The Diagnosis and Frequency of Beta and Alpha Thalassemia Mutations and other C, D, and S Common Hemoglobinopathies in Ahvaz Volunteer Patients
Khodamorad Zandian*, Mohammad Pedram*, Bizhan Keykhaei**

* Professor at Children Blood and Cancer Specialty Department, Thalassemia and Hemoglobinopathy Research Center in Ahvaz Jondi Shapoor University of Medical Sciences
* Assistant Professor at Children Blood and Cancer Specialty Department, Thalassemia and Hemoglobinopathy Research Center in Ahvaz Jondi Shapoor University of Medical Sciences

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ABSTRACT

Background and objective: Khuzestan Province have major and various gene reservoir of Beta and Alpha Thalassemia and C, D, and S Hemoglobinopathies and other types of Hemoglobinopathies. The purpose of this study was analyzing the frequency of common mutations among the volunteers for before-birth prevention.

Methodology: Thus, in this prospective study which took 4 years (2001-2005), 93 Ahwazi volunteers were sent to Tehran medical Genetics private centers for conducting necessary experiments for before-birth diagnosis, and diagnosis of mutation in carrier parents and suffering fetuses. The participants covered the expenses themselves. The results obtained from existing mutations were analyzed. The experiments were conducted on a voluntarily basis and no selection was at work. Thus, it can be claimed that the above pattern (due to its relative high costs) was related to those who afforded to pay for the expenses.

Results: There were 11 patient fetuses (4 cases of Beta Thalassemia, 2 cases of Cycle Thalassemia, 2 cases of Cycle Hemoglobin D, 1 case of Cycle Hemoglobin C, and 1 case of Hemoglobin H). The results of frequency of mutation variety in this study showed that Major Beta Thalassemia, Hemoglobin S, Alpha Thalassemia, Hemoglobin D, and Hemoglobin C consisted 55, 16, 9, 3, and 1 percent of this sample respectively. More research is needed to determine the remaining unknown 18%.

Conclusions: The results of the present study showed that a small percentage of pregnant women carry fetuses with Major Thalassemia (it is obvious that the majority of these fetuses, about 75%, do not need therapeutic abortion). Due to the long distance between Tehran and Ahvaz, before-birth Thalassemia preventive experiments should be provided in Ahvaz. In addition, the existence of 18% of unknown genes in carriers of Thalassemia highlights this need more.

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Introduction:
Khuzestan Province, and Ahvaz, which is a populated city, have an ethnically mixed population due to communications with the outside world though water and land, existence of Arab, Bakhtiari and other ethnic groups of people, its geographical location near the Persian Gulf, its hot weather near the equator, and the Malaria epidemics in the past. In addition, the outbreak of various wars in this region has caused Khuzestan to be a major and various gene reservoir of types of Thalassemia and other Hemoglobinopathies (1, 2, and 3). It is obvious that there are various Asian, Indian, Mediterranean, Arab, and Iranian specific genes in it (3, 4, and 5). The diagnosis of common carriers of the region is among the main and fundamental principles of before-birth prevention in hygiene. In regions with high distribution and frequency of Thalassemia, universal screening
requires precise organizational plans with complete technology and enough financial resources which have not been sufficiently provided for Khuzestan Hygiene so far (6). Thus, in the absence of organizations, facilities, and budget, some volunteers were sent to Tehran Genetics private centers for determining the types of Hemoglobin genetic mutations according to their need and necessity during 4 years (2001-2005). They themselves covered the expenses or covered them with the help of existing insurances. Some individuals participated for the first phase of prevention, carrier mothers in suffering families participated for the second phase of prevention and Chrionic Villus Sampling form Beta Thalassemia fetuses and other common Hemoglobinopathies. The data were collected and statistically analyzed. Regarding the 7 to 10 percent frequency of Beta Thalassemia (2) and the 1 to 3 percent frequency of Cycle Tuberculosis carriers (1) in the region, it is suggested to do significant investments for the prevention of Thalassemia and other Hemoglobinopathies.

**Method:**
The method of this experimental study was prospective with clinical trial. The data of the experiments with Ploymerase Chain Reaction/Amplification Refractory Mutation System Method were gathered from valid genetics laboratories of Tehran (2-4). About 186 patient chromosomes were experimented.

**Results:**
The frequency of Thalassemia common mutations and other Hemoglobinopathies of this report are shown in tables 1 and 3. Among the 44 fetuses, 10 fetuses were healthy and 11 were sick (see table 3) and 23 were carriers (see table 4).

**Discussion:**
As shown in table 2, the first common disease is major Beta Thalassemia. The second common disease is Cycle Tuberculosis and the third one is Alpha Thalassemia. Other Hemoglobinopathies such as C and D also exist in the region. It is obvious
that most of the diseases are congenital chronic anemia during life that have severe problematic symptoms for families and the society. Due to the variety in Hemoglobinopathy mutations, their Homozygotes and Heterozygotes were classified based on clinical severity in table 5. In moderate-severe and very severe conditions such as Major Beta Thalassemia before marriage and before pregnancy, PND experiment and abortion with parents’ consent are suggested. Thus, each hygienic, legal, and insurance measure which is taken should be expert, steady, and universal; and include all Cycle Tuberculosis and Hemoglobinopathy diseases 1. Thalassemia, S, D, C, E, etc. Hemoglobinopathies, and types of point disorders of Betaglobin Gene are placed on Chromosome 11 and are transferred from parents to children in autosomal form (6, 7 and 8). Those who are the carriers of a disease are mostly like healthy people most of whom are unaware that they are carriers. Their being carriers can be confirmed through necessary experiments before marriage or pregnancy. Genetic counseling before marriage has become a health civil law in Thalassemia-prone regions of the world (8, 9, and 10). The above-mentioned law is administered by the Hygiene Center in Iran before marriage. The experiment for the diagnosis of the type of Thalassemia or Hemoglobinopathy mutation should be conducted before marriage or pregnancy in carriers or suspicious people to make time for studying the mutation in the suffering fetus during the first three month of pregnancy. Now, these diseases are controlled by hygiene technology in many points of the world (1-2) including Iran in which common mutations are investigated and recorded with the significant growth in technology, and scientific and practical genetics. Effort is continued for studying unknown genes. With the growth of necessary budget indexes in hygiene, access to resolution of general problems will be easier (11, 12, and 13).

Acknowledgements:

1. Thalassemia has long been a known disease in Iran law, but other Hemoglobinopathies such as Cycle Tuberculosis, C, D, E, O, Arab, etc. Hemoglobins have not been attended to, while they are all point mutation disorders of Russian Beta Thalassemia Gene Chromosome 11.

References:


