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The High Birth Rate of Thalassemia Major and the Associated Problems in Sistan and Baluchistan Province, Iran

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Dear Editor,

Thalassemia syndromes are congenital types of anemia resulting from genetic mutations affecting normal production of globin genes. These genetic based disorders are known as the most common causes of inherited anemia worldwide, including Iran. Thalassemia is inherited in an autosomal recessive manner; in other words, the two defective alleles need to be transmitted from both mother and father. Parents with mutated forms of either α or β globin genes are called thalassemia minor or thalassemia carrier. This is clinically characterized with very mild microcytic hypochromic anemia. Thalassemia minor is a relatively common clinical situation observed in all regions of Iran. Prevalence of thalassemia trait is estimated to reach the most in the northern and southern areas with frequency of about 10% and 4% - 8% in other regions (1). Transfusion dependent (major) thalassemia, however, is more likely to result in cases of marital relationship of parents with thalassemia minor phenotype. Sistan and Baluchistan province in Iran is currently a commonplace for incidence of transfusion dependent thalassemia with an estimated number of 2050 registered patients (1). Transfusion dependent thalassemia renders an intense situation for the patients, families, as well as the society and the government. Management of patients with thalassemia major throughout their lifetime requires comprehensive medicinal measures including regular assessment of vital organs such as heart, liver, kidney and endocrine system, as these organs are the most vulnerable to iron deposits from incessant transfusions.

Present treatment and management options for thalassemia major are both expensive and exhausting. Many families complain about having difficulties convincing their children to use iron chelators as injection or tablets, though presently they are the only available therapeutic measures to secure the body to rid itself from excessive iron overload built up as a result of routine transfusion. It should be emphasized that gene therapy is out of reach and bone marrow transplantation is expensive and requires matched donors. Accordingly, preventive procedures are widely encouraged by the health professionals to cope with thalassemia related problems.

Nowadays, the preventive approaches are most widely based on pre-marital screening programs of the couples for thalassemia minor followed by molecular based prenatal diagnostic (PND) for carrier couples. These procedures are undertaken and successfully delivered by the countries with high prevalence of thalassemia such as Italy, Greece and Cyprus (2, 3). Implanting a PND program, however, necessitates understanding of thalassemia causing mutational profile of the populations. This is in progress for different Iranian ethnicities since 1993. It results in determining the most common casual mutations in majority of the ethnic groups (4). The history of PND application in Iran goes back to 1996 when Pasteur institute of Iran, for the first time and other centers thereafter, began to perform PND. To address the regions with highest thalassemia carrier rates, PND centers are established across the country, in some cases by direct governmental supports. Zahedan PND center for thalassemia was founded in Sistan and Baluchistan province in 2002. Zahedan experience and implementation of PND in other regions resulted in sharp reduction in the birth of affected children. According to the Iranian national thalassemia committee, a substantial reduction in overall annual incidence of more than 70% is observed in birth of new cases of thalassemia major indicating relatively successful implementation of screening and PND programs in Iran. In most regions this

number is nearly more than 99%. Nevertheless, about one-fourth of new cases of thalassemia in the Iran live in Sistan and Baluchistan province. This high share may be partly due to high rate of thalassemia carriers in the region; nevertheless, the role of insufficient educational packages in the province as well as other reasons to convince families to come forward for PND cannot be overlooked (5, 6). Authors previous study on 601 new cases of thalassemia major born in the province since undertaking premarital screening and PND policies, revealed that consanguineous marriage (83%) and Baloch ethnicity (90%) were the two most strongly associated parameters of the high incidence of new cases of thalassemia major. Furthermore, the studies indicated that a high proportion (78%) of the couples responsible for birth of new thalassemia cases had not been screened for thalassemia minor before marriage, despite for the compulsion of every couple for screening. This lack of premarital screening would ring the bell for the health professionals to follow such carrier couples to inform the families to come for PND on time (1). This notion highlights the flawed leading future couples, especially those with Baloch origin, to the pre-marital screening centers across the province. This may be the major explanation for rather high number of new cases of thalassemia in the region. Therefore, it is highly recommended to consider more and effective educational programs designed for Baloch population of the regions, as well as to establish more counseling centers for pre-marital screening.

Footnote

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