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Letter to the Editor

The Frequency of Beta-Thalassemia Mutations among Carriers in Dezful City, Southwest Iran

***Parvin JOOLA** [1], ***Behnaz ANDASHTI** [2], **Seyed Ahmad HOSSEINI** [3], **Seyedeh Maryam MOLAEI ZADEH** [4], **Nosrat BAHRAMI** [5]*

1. *Department of Non-Communicable Diseases, Deputy of Health Services, Dezful University of Medical Sciences, Dezful, Iran*
2. *Department of Genetic, Faculty of Science, Shahid Chamran University of Ahvaz, Ahvaz, Iran*
3. *Nutrition and Metabolic Diseases Research Center, Ahvaz Jundishapur University of Medical Sciences, Ahvaz, Iran*
4. *Deputy of Health Services, Dezful University of Medical Sciences, Dezful, Iran*
5. *Department of Midwifery, Faculty of Nursing and Midwifery, Dezful University of Medical Sciences, Dezful, Iran*

***Corresponding Author:** Email: b_andashti@yahoo.com

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Dear Editor-in-Chief

Beta- thalassemia (BT) is one of the most common monogenic disorders in the world which inherited by an autosomal recessive manner. There are more than 1.5 million carriers of BT in the world and almost 60000 new carriers are born each year (1). BT is characterized by defect in beta globin chain synthesis and imbalance of alpha/beta globin portion causes sediment of extra chains and pathophysiologic changes in patients (2). BT is more prevalent in Mediterranean countries, Middle East, India, North Africa, central and southwest Asia (3). The Allele frequency and mutation dispersion of BT are specific in each population. It often includes 4-5 common mutations that affect 90% of all cases. There are numerous rare mutations in each population (4).

Iran is located on thalassemia belt. In Iran, BT is more prevalent in the border of the Caspian Sea, Persian Gulf, Oman Sea, Khuzestan, Fars, Esfahan and Kerman provinces with a prevalence of 8-10% (1, 4). Distribution of BT mutations is not similar in different regions of Iran and allele frequency is different from north to south and west to east of the country(5, 6).

Iran is a multicultural country. Ethnic diversity is important feature of Iran, which is in 16th place with 24 percent similarity. Iranian sociologists have identified 29 nationalities in this country. Fifty-one percent of Iran population has Fars ethnicity (7). Khuzestan population is a combination of Fars, Arab, Lur and Bakhtyari races. Dezful district, with a several thousand historical documents besides Dez River, has been the source of malaria and thalassemia endemic region from far away(8, 9). Furthermore, Dezful district is a combination of different ethnicities. Therefore, it is predicted to be a wide spectrum of genetic variety in Dezful region.

In our cross sectional study, 555 carrier individuals were enrolled by census sampling method. The study was approved by Research Committee of Dezful University of Medical Sciences and all patients were consenting with the study.

Beta-thalassemia carriers with blood indexes of low MCV (< 80 Fl), low MCH (< 27 pg), and decreased or normal hemoglobin levels, were referred to medical genetic laboratories to detect beta thalassemia mutations. All BT mutations were recorded from 2008 to 2013. Data were analyzed by SPSS16 (Chicago, IL, USA) software using descriptive statistical tests.

Overall 35 BT mutations were found, which were the most common mutations based on prevalence rate, respectively, IVSII-1(G>A) (26.1%), cd36/37(-T) (18.4%), IVSI-110(G>A) (9.9%) and IVSI-5(G>C) (6.8%). There were five different races in this study, including Fars (29.7%), Bakhtiary (30.6%), Lur (23.4%), Arab (9.4%), Kord (3.2%) and other races (2.9%).

IVSII-1(G>A) was the most frequent mutation in Fars and Lur populations, cd36/37 (-T) was the most in Bakhtiary and Arab populations and cd8/9(+G) was the most in Kord population (Table 1). Fars population had the highest mutation diversity (88.5%) followed by Bakhtiary, Lur and Arab population with 66.7%, 55.6% and 30.6%, respectively.

Table 1: Frequency of common mutations in each ethnicity

Race	*Mutation*	*Frequency*	*%*
Fars	IVSII-1(G>A)*	59/165	40
Lur	IVSII-1(G>A)	40/130	27
Bakhtiary	cd36/37(-T) **	46/170	45
Arab	cd36/37(-T)	14/52	13.7
Kord	cd8/9(+G)	7/18	21.8

* IVS: intervening sequence; ** cd: codon

Mediterranean mutations had the highest prevalence rate in our study (49.4%) and Iranian mutations were the second in prevalence (19.1%) (Table 2). The most frequent mutations were IVSII-1(G>A) and cd36/37(-T) respectively which were according to other studies in Khuzestan province. On the other hand, High diversity of mutations in Fars population (85.7%), confirms the ethnic complexity in this region. Research extension in future studies may detect more kinds of β-gene mutations which is helpful in screening and prevention programs in Iran.

Table 2: Frequency of mutations by origin

Mutation source	*Frequency (%)*
Mediterranean	49.4
Iranian	19.1
Indian Asian	16.4
Azarbaiejanian	3.8
Turkish	1.3
Iranian kord	4.7
Arabian	1.8
East Asian	1.0
European	2.35

Conflict of interest

The authors declares that there is no conflict of interest.

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