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ORIGINAL ARTICLE

ATTITUDE TOWARD PRENATAL DIAGNOSIS FOR β-THALASSEMIA MAJOR AND MEDICAL ABORTION IN SOUTHERN IRAN

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□ *The aim of this study was to evaluate the degree of acceptability of prenatal diagnosis and voluntary termination of pregnancy in case of a fetus affected with β-thalassemia major (β-TM) in patients and their parents in Southern Iran. We interviewed 510 parents who had β-TM children and 254 patients. A questionnaire was used to record information regarding sex, age, parents' and patients' knowledge about prenatal diagnosis, and their decision regarding abortion of an affected fetus. Of 764 participants, 565 (73.9%) knew a little about prenatal diagnosis and 198 (25.9%) had no knowledge at all, while 711 (93%) were in favor of prenatal diagnosis and 53 (6.9%) were not. Specifically, 663 subjects (86.7%) were in favor of early termination of pregnancy in case of an affected fetus, while (13.2%) were not. Compliance with prenatal diagnosis was high, and the main reason for declining was its cost. No particular correlations were found between the size of the household, the education or the economic level, and the request for early termination of pregnancy of an affected fetus. The decision not to have a medical abortion correlated with religious beliefs only.*

Keywords Prenatal diagnosis, β-Thalassemia (β-thal), Southern Iran

INTRODUCTION

Hemoglobinopathies are a group of inherited recessive disorders of the hemoglobin (Hb) molecule. The thalassemias in particular are characterized by a defect in the synthesis of the polypeptide chains (globins) needed to assemble the Hb tetramer. Hemoglobinopathies are considered as the most common genetic disorder world wide (1). Thalassemia is most common

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in the Mediterranean and in the regions of Asia and Africa where malaria has been or still is endemic (2). About 3% of the world's population (approximately 150 million people) carry a β-thalassemia (β-thal) mutation (1). Although prenatal diagnosis for β-thal major (β-TM) has been available since 1966, the access to prevention by prenatal diagnosis has remained limited in many countries because of economical, technical or ethical reasons. The methods used for prenatal diagnosis in general can be subdivided between invasive and non invasive. The first imply chorionic villus sampling (CVS), amniocentesis or cordocentesis. The second includes maternal serum sampling for fetal DNA, α-fetoprotein measurement, ultrasonography and isolation of fetal cells from the maternal circulation (3). The alternative to prenatal diagnosis is preimplantation genetic diagnosis (PGD), done in combination with in vitro fertilization (IVF). Preimplantation genetic diagnosis is a procedure used to identify genetic disorders in an embryo through IVF (4). This alternative is complex and laborious, is usually offered to couples with fertility problems and the genetic analysis is done on the early embryo before insemination. In a study by Palomba et al. (5), women at risk for β-TM in the progeny were interviewed before CVS. All women who had a previous therapeutic abortion found PGD blastocentesis acceptable. Only 30% of the women who had no previous therapeutic abortion selected PGD blastocentesis, while 25% of primi gravida women opted for PGD. From these figures it seems that obstetric experience is an important factor in the reproductive choice of women at high genetic risk.

Iran is located in the region where the prevalence of β-thal carriers is high. The disorder is more prevalent in the north (near the Caspian Sea) and south (near the Persian Gulf). The prevalence of heterozygous β-thal in these regions is between 4 and 8% (6). In 1991, a premarital screening program for the prevention of β-TM was started. However, Iran is an Islamic country where abortion has long been illegal, and severe restrictions by religious authorities essentially prevented abortion until only a few years ago (7). This study was undertaken to evaluate the compliance with prenatal diagnosis and early therapeutic abortion of a fetus affected with β-TM in patients and their parents in Southern Iran.

MATERIALS AND METHODS

From March 2006 to March 2007 a total of 764 persons were enrolled for this study. One group consisted of 510 parents of children with thalassemia (377 mothers and 133 fathers, average age 50.3 years old). The second group consisted of 254 thalassemia patients older than 16 years (147 females and 107 males, average age 22.5 years old). The patients attended the Shiraz Thalassemia Center for transfusion therapy. A questionnaire was

designed to include demographic information such as gender, age, number of children in the family, number of children with β-TM, educational level and economical status (low: monthly income up to 200, middle: between 200–400, and high: over 400 US $), use of contraceptive methods, knowledge about prenatal diagnosis, access to prenatal diagnosis and their opinion regarding termination of pregnancy. The reasons underlying acceptance or non acceptance of prenatal diagnosis and termination of pregnancy was requested. Reasons involving emotional, economical and religious aspects, the possibility of taking care of the severely affected patients and the hope for an effective cure sometime in the future, were recorded. The questionnaire was confidential and the parents were interviewed together. Each interview took about 20 minutes and was conducted by a trained doctor. Written consent was obtained from each participant. Chi-square and Mann Whitney tests were used for statistical analysis, a p value <0.05 was considered statistically significant.

RESULTS

Thirty-three point two percent of responders were thalassemia patients and 66.7% were parents. Of 171 (67.3%) patients and 266 (52.1%) parents the education level was less than high school diploma. Economic level was middle income in 29.2–51.8% (mean 40.5%) of the families. Sixty-eight percent of the families had one thalassemic child, 23.5% had two. One family had five affected children. The quality of life was reported to be heavily influenced by β-TM by 482 (94.5%) of the parents, while only 152 (59.8%) patients had the same opinion. The factors influencing the quality of life are summarized in Table 1.

As many as 172 (67.7%) patients and 393 (77%) parents reported having received insufficient information about prenatal diagnosis, and the remaining participants had received no information at al. Of all 254 patients with β-TM, 250 (98.4%) were in favor of prenatal diagnosis, and almost the same percentage was registered in the parents (90.3%). The

TABLE 1 Factors Affecting the Quality of Life Reported by Patients with TM and Their Parents

	Parent (n = 510)		Patients (n = 254)	
	Cases	%	Cases	%
Unemployed	23	4.5	2	0.78
Psychological problems	7	1.3	13	5.10
Economic problems	13	2.5	3	1.10
Combination	440	86.2	232	91.30
Miscellaneous	27	5.3	4	1.50

TABLE 2 Reasons for Non Compliance with Medical Abortion in TM Patients and Their Parents

	Parents ($n = 44$)		Patients ($n = 57$)	
Reasons	Cases	%	Cases	%
Religious motives	34	77.2	24	42.1
Hope of future treatment	2	4.5	8	14.0
Emotional reasons	6	13.6	12	21.0
Spousal disagreement	1	2.2	2	3.5
Miscellaneous	1	2.2	11	19.2

most important reasons why parents and patients rejected prenatal diagnosis were, respectively, the cost (66 and 50%) and religious beliefs (20 and 50%).

The attitudes toward medical abortion were different. Of the parents, 466 (91.3%) accepted this option, whereas only 197 (77.5%) of the patients found it acceptable. No correlation was found between these data and the level of education or the economic status after having at least one affected β-TM child. The reasons for non compliance with therapeutic abortion are summarized in Table 2.

DISCUSSION

Early screening for thalassemia trait is one of the most important ways to decrease the incidence of this disease. The goal of thalassemia screening is to identify carrier couples prior to conception or birth of an affected child in order to offer information and prospective prevention. In Iran, a national prevention plan has been offered to the population for the last 18 years; the first started in Shiraz (South Iran) and 4 years later, in all parts of Iran. Currently, more than nine clinics provide prenatal diagnosis services to thalassemic patients (7,8).

In this study we have demonstrated that prenatal diagnosis and medical abortion are welcome in South Iranian couples at-risk for β-TM and are even accepted by most of their children. The low number of young parents with affected children indicates that the Iranian prevention program is welcome; most parents in our cohort were 35 years or older. The age of the mothers who were between 31–35 years old showed the importance of information and family planning, and 95.9% of responders were currently using contraception methods. In a study by Han et al. (9) in Myanmar in 1992, contraception methods were used only by 62% of the parents with affected children.

Our data also show that the higher the information and education level of the mothers, the less number of children (and also β-TM children) in the

families ($p < 001$). Thus, informing mothers can play a substantial role in decreasing the number of infants born with thalassemia, while the number of affected newborns correlated also with the family's economic status ($p = 0.03$).

The knowledge of prenatal diagnosis correlated directly with socioeconomic and educational status. Therefore, more attempts to train and inform carriers of thalassemia are potentially useful. The acceptability of medical abortion had no significant relationship with socioeconomic or educational levels, and in spite of religious beliefs, the acceptability of selective abortion was very high. This may be due to difficulties in the availability and quality of medical care for patients with β-TM and with the loss of hope regarding the development of new effective therapies for the disease. The belief that the birth of an affected child can negatively affect a family's quality of life may be an additional reason for the wide acceptance of selective abortion. Genetic counselors and doctors provide realistic information on this matter and therefore most patients (77.5%) and parents (91.3%) agreed with abortion even at a late gestational age of more than 6 months. In this category, parents who were not in favor of late abortion, considered religious beliefs (79.1%).

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REFERENCES

1. Borgna-Pignatti C, Galanelli R. The thalassemias and related disorders: quantitative disorders of hemoglobin synthesis. In: Greer JP, Foester J, Rodgers GM, Eds. Wintrobe's Clinical Hematology, 12th ed. Philadelphia: Lippincott, Williams and Wilkins, 2009;1083–1131.
2. Cunningham MJ, Sankaran VG, Nathan DG, Orkin SH. The thalassemias. In: Orkin SH, Nathan DG, Ginsburg D, Eds. Hematology of Infancy and Childhood, 7th ed. Philadelphia: Churchill Livingstone, 2000;1015–1106.
3. Cao A. Carrier screening and genetic counselling in β-thalassemia. Int J Hematol. 2002;76(Suppl. 2):105–113.
4. Thornhill AR, Snow K. Molecular diagnosis in preimplantation genetic diagnosis. J Mol Diagn. 2002;4(1):11–29.
5. Palomba ML, Monni G, Lai R, Cau G, Olla G, Cao A. Psychological implications and acceptability of preimplantation diagnosis. Hum Reprod. 1994;9(2):360–362.
6. Abolghasemi H, Amid A, Zeinali S, et al. Thalassemia in Iran; epidemiology, prevention and management. J Pediatr Hematol Oncol. 2007;29(4):233–238.

7. Karimi M, Jamalian N, Yarmohammadi H, Askarnejad A, Afrasiabi A, Hashemi A. Premarital screening for β-thalassaemia in Southern Iran: options for improving the programme. J Med Screen. 2007;14(2):62–66.
8. Yong KN, Wadsworth D, Langlois S, et al. Thalassemia carrier screening and prenatal diagnosis among the British Columbia (Canada) population of Chineses descent. Clin Gene. 1999;55(1):20–25.
9. Han KE, Han AM, Myint T. Thalassemia in the Outpatient Department of the Yangon Children's Hospital in Myanmar: knowledge, attitudes and practice in relation to thalassemia. Southeast Asian J Trop Med Public Health. 1992;23(2):269–272.