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ACCEPTANCE OF FIRST-TRIMESTER PRENATAL DIAGNOSIS FOR THE HAEMOGLOBINOPATHIES IN LEBANON

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SUMMARY

We have interviewed 83 couples at risk for a haemoglobin disorder, mostly β-thalassaemia, in an effort to evaluate their attitude towards first-trimester prenatal diagnosis. Most of the families had received poor education and were of low socio-economic status and more than half of the couples were not properly aware of their genetic risk. Fifty-nine per cent of the couples were definitely in favour of prenatal diagnosis, 23 per cent were uncertain at the time of the interview, and 18 per cent were opposed to such testing, because of their religious conviction against termination of a pregnancy. Another important factor which seems to influence choice was the cost of the test. Essential issues that arise from this study include the importance of a control programme adapted to particular populations, proper information and counselling, and the need for financial support in countries such as Lebanon.

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INTRODUCTION

Lebanon is a small country lying on the eastern shores of the Mediterranean. As in other Mediterranean countries, the most common genetic disorder in Lebanon is β-thalassaemia, the estimated carrier frequency being 2–3 per cent (Cabannes *et al.*, 1965). Since consanguineous marriages represent at least 25 per cent of all marriages, mostly between first cousins (Der Kaloustian *et al.*, 1980; Khlat *et al.*, 1986), the expected birth rate of homozygotes is around 0·4/1000. There are other, less frequent haemoglobinopathies, mainly sickle-cell disease, but no estimate of the frequency of these disorders is currently available.

Since the end of the civil war in Lebanon, there has been increased awareness about β-thalassaemia, via national prevention campaigns and the opening of a specialized centre (Chronic Care Centre) offering care and support to affected patients and their families. In most European countries with a high incidence of β-thalassaemia, effective control programmes involving premarital screening, identification of couples at risk, genetic counselling, and prenatal diagnosis have been implemented. Premarital screening has been recently made compulsory in Lebanon; however, there are currently no data concerning the effects of genetic counselling and acceptance of prenatal diagnosis in countries such as Lebanon. In fact, most studies have been conducted in North America and Europe and show high levels of uptake of prenatal diagnostic procedures (Modell *et al.*, 1984; Lippman *et al.*, 1985; Perry *et al.*, 1985; McGovern *et al.*, 1986; Cao *et al.*, 1987;

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Spencer and Cox, 1987, 1988), but this might not be the case in Middle-Eastern societies, because of different cultural and religious beliefs.

Lebanon is a mosaic of different religious communities, of various educational and socio-economic backgrounds. The various communities can be grouped broadly into Muslims and Christians. The Muslim communities consist of the Sunnis, the Shiites, and the less numerous Druze community. The Christian communities consist mainly of the Maronites, the Greek Orthodox, Greek Catholics, Protestants and Latins, and the Armenians. Numerically, the three predominant communities by far are the Maronites, Shiites, and Sunnis. A number of non-Lebanese groups also reside in Lebanon.

β-Thalassaemia is not evenly distributed across the different communities (Rafie and Rafie, 1986). The frequency of affected individuals is highest among the Sunnis, closely followed by the Shiites, whereas only two Druze families with β-thalassaemia are currently documented. The disorder is less frequent in Christian communities, where it is found mostly among the Maronites. The only patient with a haemoglobin disorder currently documented among Armenians has sickle cell/β-thalassaemia.

The Lebanese health system is private and patients have to pay for their medical expenses. Under Lebanese law, social abortion is theoretically illegal and abortion for medical reasons is acceptable only if the pregnancy represents a threat to the mother. However, both types of abortion are commonly performed on a private basis. Taking all these points into consideration, we decided to conduct a study to estimate the acceptance of first-trimester prenatal diagnosis among couples from a high-risk group. We therefore interviewed 83 couples at risk for a haemoglobinopathy, mostly β-thalassaemia, who have had previously at least one affected child, noting the different variables that might affect their choice, informing them about the genetic risk and first-trimester prenatal diagnosis by chorionic villus sampling (CVS), and encouraging them to express freely their attitudes towards such testing.

MATERIALS AND METHODS

Eighty-three couples of child-bearing age, with at least one affected child, were randomly selected for an interview as they presented at the centre (Chronic Care Centre). It is believed that nearly all families with a haemoglobin disorder, from a variety of backgrounds, are registered at this centre. All the interviews were conducted by an experienced geneticist or a trained nurse educator. Each interview comprised three major sections. Section I recorded information about the couple's religious, educational, and socio-economic backgrounds (based on the occupation of the father). Section II gathered information about their family and reproductive history, awareness of their genetic risk, method of contraception, and understanding of prenatal diagnosis. Their genetic risk was explained to them in case it was unknown or misunderstood. Section III started with an illustrated explanation about the CVS procedure, its advantages and risks, laboratory procedures, reliability of the test, etc. The couples were then encouraged to express freely their feelings about the test, whether they would accept it or not, and the reasons for their choice. Since most families would have to pay for the test themselves, the cost of the test (400 U.S. dollars) was estimated to be an important variable and was mentioned afterwards. The couples were then asked how this issue would affect their choice.

RESULTS

The religious distribution of the couples in our sample closely resembles the distribution of the haemoglobinopathies in Lebanon, where the majority of affected individuals (80 per cent) are Muslims, mostly Sunnis and Shiites, while among the Christian communities (20 per cent), the majority are Maronites. The geographical origin of the couples in our sample also represents the distribution of the disorder, which is mainly found around the littoral (73·4 per cent of cases) [North, Beirut (the capital), and South], where malaria was endemic, and less so in the mountains and central plains (Rafie and Rafie, 1986). The majority of the couples were poorly educated: 71 per cent of husbands and 67·4 per cent of wives had not received secondary schooling. Most couples (80·7 per cent) were of low socio-economic status, as assessed by the occupation of the husband.

Table I summarizes the medical history of the couples studied. The great majority of couples (90·4 per cent) were at risk for β-thalassaemia. Eight couples were at risk for sickle-cell anaemia or sickle thalassaemia. Social background, family

Table I—Medical history of the study population

	No. (percentage)	
Referral		
β-Thalassaemia	75 (90·4%)	
Sickle-cell anaemia	5 (6·02%)	
Sickle/β-thal	3 (3·6%)	
Parental consanguinity		
Negative	29 (34·9%)	
First cousins	42 (50·6%)	
Second cousins	9 (10·8%)	
Father	3 (3·6%)	
No. of affected children		
1	58 (70%)	
2	19 (23%)	
3	5 (6%)	
>3	1 (1%)	
Family size (number of children)		
1	8 (9·6%)	
2	25 (30%)	
3	22 (26·5%)	
4	14 (16·7%)	
5	6 (7·2%)	
6	4 (4·8%)	
7	2 (2·4%)	
8	1 (1·2%)	
10	1 (1·2%)	
Pregnancy losses	SA	TOP
None	54 (66%)	66 (79·5%)
1	19 (22·9%)	15 (18%)
2	5 (6%)	2 (2·4%)
3	4 (4·8%)	—
4	1 (1·2%)	—
More children wanted		
Yes	32 (39%)	
Maybe	15 (18%)	
No	36 (43%)	
Contraception (in couples who did not want additional children)		
Reliable	22 (61%)	
Unreliable	10 (27·8%)	
None	4 (11·2%)	

SA = Spontaneous abortion; TOP = termination of pregnancy.

Table II—Awareness of the disorder in the study population

	No. (percentage)
Other affected family members	
No	62 (74·7%)
Yes	21 (25·3%)
Awareness of genetic risk	
Good	39 (47%)
Little	27 (32·5%)
Wrong	6 (7·2%)
None	11 (13·3%)
Awareness of prenatal diagnosis	
Good	53 (64%)
Not clear	18 (21·6%)
None	12 (14·5%)
Acceptance of prenatal diagnosis	
Yes	49 (59%)
Uncertain	19 (23%)
No	15 (18%)

size, and responses from these couples were similar to those at risk for β-thalassaemia and therefore the results were grouped. Sixty-five per cent of couples were consanguineous and, among these, 77·8 per cent were first cousins. The average paternal age was 36·7 years and the average maternal age was 30·6 years. All couples studied had at least one affected child, but 30 per cent had more than one. Eight families in total had experienced the death of at least one child; in four of these families, the death was because of β-thalassaemia, and the number of affected deceased children ranged from 1 to 4. Sixty-six and 79·5 per cent of couples had not experienced a spontaneous abortion or a pregnancy termination, respectively. In 16 out of the 17 pregnancy terminations, the reason was fear of giving birth to an affected child. When asked whether they wanted additional children, 32 couples (39·7 per cent) expressed such a wish. All couples who had only one child were in this category, and the rest had a variable number of children (data not shown). Three of the women were actually pregnant when interviewed. Forty-three per cent of the couples did not want more children and in fact all the couples in this group already had two or more children. However, approximately 40 per cent of the couples in this group were not using a reliable method of contraception.

Table II summarizes the answers to questions concerning awareness in the couples about the disorder and their attitudes towards prenatal diagnosis. Twenty-five per cent of the couples had previously heard of the disorder because of another affected family member, but had not thought that they were at risk. The majority of the

couples, however (75 per cent), had never heard of the disorder in their family before. When asked about genetic risk, 53 per cent of the couples had little or no awareness of, or a complete misunderstanding about their genetic risk. This ranged from not knowing that they were at risk at all to thinking that their risk was 90 per cent or misinterpreting the 1 in 4 risk as a guarantee that once they had had an affected child, the following three would be normal. On the other hand, 64 per cent of the couples had heard of prenatal diagnosis, either through their doctor, a friend, or more recently from the centre. Seven women had actually undergone prenatal diagnosis previously. However, 36 per cent of the couples interviewed either had a wrong idea about prenatal diagnosis (21·6 per cent) or had not heard of it previously (14·5 per cent). Misconceptions about prenatal diagnosis included a high risk to the fetus or the mother, low accuracy of the results, and difficulty of the procedure.

The procedure of CVS and its advantages and risks, as well as the laboratory procedures, were explained to all couples in detail before they were asked to express their attitudes to such testing and whether they would opt for it. Fifty-nine per cent of couples agreed that they would definitely request prenatal diagnosis in a future pregnancy. However, 23 per cent of couples were uncertain at the time of the interview and 18 per cent were definitely opposed to prenatal diagnosis.

Among couples who favoured prenatal diagnosis, 37 per cent expressed concern about its cost and said that they would need to secure the required amount before asking for the test. Of the 19 couples who were uncertain about the test at the time of the interview, the reasons for their indecision were a disagreement between the husband and wife in nine cases, the elevated cost of the test in four cases, a personal choice in three cases, a religious dilemma in two cases, and a previous unpleasant experience with prenatal diagnosis in one case.

In Table III, the profile of couples opposed to prenatal diagnosis is summarized. All 15 couples in this group are at risk for β-thalassaemia. However, comparisons are made difficult by the very fact that the disorder is not evenly distributed across all religious communities, and that most families with β-thalassaemia are in fact of poor education and low socio-economic background. Still, some observations can be made. In our study population, 80 per cent of couples were of the Muslim faith and 20 per cent were Christians. Of the couples opposed to prenatal diagnosis, all but one (7 per cent) were Muslims, equally divided between the Sunni and the Shiite sects. The level of education in this group was variable and all couples were of low or middle socio-economic status. Nine couples had one affected child, five had two, and one couple had three affected children. When asked about the reason for their opposition to prenatal diagnosis, all couples admitted that it was essentially because of religious convictions against termination of pregnancy. In other words, none of these couples was opposed to prenatal diagnosis itself, but rather to termination of pregnancy in case of an affected fetus. When asked what they would do in their next pregnancy, all the couples who were opposed to prenatal diagnosis, including one expecting a baby at the time of the interview, said that they would continue the pregnancy and take the risk. On the other hand, couples who expressed unease about the cost of the test said that they would either secure the cost before planning the next pregnancy or take the risk if the sum was not available.

Table III—Profile of couples opposed to prenatal diagnosis (total: 15)

Religion		
Muslims	14 (7 Sunnis, 7 Shiites)	
Christian	1 (Maronite)	
Education		
	Husband	Wife
None	—	2
Primary school	9	6
Middle school	2	4
Secondary school	2	3
Higher	2	—
Socio-economic background		
Low	13	
Middle	2	
High	—	
No. of affected children		
1	9	
2	5	
3	1	

DISCUSSION

The haemoglobinopathies are the most common hereditary disorders in the world. However,

through appropriate control programmes, prevention at the community level is possible (Modell, 1994). An important aspect of such control programmes is genetic counselling and the offer of prenatal diagnosis. The level of demand for prenatal diagnosis in a particular population is one important variable in the efficiency of such a programme (Modell, 1994). Most studies on the acceptance of prenatal diagnosis have been conducted in North America or Europe. More specifically, acceptance of first-trimester prenatal diagnosis for β-thalassaemia has been evaluated, for instance, in Sardinia (Cao *et al.*, 1987), where it was found that practically all couples (99·1 per cent) would choose CVS, as opposed to a second-trimester diagnosis, probably because a first-trimester termination of pregnancy is morally more acceptable to this Catholic population. In addition, first-trimester diagnosis allows privacy and reduces anxiety. Another study was done in a Muslim community in Europe, the British Pakistanis (Petrou *et al.*, 1990). The study showed that, contrary to unfounded misconceptions, many British Pakistani families at risk for β-thalassaemia would request first-trimester diagnosis. A study of a sample of thalassaemic families in Lebanon has been conducted previously in order to evaluate aspects of burdens on families and to assess their awareness of the disease and prenatal diagnosis (Der Kaloustian *et al.*, 1987). In this study, it was found that approximately half of the 44 families interviewed would opt for prenatal diagnosis. However, the study does not specify the prenatal diagnostic procedures suggested nor does it discuss the reasons governing the choice that the families were making regarding these procedures.

It appears from our study that the haemoglobinopathies are mostly found among families of lower socio-economic status and poorer education in Lebanon. This might correlate with reduced awareness of genetic risk, increased parental consanguinity, and reduced acceptance of prenatal diagnosis. In our study, 59 per cent of the couples interviewed would definitely request prenatal diagnosis, while 18 per cent would be opposed to it on religious grounds. All couples in the latter category would be willing to take the risk in their next pregnancy. One important factor that seems to influence couples who either accept or were uncertain (23 per cent) about prenatal diagnosis is the cost of the test. This is not surprising, since most families at risk for a haemoglobinopathy have a low income and cannot afford this cost. Although first-trimester prenatal diagnosis procedures can currently be performed in Lebanon, samples have to be sent abroad for analysis in specialized laboratories, usually to the United Kingdom or Cyprus, adding to the basic cost.

Parental consanguinity seems to play an important role in the incidence of the disorder in Lebanon. Sixty-five per cent of couples studied were consanguineous, 77·8 per cent of them being first cousins, which is higher than the percentage among the general population of Beirut (25 per cent) previously reported (Khlat *et al.*, 1986). However, the issue of genetic counselling in relation to consanguineous marriage is not an easy one to approach. Consanguineous marriage has important social roles, especially to women, which cannot be overlooked in various communities (Khlat *et al.*, 1986) and many geneticists agree that the associated genetic risk does not provide adequate grounds for a campaign against consanguineous marriage (Bittles, 1980; Petrou *et al.*, 1990; Modell and Kuliev, 1992). The problem for the haemoglobinopathies can probably be approached by offering, as a first step, premarital screening, which has been recently implemented in Lebanon. Before receiving a marriage licence from their respective religious authority, couples now have to undergo a series of required blood tests, which include MCV (mean corpuscular volume). In the case of an abnormal MCV, haemoglobin electrophoresis is performed. However, proper counselling following this initial screening is equally important and cannot be overlooked (Fletcher *et al.*, 1985). Currently, no guidelines are followed regarding this counselling in Lebanon and it is not known how the counselling, if any, is provided.

Important aspects that seem to arise from this study are the need to adapt particular control programmes to particular populations. Providing proper information, screening, and counselling are important issues, but equally important is the provision of care and support regardless of the choice that the family makes, taking into consideration social, ethical, moral, and religious dilemmas that might specifically characterize each couple. Since the health system in Lebanon is private and so far couples have to secure the money to pay for prenatal diagnosis, financial support is also required in order to better implement such a control programme.

The couples included in our study were all at risk for a haemoglobin disorder, mainly β-thalassaemia. Although our sample seems to be representative for the thalassaemic families in Lebanon, it does not represent the whole Lebanese population, and therefore our data cannot be extended to the entire population, nor to other disorders or risk groups.

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