

## Prevalence of $\beta$ -thalassemia trait in premarital screening in Al-Hassa, Saudi Arabia

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**BACKGROUND:** The Al-Hassa area is one of the regions in Saudi Arabia where hemoglobinopathies are prevalent. The Saudi Ministry Of Health designed a protocol for premarital testing after the royal decree in December 2003. The protocol was implemented in a February 2004 order. The aim of this study was to determine the prevalence of  $\beta$ -thalassemia trait among subjects coming for premarital screening in the Al-Hassa area.

**SUBJECTS AND METHODS:** From February 2004 to November 2004, healthy subjects coming to six marriages consultation centers in the Al-Hassa area underwent routine mandatory tests. Subjects were considered to have  $\beta$ -thalassemia trait if they had a mean corpuscular volume (MCV)  $<80$  fL and/or a mean corpuscular hemoglobin (MCH)  $<27$  pg and a hemoglobin A2 level  $>3.2\%$ . Venous blood was taken into an EDTA tube and the complete blood count and red blood cell indices were measured by a Coulter automated cell counter on the same day of hemoglobin collection. Electrophoresis was done on cellulose acetate.

**RESULTS:** All Saudi participants (n=8918), including 4218 (47.3%) males and 4700 (52.7%) females were screened. The prevalence of  $\beta$ -thalassemia trait with high hemoglobin A2 and microcytic hypochromic anemia was 3.4% (307/8918).

**CONCLUSION:** In countries with a high prevalence of hemoglobinopathies, a premarital screening program is helpful for identification and prevention of high-risk marriages. With a 3.4% prevalence of  $\beta$ -thalassemia trait in premarital couples, future comprehensive programs are needed to know the actual prevalence of  $\beta$ -thalassemia in Al-Hassa.

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$\beta$ -thalassemia, one of the thalassemia syndromes, is an autosomally recessive inherited blood disease characterized by a reduction in the synthesis of the  $\beta$ -globin chain, which leads to a reduction in  $\beta$ -globin chain synthesis and eventually produces hypochromic microcytic anemia.<sup>1,2,3</sup>  $\beta$ -thalassemia is widespread throughout the Mediterranean region, Africa, the Middle East, the Indian subcontinent and Southeast Asia.<sup>3,4,5</sup> Due to a high prevalence of inherited blood disorders in Saudi Arabia (sickle cell disease and thalassemia), the Saudi Ministry Of Health designed a protocol for premarriage testing after a royal decree in December 2003, which was implemented by a February 2004 order. The aim of the study was to determine the prevalence of  $\beta$ -thalassemia trait among subjects coming for premarital screening in the Al-Hassa area.

## Subjects and methods

From February 2004 to November 2004, healthy subjects attending six marriages consultation centers in the Al-Hassa area underwent routine mandatory tests. The mandatory tests included complete blood count (CBC), sickle cell test, and hemoglobin electrophoresis (even when subjects had normal indices) for both partners. The data in the premarital form included name, age, sex, national number, address, and telephone number. Subjects were considered to have β-thalassemia trait if they had MCV <80 fL and/or MCH <27 pg and a hemoglobin A2 level >3.2%. Venous blood was taken into an EDTA tube, the CBC and red blood cell indices were measured by a Coulter automated cell counter on the same day of collection. Hemoglobin electrophoresis was done on cellulose acetate (Helena Biosciences, Texas, USA, kit SAS 3).

## Results

We screened all Saudi participants (n=8918), including 4218 males (47.3%) and 4700 females (52.7%). The prevalence of β-thalassemia trait with high Hb-A2 and microcytic hypochromic anemia was 3.4% (307/8918) overall and included 164 (53.4%) males and 143 (46.6%) females.

## Discussion

As in other Mediterranean and Middle Eastern countries, β-thalassemia is an important health problem in the Kingdom of Saudi Arabia. This study determined the prevalence of β-thalassemia among premarital couples that came to a consultation centers for the purpose of preventing unsafe marriages that would lead to the possibility of birth children with thalassemia major. β-thalassemia major causes serious physical and emotional problems for patients and families, and a financial burden for health services.<sup>6</sup> Premarital screening is very useful for detecting carriers of β-thalassemia and for controlling thalassemia major.

To our knowledge there is no national study showing the actual prevalence of β-thalassemia trait in Saudi Arabia, but the prevalence of the β-thalassemia gene is estimated to range between 0.01 to 0.15 in various areas of Saudi Arabia.<sup>6,7,8,9</sup> The molecular pathogenesis of both the Mediterranean and

**Table 1.** Hematological values in subjects with β-thalassemia trait.

	Mean±SD	Range
Age (years)	24.8±6.7	14-47
Red blood cells (× 10 <sup>6</sup> /μL)	6.53±0.75	4.1-7.91
Hemoglobin (g/dL)	12±1.6	9.2-15.1
Mean corpuscular volume (fL)	61±6.8	50.4-79.4
Mean corpuscular hemoglobin (pg)	20.27±2.4	16.8-26.9
Hb-A2 (electrophoresis) (%)	5.3±1.2	3.2-6.06
Hb-F (%)	0	0 (majority)-2.35

Asian β-thalassemia mutations overlap in Saudis, with the most frequent mutations for β-thalassemia being IVS-110, IVSII-1, CD39, IV11-3 and CD6.7 This overlapping was confirmed in another study done in the Qatif area.<sup>10</sup> Several reports from Arab countries indicate that β-thalassemia carriers have common genetic abnormalities, and the frequency of this disorder varies from country to country in the Middle East and is reported to be between 1% to 15%.<sup>6,11</sup> The prevalence of 3.4% found in our study is in keeping with that reported in Arab countries. High hemoglobin A2 with an absence of hemoglobin F (in most patients) is the most commonly encountered type of β-thalassemia trait in Saudi Arabia, and is typically characterized by the microcytic and hypochromic red blood cells found in this study. Although β-thalassemia is an autosomally recessive disorder, we found a nonsignificant gender difference between males (n=164, 53.4% vs n=143, 46.6%) and females.

In countries with a high prevalence of hemoglobinopathies, premarital screening is helpful for identification and prevention of high-risk marriages. With a 3.4% prevalence of β-thalassemia trait, future comprehensive programs are needed to know the actual prevalence of β-thalassemia in Al-Hassa.

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