



Evaluation of hemoglobinopathy screening results of a six year period in Turkey

Seçil Gunher Arıca, Ebru Turhan, Cahit Özer, Vefik Arıca, Dilek Benk Şilfeler, İbrahim Şilfeler, Ayşe Betül Altun

International Journal of Collaborative Research on Internal Medicine & Public Health
Vol. 4 No. 2 (February 2012)

International Journal of Collaborative Research on Internal Medicine & Public Health (IJCRIMPH)

ISSN 1840-4529 | Journal Type: Open Access | Volume 4 Number 2

Journal details including published articles and guidelines for authors can be found at:

<http://www.iomcworld.com/ijcrimph/>

To cite this Article: Arıca SG, Turhan E, Özer C, Arıca V, Şilfeler DB, Şilfeler I, Altun AB. Evaluation of hemoglobinopathy screening results of a six year period in Turkey. *International Journal of Collaborative Research on Internal Medicine & Public Health*. 2012; 4(2):145-151.

Article URL: <http://iomcworld.com/ijcrimph/ijcrimph-v04-n02-07.htm>

Correspondence concerning this article should be addressed to Assistant Professor Dr. Vefik ARICA; Medical Faculty of the Mustafa Kemal University, 31100, Serinyol, Antakya, Hatay, Turkey | Email: vefikarica@hotmail.com | Phone: +90 326 2291000 Fax: +90 326 2455654

Paper publication: 18 February 2012

International Journal of Collaborative Research on Internal Medicine & Public Health

Editors-in-Chief:

Asst. Prof. Dr. Jaspreet S. Brar (University of Pittsburgh, USA)
Forouzan Bayat Nejad

Executive Editor: Mostafa Nejati

Deputy Editor: Dr. Mensura Kudumovic (University of Sarajevo, Bosnia & Herzegovina)

Evaluation of hemoglobinopathy screening results of a six year period in Turkey

Seçil Gunher Arıca (1), Ebru Turhan (2), Cahit Özer (1), Vefik Arıca (3) *, Dilek Benk Şilfeler (4), İbrahim Şilfeler (5), Ayşe Betül Altun (6)

1) Mustafa Kemal University Medical Faculty, Department of Family Medicine, Hatay, Turkey

2) Mustafa Kemal University Medical Faculty, Department of Public Health, Hatay, Turkey

3) Mustafa Kemal University Medical Faculty, Department of Pediatric, Hatay, Turkey

4) Mustafa Kemal University Medical Faculty, Department of Obstetrics and Gynecology, Hatay, Turkey

5) Hatay Hassa State Hospital, Department of Pediatric, Hatay, Turkey

6) Hatay Health Directorate, Child Health and Family Planning Directorate, Hatay, Turkey

* Corresponding Author

ABSTRACT

Background and Objectives: Hemoglobinopathies are autosomal recessive inherited diseases more commonly seen in Mediterranean countries. Hereditary blood diseases including B-thalassemia and sickle cell anemia are important health problems. In our study we aimed to analyze the results of the premarital hemoglobinopathy screening test for a 6 years period in Hatay region.

Material and Methods: The study sample comprised the couples attending to the Mother and Child Health Care Center in Hatay for premarital hemoglobinopathy screening from 2004 to 2009. Hemoglobin chain analyses of 87.830 couples were evaluated.

Results: 175.660 people were screened at total. The prevalence of beta thalassemia trait, sickle cell anemia trait, sickle cell anemia, beta thalassemia major, beta-thalassemia intermedia, alpha-thalassemia, alpha-thalassemia trait was found as 13.921 (7,9%), 6.074 (3,4%), 631 (0.35%), 132 (0.07%), 118 (0.06%), 9 (0.005%), 150 (0.08%) respectively. 72 newborns with beta-thalassemia were diagnosed as a result of the marriage of the carrier couples in 6 years.

Conclusions: Hatay is a high risk region for beta-thalassemia and sickle cell anemia trait. In countries with high prevalence of hemoglobinopathies, a premarital screening program and counseling is needed to decrease the prevalence.

Keywords: Hemoglobinopathy, Thalassemia, sickle cell anemia

Introduction

Hemoglobinopathies are amongst the most prevalent of all human genetic disorders resulting from structural changes or abnormal synthesis of polypeptide chain of the hemoglobin molecule (1). Although it is highly prevalent in tropical and subtropical regions, became a common problem all around the world because of the migrations (2-4). Hemoglobinopathy is amongst the most common hereditary hematologic disorder in country and especially in Cukurova region an important health problem. In Turkey, there are approximately 1.300.000 carriers and 4000 patients (5, 6). According to the results of the survey carried out by Ministry of Health and National Hemoglobinopathy Council, mean beta-thalassemia trait prevalence of the sixteen mostly common centers in Mediterranean region was 4.3% (4-7). Prevention of hemoglobinopathies that are important causes of mortality and morbidity with premarital screening and prenatal diagnose is possible. Thus determinations of carriage and premarital screening tests in highly prevalent regions have great importance (8). For this reason a first step Hemoglobinopathy Control Program was established in 33 provinces including Hatay regarding the disease prevalence rates in 2002 (4).

The aim of this study is to determine the prevalence of hemoglobinopathy trait evaluating premarital screening results of Hatay region.

Material and Methods

The study sample comprised the couples attending to the Mother and Child Health Care Center in Hatay for premarital hemoglobinopathy screening from 2004 to 2009. Hemoglobin chain analyses of 87.830 couples were evaluated. The blood samples

were collected at the main Center in Antakya. The blood samples at the districts are sent to the center with a carrier by obeying the cold transport and their evaluated results at the center again transported to the districts by a carrier. The required information about the subject is given during the application and the results are delivered in accordance with the privacy rules. All of the applicants were subjected to the complete blood count and hemoglobin electrophoresis. Hematological parameters were analyzed by Coulter counter and hemoglobin electrophoresis has been performed by MALTA Celloclear Alkaline Electrophoresis. Hemoglobin A₂ was analyzed by micro column technique from Bio Systems kits and hemoglobin F was assayed by alkaline denaturation technical. The blood samples with mean corpuscular volume (MCV) levels below 80 fL were analyzed for hemoglobin A₂ (HbA₂) and hemoglobin F (HbF). People who had elevated HbA₂ ≥ 3.5 and/or HbF ≥ 2.0 were diagnosed as β thalassemia trait. Sick cell Hemoglobin (HbAS) indicates as sick cell trait and Sick cell disease (HbSS) indicates as sick cell anemia with hemoglobin electrophoresis. Because of the high costs, gene analyses were done only cases those with normal HbA₂ levels but MCV < 80 fl or HbF > 2%

Ethical consent was obtained (Mustafa Kemal University Medical Faculty Ethics Committee (02.10.2010/153).

Results

175.660 people (87.830 male, 87.830 female) were screened as a part of premarital screening program in Hatay region in six years. Beta-thalassemia trait was found in 13.921 people (7.9%). Total sickle cell anemia trait was found in 6.074 people (3.4%). 3208 (3.6%) of them were male, while 2.866 were female (3.3%) (**Table I**). The prevalence of

sickle cell anemia, beta-thalassemia major, beta thalassemia, alpha thalassemia, alpha thalassemia trait was found 0.35% (n=631), 0.07% (n=132), 0.06 % (n=118), 0.005% (n=9), 0.08% (n=150), respectively.

Beta-thalassemia trait prevalence in districts of Hatay was found as; Antakya 4.409 (2,5%), Reyhanlı 1.450 (0,8%), Altınözü 912 (0,5%), İskenderun 1.160 (0,7%), Belen 7 (0.003%), Erzin 97 (0.05%), Samandağ 1.369 (0,7%), Yayladağ 46 (0.03%), Kırıkhan 1.360 (0,7%), Dörtöyl 976 (0,6%), Hassa 681 (0,4%), others 1.453 (0,8%).

Sickle cell anemia trait in districts was found (**Table II, Figure I**). The age intervals of the subjects were calculated as follows: age: 17-20, 14.322 (8.2%), age 21-29, 126.117 (71.8%), age 30-40, 20.074 (11.4%), age over 40, 15.147 (8.6%). Premarital hemoglobinopathy screening was carried out in 27.612 people in 2004, in 28.261 in 2005, in 28.839 in 2006, in 30.733 in 2007, in 32.016 in 2008, in 28.199 in 2009. Formal marriage numbers according to years in Hatay region was as follows: in 2004 27.411, in 2005 26.852, in 2006 28.296, in 2007 30.094, in 2008 28.730, in 2009 27.696. In the study period 182, 291, 417, 502, 598, 644 couples were found to be carriers, respectively (**Table III**). 72 newborns with beta-thalassemia were diagnosed as a result of the marriage of the carrier couples in 6 years.

Discussion

Thalassemia was first described in a child with deep anemia, splenomegamia and characteristic bone changes in 1925 by Thomas Cooley and Peael Lee (9). The name was originated from the Greek word "thalas" which means Mediterranean because formerly it was thought to be prevalent in the Mediterranean region. Hemoglobinopathies

can be classified into thalassemias and abnormal hemoglobinopathies. The latter is consisting of aminoacid change on the globin chain of hemoglobin molecule. The most prevalent abnormal hemoglobins are Hb S, Hb D, Hb E, and Hb C (1). Thalassemia is a genetic disorder characterized with a decrease or a complete loss of synthesis of the globin chains of hemoglobin (10). The most prevalent types are α ve β -thalassemia. β -thalassemia is common in Turkey (11). Abnormal hemoglobins are generally specific to regions and ethnicity. In Turkey, HbS is the most prevalent abnormal hemoglobin type which causes to sickle cell anemia (11). Thalassemia syndromes are the most common monogenetic disorders in human which were described in detail. In the light of the knowledge, not only managing the broad clinical spectrum but also precise decisions using prenatal diagnosis would be possible (12).

The effective methods to prevent autosomal recessive hereditary diseases such as hemoglobinopathies are community education about the disease, determining the carriers by screening, genetic counseling, and prenatal diagnosis (4, 13).

Hemoglobinopathies were controlled; number of new cases was decreased, even eradicated in some countries including Cyprus, Greece and Italy (4, 8). Previous studies showed the efficacy, efficiency and, importance of premarital screening in high risk populations (14).

Beta-thalassemia trait prevalence of Turkey and other Middle Eastern countries were compared and showed in **figure II**. We can see the high prevalence in Turkey (15). The prevalence of beta-thalassemia trait and sickle cell trait is shown in **figure III**. They're prevalent in Mediterranean region. Beta-thalassemia prevalence was found 7.7 % while it was 4.3% in Turkey. Sickle cell anemia trait

prevalence was found 10.0% in Adana and 13.6% in Mersin-the cities in Mediterranean region-(4-7). In the present study sickle cell anemia trait prevalence was found 3.4% which is lower than Mediterranean region but higher than Turkey average.

Thalassemia and hemoglobin disorders are one of the common hereditary disorders in our country and all over the world. Although these hereditary disorders are more common in Mediterranean region, studies showed the presence at various rates all around the world. Today the only way for prevention is prenatal diagnosis, therefore hemoglobinopathy control programs were established not only in our country but also all over the world. The main approach of these control programs were identification of the individuals especially in the premarital age.

Antenatal screening programs is common in the Mediterranean populations. The successes of these programs depend on public health education and effective screening programs. Application of these programs in crowded Asian populations like India and southeast countries might be difficult because of faith, culture, and economical factors. Initially education programs should be established and voluntary screening programs should be supported. Following these programs would compose the basic approach on establishment of the development of the national population control programs (9). Public education and acknowledgement studies should be given priority in countries which are composed of different religious, ethnic and cultural diversity like ours.

As a conclusion Hatay is a high risk region for beta-thalassemia and sickle cell anemia. A premarital screening program is of great value to prevent high risk marriages to decrease prevalence of hemoglobinopathies in high risk regions. Also counseling about the antenatal screening and hereditary diseases should be

given to disease carrying couples and to the whole population.

Conflict of Interest: None to declare

References

- 1- <http://library.cu.edu.tr/tezler/6490.pdf> last access: 20.11.2010
- 2- Yapıcı G, Aksoz F, Kızılok A. Frequency of Carriers for Beta Thalassemia and Sickle Cell Trait in Mersin. The Anatolian Journal of Clinical Investigation (AJCI) 2007; 1(4): 255-259.
- 3- Guidelines for the control of hemoglobin disorders. WHO/HDP/HB/GL/94, 1. Control of Hereditary Diseases 1996; Genova WHO.
- 4- http://rnaportal.com/RNAFTP/Upload/Guncel_Cocuk_Sagligi.pdf last access: 20.11.2010
- 5- Canatan D, Kose MR, Ustundag M, Haznedaroglu D, Ozbas S. Hemoglobinopathy control program in Turkey. Community Genet 2006; 9: 124-6.
- 6- Gulleroğlu K, Sarper N, Gokalp A. Public education for the prevention of hemoglobinopathies: a study targeting Kocaeli University students. Turk J Hematol 2007; 24:164-170.
- 7- Guler E, Garipardic M, Dalkiran T, Davutoglu M. Premarital screening test results for β -thalassemia and sickle cell anemia trait in east mediterranean region of Turkey. Pediatric Hematology and Oncology 2010; 1(6): 1-5.

- 8- Guler E, Davutoglu M, Karabiber H, Celik M, Ekerbicer H, Paksoy MF. Evaluation of the Results of Premarital Screening of Hemoglobinopathies Trait in Kahramanmaras. TAF Prev Med Bull 2008; 7(3): 243-244
- 9- Weatheral DJ. The Thalassemias. In: Stamatoyannopoulos G, Majerus PW, Perlmutter RM, Varmus H. Eds. The Molecular Basis of Blood Disease, 3th Ed., Philadelphia: WB Saunders Comp, 2001: 183-226.
- 10- http://www.talasemi.org/pdf/tani/cansinT_edavi-9pdf.pdf last access: 20.11.2010
- 11- Basak N. Molecular Genetics of Thalassemia. http://www.thd.org.tr/doc/kurs_pdf/nazlib_asak.pdf last access: 18.11.2010
- 12- Thein SL. Genetic modifiers of the b-hemoglobinopathies. Br J Haematol 2008; 141: 357-366.
- 13- Screening programs and methods. http://www.talasemi.org/pdf/tani/cansinT_edavi-5.pdf. Last access: 20.11.2010.
- 14- Al-Suliman A. Prevalence of beta-thalassemia trait in premarital screening in Al-Hassa, Saudi Arabia. Ann Saudi Med 2006; 26: 14-6.
- Thalassemia and abnormal hemoglobins in the world and in Turkey. <http://www.talasemi.org/pdf/tani/1cansinTani.pdf>. Last access: 20.11.2010.

Table I: Prevalence of disease carriage in both sexes

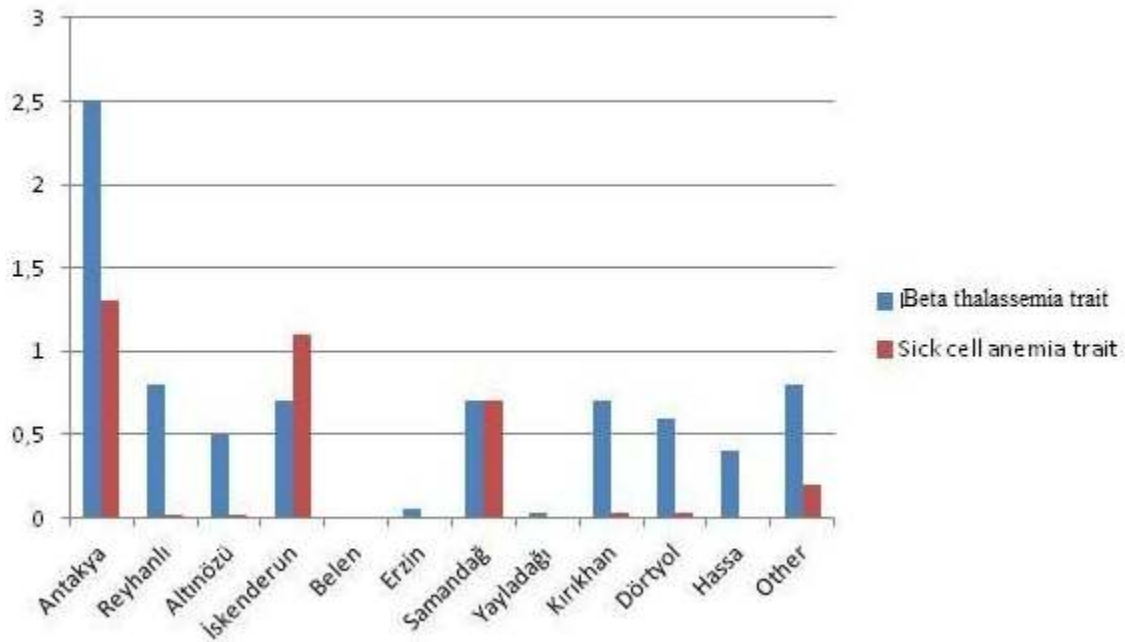
	Male		Female		Total	
	n	%	n	%	n	%
Beta Thalassemia trait	5709	6.5	8212	9.3	13921	7.9
Sickle cell anemia trait	3208	3.6	2866	3.3	6074	3.4
Total	87830	100	87830	100	175660	100

Table II: Screening results according to birth place

	Beta Thalassemia trait (n)	%	Sickle cell anemia trait (n)	%
Antakya	4409	2.5	2356	1.3
Reyhanlı	1450	0.8	37	0.02
Altınözü	912	0.5	42	0.02
İskenderun	1160	0.7	1898	1.1
Belen	7	0.003	2	0.001
Erzin	97	0.05	2	0.001
Samandağ	1369	0.7	1224	0.7
Yayladağ	46	0.03	4	0.002
Kırıkhan	1360	0.7	45	0.03
Dört Yol	976	0.6	55	0.03
Hassa	681	0.4	5	0.003
Other	1454	0.8	404	0.2
Total	13921		6074	

Table III: Premarital screening number according to years, number of married people and number of carrier couples

	2004	2005	2006	2007	2008	2009
Premarital screening number	27.612	28.261	28.839	30.733	32.016	28.199
Number of married people	27.411	26.852	28.296	30.094	28.730	27.696
Number of carrier couples	182	291	417	502	598	644

**Figure I:** Screening results according to birth place

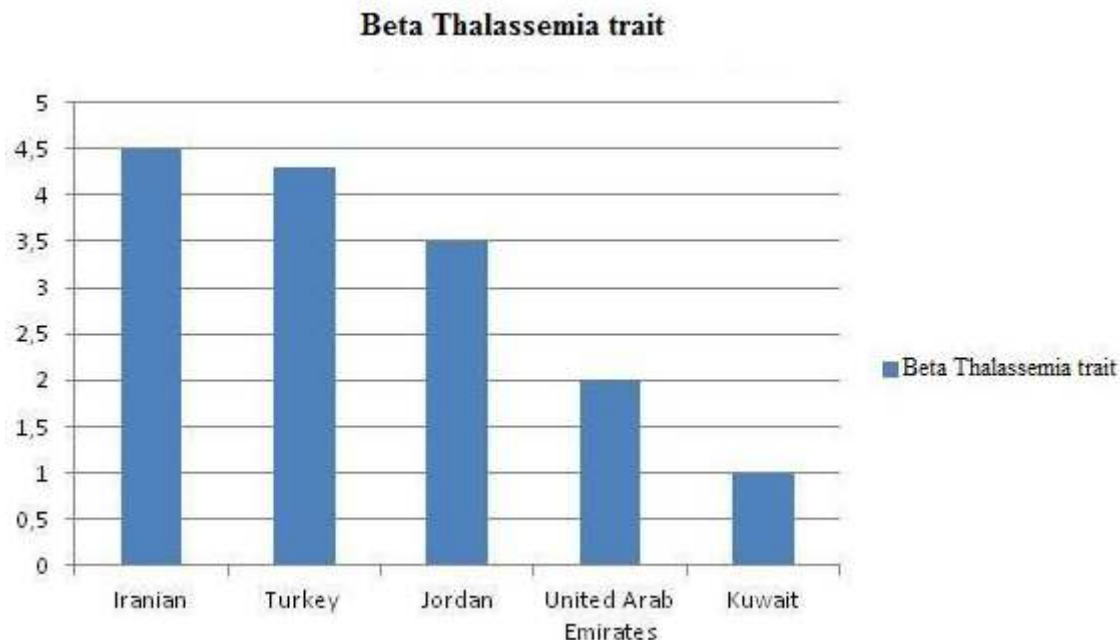


Figure II: Comparison of beta-Thalassemia trait prevalence of Turkey with Middle Eastern Countries

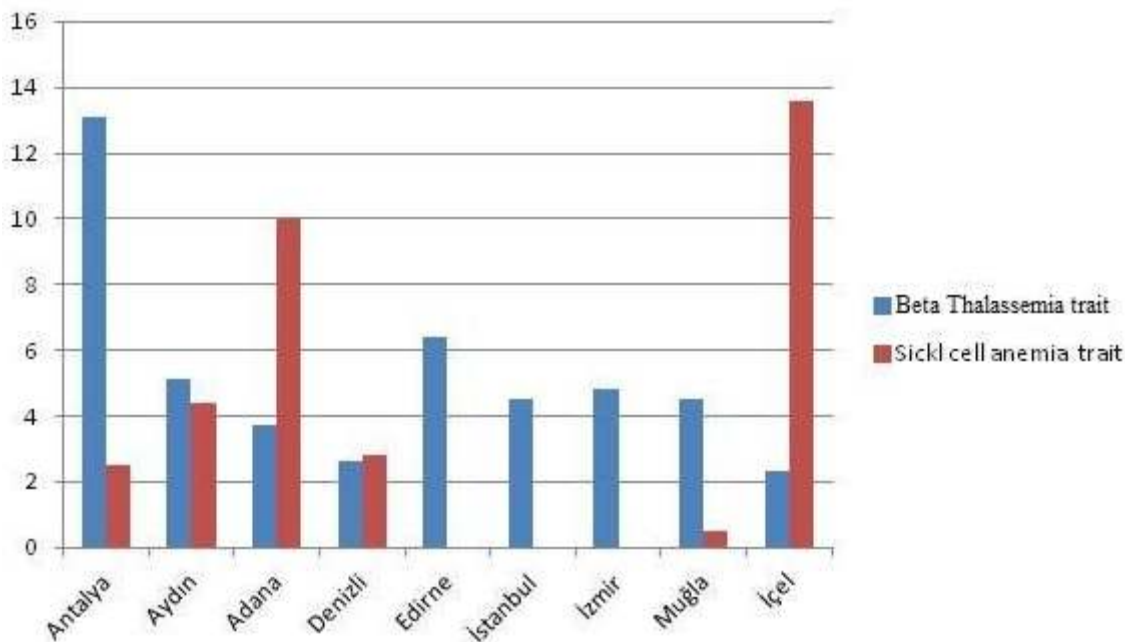


Figure III: Prevalence of beta-Thalassemia trait and Sickle cell trait in Turkey