (5-8).

There are three types of beta thalassemia depending on their effects they are classified in the body from mild to severe. a) Minor Thalassemia: in this case protein deficiency is not sufficient to disrupt the hemoglobin function. A person with this disease carries a genetic property of thalassemia. This person will not experience a problem other than a slight anemia in some cases. As with mild alpha thalassemia, physicians treat red blood cells of a person with

Residence place	Number of Referred Individuals	Incidence (of total, %)	HbA2 measured (n)	Number of thalassemia cases	Prevalence rate (based HbA2, %)
Ardabil city	2886	1.63	1402	47	3.35
Other cities	1076	4.46	523	48	9.2
Total	3962	2.4	1925	95	4.94

Table 1. The incidence rate of minor  $\beta$ -thalassemia in Ardabil province

beta minor thalassemia as a sign of iron deficiency anemia with inappropriate iron supplementation. b) Intermediate thalassemia, In this case the deficiency of beta protein in the hemoglobin is so high that it leads to relatively severe anemia and significant impairment in the health of the individual such as bone malformations and enlargement of the spleen. There is a wide range of symptoms at this stage (3-5). A slight difference between the symptoms of intermediate thalassemia and the severe form (major thalassemia) or large thalassemia may be confusing. Because of the patient's dependence on blood transfusion, the person is placed in the major thalassemia group. Patients with intermediate thalassemia require a blood transfusion to improve their quality of life and not to save. c) Major thalassemia or major thalassemia anemia (Cooley's Anemia), this stage is the most severe form of beta thalassemia, which results in a severe deficiency of beta protein in hemoglobin, which leads to an anemia that is life-threatening and needs a lot of regular blood transfusions and medical care. Repeated blood transfusions in life duration lead to excessive iron agglomeration that they should be treated with prescribing chelator agents to help prevent death and organ failure. Thalassemia is detected by examining the level of hemoglobin in humans. The number of red blood cells and hemoglobin in people with thalassemia is lower than normal rate (12-14).

For the primary diagnosis of beta and alpha thalassemia and iron deficiency anemia electrophores hemoglobin and CBC blood tests are used. In minor  $\beta$ -thalassemia, the MCV below 80, MCH below 27 and HbA2 greater than 3.5. If a person had all of the above conditions, would be considered a definite carriers of  $\beta$ -thalassemia and if intend to marry a person with  $\beta$ -thalassemia, he must has a genetic test. In alpha thalassemia, the indexes will be similar to beta only with the difference that the HbA2 level below 3.5. In iron deficiency anemia the indices will be similar to alpha minor thalassemia with this different that the total amount of hemoglobin displayed with the Hb index is lower than 10. Also in these people, the amount of ferritin is at minimum rate (15-18). There are over 200 different mutations for  $\beta$ -thalassemia and the result is a reduction or loss of globin, although rare. About 20 common alleles make up 80% of known thalaseemics around the world, 3% of the world's population carry genes associated with  $\beta$ -thalassemia. In Southeast Asia, 5-10% of people carry  $\alpha$ -thalassemia genes. In the United States, nearly 2,000 people have  $\beta$ -thalassemia. Currently the most effective way to prevent thalassemia is to screening disease at the population level and then to evaluate the molecularity of the individual which in screening program known as thalassemia carriers. The thalassemia prevention program in

Iran is based upon premarital screening of  $\beta$ -thalassemic couples (carriers) in order to encourage them to participate in counseling and perform prenatal diagnosis (PND) (19).

## 2. AIM

Therefore, the aim of this study was to identify the incidence of minor  $\beta$ -thalassemia among Individuals participated in Premarital Screening Program in Ardabil province: North-west of Iran.

## 3. METHODS

### Study design

In this cross-sectional study, 3962 individuals participated in the national screening program for minor  $\beta$ -thalassemia and of them, 1925 cases (48.6%) had MCV<80. All of cases with MCV<80 were checked for HbA2 concentrations. A checklist including MCV, MCH and HbA2 indices and residence of place, sex was completed for all patients and the prevalence of  $\beta$ -thalassemia was estimated based on the results of the HbA2 concentration.

### Statistical Analysis

Collected data were analyzed using statistical methods in SPSS version 19. This research was approved by ethical committee of Ardabil university of medical sciences with the code IR.ARUMS.REC.1396.236.

## 4. RESULTS

In 1925 individuals with MCV<80 the, HbA2 concentrations were measured that of them 95 case (4.94%) had minor  $\beta$ -thalassemia. Of all cases with minor  $\beta$ -thalassemia, 48.4% were women and 51.6% were men. Also, the total incidence of minor  $\beta$ -thalassemia in all participated was 2.4%.

The average HbA2 was  $4.6 \pm 0.4$  in men and  $4.2 \pm 0.5$  in women and the difference was significant.

## 5. DISCUSSION

In the present study, the total incidence of minor  $\beta$ -thalassemia among all participated cases was 2.4% in Ardabil province that this rate in Ardabil city was 1.63% and in other cities was 4.46%. The mean of HbA2 was  $4.6 \pm 0.4$  in men and  $4.2 \pm 0.5$  in women and the difference was significant. ( $p=0.001$ ) In the study of Afrouz et al, the prevalence of alpha-thalassemia with 78.4%, non-alpha beta with 9% and beta-thalassemia with 8.1% had the highest rate orderly. The average of MCV was significantly higher in patients with alpha-thalassemia than patients with  $\beta$ -thalassemia. There was a significant correlation between MCV and MCH among all patients ( $r=0.83$ ,  $p=0.001$ ) (20). In the study of Hayatbakhsh et al in Kerman, 5.7% of individuals had minor beta-thalassemia that of them 51.6% were men and rest of them were women and the difference wasn't significant and

this rate was upper than our study results (21). In the study of Chehkandi et al, of all individuals, 7.1% had microcytosis that after receiving iron as orally, 41.2% of them weren't corrected which 35.7% had HbA2>3.7% and called carriers of  $\beta$ -thalassemia (22).

In the study of Madmouli et al in 2017, 13.2% of individuals had intermediate and 86.8% major thalassemia . There was a significant relationship between thalassemia type and diagnosis age of thalassemia ( $P = 0.001$ ) . There was a significant positive correlation between age and distance between transfusions (23).

In a study by Nabipoor et al, the overall incidence of  $\beta$ -thalassemia was 2.6% which was similar to our study results (2.4%) (24).

Some studies in other places of country are similar to results of the present study (25-27) but in some studies the prevalence of minor  $\beta$ -thalassemia was greater than our study results (20, 28). Also in the study of Rahmani et al, the prevalence of minor  $\beta$ -thalassemia was significantly lower than our study rate (29).

## 6. CONCLUSION

Results showed that the total incidence of minor  $\beta$ -thalassemia in the studied years based HbA2 concentration in Ardabil province was 2.4%, in Ardabil city was 1.63% and other cities was 4.46% which was lower than other studies within country. It is suggested that a study be conducted on a larger dimension among school age students in the future.

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