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An Overview of Hereditary Diseases in Khuzestan Province, Southwest Iran

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The province of Khuzestan, located southwestern of Iran, is one of the country's largest provinces with a population of over four million people and is composed of various ethnic groups such as Arab, Persian, Bakhtiari/ Qashqai. Consanguineous marriage is common in this province and in some ethnic groups, particularly among the Arab population, its rate is much higher than the country's average rate of 38.6% (1). This has led to a high incidence of certain genetic disorders in the province.

According to annual reports compiled of data taken from patients who had referred to the Genetic Counseling Center of Ahvaz Welfare Organization between 2011 and 2013, thalassemia disease, metabolic disorders, deafness and chromosomal abnormalities such as Down syndrome are the most common complications in the studied pedigrees. Based on these reports over 75% of couples had a kinship relation and more than 50% of them were first cousins, and this accounts for some of the aforementioned diseases.

Iran is one of the countries located on the thalassemia belt of the world (2), with the disease being the most common disease of the country, particularly in the Khuzestan province (3). Thalassemia is a congenital hemolytic anemia, which starts with anemia and is associated with growth retardation and bone deformities. According to previous official reports by the Health Ministry and Iran Blood Transfusion Organization, the number of patients with major thalassemia in Iran is about 18,500 people (4), of which 1800 patients (about 10%) are from the Khuzestan province, making the region second in regards to the number of thalassemic cases.

Furthermore, α-thalassemia, β-thalassemia, Hb S, Hb C, Hb D, Hb E are the most common hemoglobin disorders, of which α-thalassemia is the most frequent in this province. On the other hand, the prevalence of β-thalassemia gene (the second most common hemoglobin disorder in the province) is between 5 to 7%. In the study carried out by Latifi and Zandian (2010) on 109 patients with major thalassemia in this province, 65.1% of parents were first cousins, 8.3% were second cousins and 26.6% were non-relatives (2). This indicates that the high rate of consanguineous marriage in this province is one of the reasons behind the incidence of thalassemia.

Other common genetic disorders in this area include a variety of metabolic disorders that can have an effect on the metabolism of amino acids, such as Phenylketonuria (PKU), which has an autosomal recessive inheritance pattern and in general is characterized by mental retardation, fair skin, eczema and epilepsy. Incidence of PKU is approximately 1.6 per 10,000 people in southern Iran (5). In a study on 40 PKU patients in the Khuzestan province, IVS10-11 G > A mutation (similar to other studies in Iran and Mediterranean populations) was the most frequent mutation among patients. Furthermore 12 different mutations were observed in this study (6).

Frequency of GSD-I (another metabolic disorder in the province) is higher than other types of Glycogen Storage Diseases (GSD). Glycogen Storage Diseases have an autosomal recessive pattern and are associated with symptoms such as hepatomegaly and hypoglycemia. GSD-I is caused by defection in glucose-6-phosphatase enzyme.

Niemann-Pick disease has also been observed in this area and types A and B are more common. This disease is caused by deficiency of the sphingomyelinase enzyme and has an autosomal recessive inheritance pattern, with symptoms such as failure to thrive, hepatomegaly and developmental regression.

Deficiency of G6PD is the most common enzyme defect in X-linked recessive inheritance pattern in the world, which causes a variety of symptoms including increased bilirubin levels and acute or chronic hemolysis. In Khuzestan, frequency of G6PD deficiency is estimated to be about 7.6% (7). Also, the frequency of the Mediterranean, Chatham and Cosenza variants were 73.2%, 8.66%, and 2.6%, respectively (7-9).

Another condition that can be observed in the studied province with high frequency is hearing loss. Based on statistics, of 300 born children, one is affected by this condition, whereas 1 or 2 cases per 1000 births is affected elsewhere in the world (10, 11). Hearing loss can be divided into two different types of syndromic and non-syndromic. Also in terms of inheritance pattern it is divided into Autosomal Recessive (AR), Autosomal Dominant (AD) and X-linked. In rare cases, a mitochondrial pattern of inheritance can also be observed. More than 70% of cases are non-syndromic in nature, of which over 80% are inherited with an AR pattern and are said to have Autosomal Recessive Non-syndromic Hearing Loss (ARNSHL) (12). Preliminary studies show that the prevalence of GJB2 mutation rate (the most common cause of deafness in the world) is very low in Khuzestan's population (0 - 4%) (13, 14).

As previously mentioned, the high rate of consanguineous marriage is one of the main reasons behind high prevalence of these diseases. Each of the mentioned diseases could cause psychological, emotional and economic consequences for the patient's family and community. Premarital and prenatal genetic counseling could play an important role in prevention of genetic abnormalities. Therefore increasing public awareness about the importance of genetic counseling could prevent harmful effects.

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References

1. Saadat M, Ansari-Lari M, Farhud DD. Consanguineous marriage in Iran. *Ann Hum Biol.* 2004;**31**(2):263–9.
2. Latifi SM, Zandian KH. Survival analysis of B-thalassemia major patients in Khouzestan province referring to Shafa hospital. *Jundishapur Scient Medic J.* 2010;**9**(1):84–92.
3. Habibzadeh F, Yadollahie M, Merat A, Haghshenas M. Thalassemia in Iran; an overview. *Arch Irn Med.* 1998;**1**(1):27–33.
4. Iran Blood Transfusion Organization.Thalassemia in Iran . Iran Blood Transfusion Organization. Iran: Iran Blood Transfusion Organization; [cited May 2015]. Available from: http://www.ibto.ir/HomePage.aspx?site=ibto&tabid=1&lang=fa-IR.
5. Habib A, Fallahzadeh MH, Kazeroni HR, Ganjkarimi AH. Incidence of phenylketonuria in Southern Iran. *Iran J Med Sci.* 2015;**35**(2):137–9.
6. Ajami N, Kazeminezhad SR, Foroughmand AM, Hasanpour M, Aminzadeh M. A preliminary mutation analysis of phenylketonuria in southwest Iran. *Genet Mol Res.* 2013;**12**(4):4958–66.
7. Kazemi Nezhad SR, Mashayekhi A, Khatami SR, Daneshmand S, Fahmi F, Ghaderigandmani M, et al. Prevalence and molecular identification of Mediterranean glucose-6-phosphate dehydrogenase deficiency in Khuzestan province, Iran. *Iranian J Public Health.* 2009;**38**(3):127–31.
8. Gandomani MG, Khatami SR, Nezhad SR, Daneshmand S, Mashayekhi A. Molecular identification of G6PD Chatham (G1003A) in Khuzestan province of Iran. *J Genet.* 2011;**90**(1):143–5.
9. Kazemi Nezhad SR, Fahmi F, Khatami SR, Musaviun M. Molecular Characterization of Cosenza Mutation among Patients with Glucose-6-Phosphate Dehydrogenase Deficiency in huzestan Province, Southwest Iran. *Iran J Med Sci.* 2011;**36**(1):40–4.
10. Morton NE. Genetic epidemiology of hearing impairment. *Ann N Y Acad Sci.* 1991;**630**:16–31.
11. Dror AA, Avraham KB. Hearing loss: mechanisms revealed by genetics and cell biology. *Annu Rev Genet.* 2009;**43**:411–37.
12. Finsterer J, Fellinger J. Nuclear and mitochondrial genes mutated in nonsyndromic impaired hearing. *Int J Pediatr Otorhinolaryngol.* 2005;**69**(5):621–47.
13. Hosseinipour A, Hashemzadeh Chaleshtori M, Sasanfar R, Farhud DD, Tolooi A, Doulati M, et al. Report of a new mutation and frequency of connexin 26 gene (GJB2) mutations in patients from three provinces of Iran. *Iranian J Public Health.* 2005;**34**(1):47–50.
14. Galehdari H, Foroughmand AM, Soorki MN, Mohammadian G. Absence of mutations in GJB2 (Connexin-26) gene in an ethnic group of southwest Iran. *Indian J Hum Genet.* 2009;**15**(1):9–12.