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Genetic Counseling for Childless Women at Risk for Duchenne Muscular Dystrophy

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The aim of the present study was to assess the impact of genetic counseling in young women at risk to have Duchenne muscular dystrophy (DMD) children prior to childbearing. A total of 263 potential DMD carriers, who had had genetic counseling and were given different genetic risks, were included in this investigation. Their reproductive outcome and future plans as well as their requests for DNA tests (for carrier detection and prenatal diagnosis) were analyzed according to genetic risk magnitude, comprehension of genetic counseling issues, family and personal history, socio–educational level, and subjective opinion about selective abortion. We noted that genetic risk magnitude had no significant influence on reproductive plans or outcome nor on the request for additional DNA testing, even considering only those clients with good comprehension and retention of issues discussed during genetic counseling. On the other hand, counselees who had more than one affected or at least one deceased DMD case in their family understood genetic counseling significantly better, suggesting that "learning with life" has a stronger impact than genetic counseling. Am. J. Med. Genet. 86:447–453, 1999. © 1999 Wiley-Liss, Inc.

KEY WORDS: family planning; genetic counseling; Duchenne muscular dystrophy; psychosocial

INTRODUCTION

Most articles concerning reproductive decisions after genetic counseling assess clients who already have had children, affected or not, prior to counseling [Eggers and Zatz, 1998; Sissine et al., 1981; Sorenson et al., 1987; Swerts 1989; Wertz and Sorenson, 1986; Wertz et al., 1984] and most of them include many different diseases in their cohorts. In the present investigation we analyzed the understanding of genetic counseling and the reproductive outcomes of 263 women childless before genetic counseling who were at risk for Duchenne muscular dystrophy (DMD).

During the past 30 years more than 1000 DMD families have been diagnosed and counseled in our center. Many of them have been followed by the same team for many years, allowing us to evaluate the long-range effects of counseling in young generations who knew about their genetic risks before reproducing. Because our laboratory is connected to the Brazilian Association for Muscular Dystrophy (ABDIM) where patients come periodically for physiotherapy, art therapy, and specific group activities, a continuous follow-up and contact with professionals from the responsible team is possible. The aim of this study was to assess the impact of genetic counseling on women related to boys affected with DMD prior to childbearing, focusing on their future reproductive plans, their requests for DNA tests, and their reproductive outcome considering genetic risk magnitude, comprehension of genetic counseling issues, family and personal history, socio–educational level, and opinion about selective abortion.

PATIENTS AND METHODS

The present study focused on 263 women at risk for DMD who were counseled in our Center at least 3 years ago before they had children. All were older than 18 at the last contact with our staff. Most of them are sisters of DMD patients (64%) whereas the rest are cousins, aunts, or nieces.

The diagnosis of DMD was based on clinical and neurological evaluation, family history, course of the disease, serum creatine kinase activities, DNA analysis, and dystrophin assessment in muscle biopsy.

Genetic risk, defined here as the risk for DMD in a male baby (or fetus in the case of prenatal diagnosis), was estimated taking into account family history, serum creatine kinase levels, and the mutation of the propositus. The probability of germinal mosaicism ac-

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cording to the site of the deletion in the dystrophin gene [Passos-Bueno et al., 1992] was also considered. In cases that had been counseled before the introduction of DNA technology, risk estimations were based only on the family history and on serum creatine kinase levels. Genetic risks were classified as very low (1–4%), low (5–9%), intermediate (10–24%) and high (>25%) for simplicity at reporting our findings. These judgmental statements do not, however, reflect the counselees' opinion about the magnitude of the risk.

Data were collected on the counselee's personal and family history and on several aspects observed during genetic counseling and follow-up sessions, as summarized below:

Genetic counseling included at least one session when the etiology, prognosis, and management of the disease, the counselee's genetic risk, carrier detection, prenatal and preclinical diagnosis, psychosocial and ethical aspects, her interest and emotional status were discussed according to the client's professional and cultural level. The genetic risk was given in percent and, if necessary, this figure was explained with the aid of simple examples. Whenever the counselee was married or had a stable relationship, both were encouraged to attend the sessions. If the couple decided that they do not want to risk an affected child, the counselor talked about different contraceptive methods (including selective abortion). All counselees were given a written report on the main aspects discussed during the genetic counseling, including the estimated genetic risk in percent.

After the first counseling session the counselees were recontacted about their understanding of the information discussed during genetic counseling. For the comprehension and retention of information received during genetic counseling, the following arbitrary scores were given by one of us (R. C. M. P.): 0 = very poor (if the counselee thought that DMD was transmitted by a virus, for example); 1 = reasonable (if she knew about her genetic risk, but thought she "should not" have children); and 2 = good (if she knew about the magnitude of her genetic risk and could explain the type of inheritance in her own words).

Reproductive plans were classified as "wants children" when the counselee expressed the wish for biological children during follow-up sessions. If she stated that she wanted no children at all, or wanted to adopt children, she was considered as wanting no children. In addition, a group who had undergone tubal ligation also was classified as "wanting no children" for statistical purposes.

Reproductive outcome represents the number of children born after genetic counseling, including healthy as well as DMD affected children.

In the item *requests for DNA tests*, prenatal diagnosis and carrier detection tests were considered together for statistical purposes. *Attitude towards abortion* was classified as "favorable" (if the client and her husband or partner accepted it), "unfavorable" (if they were against it), and "other" (if there was no consensus among the couple or if the husband's opinion was unknown). The criteria established for the Brazilian Census from [IBGE, 1980, 1996] were used for the classification of propositi according to their *Socio–educational level*. *Statistical analysis* included chi-square and contingency tables.

RESULTS

Demographic data (Table I) showed no significant difference among the four groups classified according to genetic risks, except for the higher socio–economic level of counselees with intermediate risk.

Genetic risk magnitude and reproductive outcome: The mean number of children per recontacted counselee was 0.47 with no significant difference, $P = 0.46$, among the four groups, but most of these women (65%; mean age: 25 years) have not had children yet. Considering the remaining 35%, that is, the 91 women who reproduced after counseling, the mean number of children was 1.39, with no significant difference, $P = 0.73$, among the four groups. Women with a genetic risk >25% did not reproduce significantly less after genetic counseling, did not request DNA tests more frequently, and did not express fewer reproductive plans than those with genetic risks (≤24%) (Table II), even con-

TABLE I. Data From Counselees at Different Risks for DMD[†]

Genetic risk	Very low <5%	Low 5–9%	Intermediate 10–24%	High >25%	Total *N*
Number of counselees	72	81	43	67	263
Mean age at genetic counseling	20.43 ± 7.49	16.94 ± 7.04	18.89 ± 5.17	16.45 ± 8.42	17.41 ± 6.85
Mean age at assessment	25.38 ± 5.47	25.09 ± 5.83	24.77 ± 5.84	24.78 ± 6.25	25.01 ± 5.85
Married (%)	57	63	58	67	162
Had children after genetic counseling (%)	33	33	30	40	91
Number of normal children	30	40	13	33	116
Number of boys with DMD	0	3	1	4	8
Children/married woman	0.73	0.84	0.56	0.82	0.72
Requested DNA test (%)	24	30	47**	19	74
Carriers among those tested (%)	23	29	20	62	23
Against abortion (%)	41	35	16	44	41/113
Wants (more) children (%)	75	48	84	67	60/90
Good comprehension/retention (%)	64	70	79	67	106/154
Lower socio–educational level (%)	65	63	49*	70	166/263

[†]Data from cases with complete information.
*$P < 0.05$; **$P < 0.01$.

TABLE II. Genetic Risks, Reproductive History, Socio–Educational Level, and Reproductive Decisions*

	Yes (%)	No (%)	*N*	*P*
All counselees:				
Genetic risk (%)	Had children after genetic counseling?			
>25	40	60	67	ns
≤24	33	67	196	
	Requested DNA test?			
>25	19	81	67	ns
≤24	31	69	196	
	Wants (more) children?			
>25	68	32	28	ns
≤24	66	34	62	
Had children after genetic counseling?				
Yes	63	37	19	ns
No	68	32	71	
Requested DNA test?	Against abortion?			
Yes	18	82	40	<0.01
No	46	54	73	
Has lower socio–educational level?	Had children after genetic counseling?			
Yes	39	61	166	ns
No	28	72	97	
	Requested DNA test?			
Yes	19	81	166	<0.01
No	43	57	97	
	Good comprehension of genetic counseling?			
Yes	57	43	91	<0.01
No	86	14	63	
	Against abortion?			
Yes	52	48	62	<0.01
No	18	86	49	

*N, cases with complete information; ns, not significant.

sidering only those who showed a good comprehension of information received during genetic counseling (Table III). Sisters of DMD patients did not reproduce less or request DNA tests more often than aunts, cousins, or nieces (Table IV), although significantly, $P < 0.01$, more of them (75.0% vs. 28.72%) had an intermediate to high risk (data not shown). The only factor that was significantly associated with a reduction in the reproductive outcome was the good comprehension of genetic counseling issues, which, in turn, is associated with a higher socio–educational level (see below). However, it is important to note that even in this subgroup of women there was no correlation of the magnitude of the risk and the reproductive outcome (Table III).

The *comprehension of genetic counseling* issues was significantly correlated with the socio-educational level (Table II), as well as with the number of affected and deceased DMD relatives (Table IV). *DNA tests* were significantly more often requested by women who belonged to higher socio–educational levels, who were favorable inclined towards selective abortion (Table II), and who had more than one affected and/or at least one DMD deceased relative in their family (Table IV). The differences among counselees seeking carrier detection (n = 12) in comparison to those requesting prenatal diagnosis (n = 62) will be analyzed in a future study.

Future family planning was not found to be significantly influenced by any of the factors analyzed, probably due to the relatively small sample size. One hundred two counselees were not yet sure about their reproductive plans. They were excluded from this particular analysis and will be studied in a different setting.

DISCUSSION

Our former study on the effects of genetic counseling in DMD [Zatz, 1983] showed that mothers had a better knowledge of issues discussed during genetic counseling and also reproduced significantly less than sisters of boys affected with DMD. Different from that or other reports [for example, Sissine et al., 1981; Sorenson et al., 1987; Swerts, 1987 Wertz and Sorenson, 1986; Wertz et al., 1984], the present study only includes women childless before genetic counseling. This design was chosen to ensure that past reactions toward pregnancy and the birth of affected or normal children would not interfere with family planning. Furthermore, we noted that almost half of DMD mothers were already sterilized when referred to genetic counseling. Tubal ligation is a common parental decision in Brazil when no further children are desired. This option is more often chosen by women of lower socio–educational levels and is usually carried out during Caesarean section (for which Brazil accounts with one of the highest rates worldwide). The fact that almost half of DMD mothers are sterilized at genetic counseling obviously invalidates any attempt to evaluate their further reproductive outcome afterwards. In addition, the present study also has the advantage of focusing on a large

TABLE III. Comprehension of Genetic Counseling Information and Reproductive Decisions*

	Yes (%)	No (%)	*N*	*P*
Counselees with good versus reasonable or bad comprehension of genetic counseling:				
Good comprehension (any genetic risk)?	Had children after genetic counseling?			
Yes	39	61	106	<0.01
No	70	30	48	
	Is against abortion?			
Yes	19	81	62	<0.01
No	59	41	27	
	Requested DNA-test?			
Yes	37	63	106	ns
No	29	71	48	
	Wants more children?			
Yes	56	44	63	ns
No	63	37	19	
Counselees with good comprehension of genetic counseling:				
Genetic risk (%)	Had children after genetic counseling?			
>25	36	64	33	ns
≤24	29	71	73	
	Requested DNA test?			
>25	27	73	33	ns
≤24	42	58	73	
	Wants (more) children?			
>25	56	44	25	ns
≤24	55	45	38	

*N, cases with complete information; ns, not significant.

group of women (N = 263) at different risks for the same disease, and who have been studied and counseled by the same professionals. For these reasons, the present study is not comparable to others.

According to Kessler [1989] genetic counseling is effective in educating counselees about recurrence risks and diagnostic issues, but improvement is needed because many remain poorly informed. On the other hand, seeking advice about reproduction is frequently observed even among well-informed counselees and often leads to problems related to the issue of nondirectiveness during genetic counseling [Kessler, 1992, 1997]. Specifically concerning reproductive outcome after genetic counseling, some investigators report that genetic risk estimates have influenced family planning [Abramovsky et al., 1980; Evers-Kiebooms and van der Berghe, 1979], whereas others report that the past reproductive history has been more important than the genetic risk [Eggers and Zatz, 1998; Sissine et al., 1981; Sorenson et al., 1987; Swerts 1989; Wertz and Sorenson, 1986]. Another perspective on this issue was given by Wertz et al. [1984] where uncertainties concerning issues such as the ideal family size as well as the effects of caring for affected children reported before genetic counseling were considered to be good predictors for reproductive uncertainty after genetic counseling. Because these factors lie outside the scope of genetic counseling, however, it was argued that for many clients reproductive uncertainty could not be eliminated by improving counseling techniques.

Mean Reproductive Outcome After Genetic Counseling

Although 65% of the recontacted females at risk for DMD who were childless before genetic counseling do not have children yet, the mean number of children (n = 0.47) considering all recontacted women (N = 263) is significantly ($P < 0.01$) less than the one observed among the general female population from São Paulo of comparable age and socio–educational level (1.16) [IBGE, 1996]. Postponing (or even refraining from) reproduction might be related to the impact of experiencing (through brother, uncle, or nephew) that DMD is hereditary and lethal, to the effect of genetic counseling which is continuously rediscussed and reinforced by the staff from ABDIM for a great number of these counselees as well as due to the contact and life experience with other affected families. However, the question of whether these women are refraining from or just postponing reproduction remains to be answered in a future study.

Genetic Risk Magnitude

Reproductive decisions after genetic counseling are complex and sometimes unexpected. In the present study, no significant difference was found between lower (<25%) and higher (>25%) risk groups regarding reproductive outcome after genetic counseling, proportion of counselees requesting DNA tests, and desire for (further) children. This is also true for women with good comprehension and retention of the information

TABLE IV. Personal Experience With DMD and Reproductive Decisions*

	Yes (%)	No (%)	*N*	*P*
Sister of affected DMD?	Had children after genetic counseling?			
Yes	35	65	168	ns
No (aunt, cousin, niece)	33	67	95	
	Requested DNA test?			
Yes	26	74	168	ns
No	32	68	95	
	Against abortion?			
Yes	34	66	77	ns
No	42	58	36	
Has only one DMD affected relative?	Had children after genetic counseling?			
Yes	39	61	137	ns
No (has 2–13)	29	71	126	
	Requested DNA test?			
Yes	21	79	137	<0.05
No	36	64	126	
	Good comprehension of genetic counseling?			
Yes	61	39	72	0.05
No	76	24	82	
	Wants (more) children?			
Yes	67	33	42	ns
No	67	33	48	
Has no deceased DMD relatives?	Had children after genetic counseling?			
Yes	38	62	154	ns
No (has 1–11)	29	71	109	
	Requested DNA test?			
Yes	20	80	154	<0.01
No	39	61	109	
	Good comprehension of genetic counseling?			
Yes	59	41	78	0.01
No	79	21	76	
	Wants (more) children?			
Yes	67	33	42	ns
No	67	33	48	
	Against abortion?			
Yes	45	55	51	ns
No	29	71	62	

*N, cases with complete information; ns, not significant.

given to them during counseling. These apparent contradictions were also observed by us [Eggers and Zatz, 1998] in a study on the effects of genetic counseling for adult males affected with progressive muscular dystrophies (who had different recurrence risks for their offspring) as well as by other authors for different genetic diseases [Evers-Kiebooms and van der Berghe, 1979; Frets et al., 1990a; Frets et al., 1990b; Sorenson et al., 1987; Sorenson et al., 1981]. The lack of association between the magnitude of the genetic risk and the reproductive outcome after genetic counseling could be due to differences in the risk perception and complex decision processes prior to reproduction unconscious to a great extent [Frets et al., 1991; Kessler, 1989].

The fact that the intermediate risk group (10–24%) analyzed in the present study requested significantly more DNA tests than the other groups might represent an ascertainment bias since significantly more females in this risk group had a higher socio–educational status. In order to assess this possibility all analyses were repeated excluding this group. Interestingly, all previous associations remained unchanged (data not shown) with two exceptions: a) counselees belonging to lower socio–educational levels reproduced significantly more ($P < 0.01$) and b) counselees with at least one deceased DMD relative reproduced significantly less after genetic counseling ($P < 0.05$). The first observation is not surprising because in all parts of the world lower socio–educational level correlates with higher reproductive rates [Cleland, 1990; Hill, 1990]. However, the fact that counselees reduce reproduction after the death of a DMD relative was apparently not reported previously.

Comprehension of Issues Discussed During Genetic Counseling

As expected and already reported, comprehension of different genetic counseling issues was significantly better among counselees from a higher socio–educational level [Rona et al., 1994], but also among those with more than one affected or at least one DMD relative already deceased. In a previous study [Zatz, 1983], including mothers and sisters of DMD patients, it was observed that not only the reproductive outcome after genetic counseling was lower but also the compre-

hension of genetic risks was better among mothers than sisters. This tells us that the responsibility and the suffering of a mother raising a DMD child have a stronger influence than living with an affected brother. These observations suggest that "learning from life" may have a greater impact than learning during genetic counseling.

Requests for DNA Tests

As expected, DNA tests were more frequently requested by counselees from higher socio-educational levels and with more than one DMD relative, which suggests that the impact of the disease is greater for reproductive decisions if the counselee has already experienced its recurrence. However, the observation that DNA tests are requested more often in cases where an affected relative is already deceased might be related to a higher awareness of the severity and lethality of DMD, or be caused by a feeling of guilt that would deter them emotionally to abort a fetus carrying the same disorder of a dear relative. For example, one young DMD sister who knew she was a carrier wanted to be sterilized when her brother was dying, stating that she would never interrupt a pregnancy. Three years later she returned pregnant to our service requesting prenatal diagnosis and stated she would perform selective abortion in the case of an affected fetus.

In conformity with former results on the lack of association of high risks for different genetic diseases and low reproductive outcomes [Eggers and Zatz, 1998; Evers-Kiebooms and van der Berghe, 1979; Frets et al., 1990a; Frets et al., 1990b; Sorenson et al., 1987; Sorenson et al., 1987], DMD sisters did not show a significant lower reproductive outcome or requested DNA tests more often than aunts, cousins, or nieces despite their higher genetic risks. One possible explanation is that they are too young to realize how difficult it is to raise a child with a lethal degenerative disorder. On the other hand, the need to fulfill motherhood at least once may be greater than the fear to have an affected child, as illustrated in the following examples. Two very young, at-risk women were opposed to prenatal diagnosis when they first became pregnant. One of them had an affected child but the other had a normal son. Five years later they returned to our service asking for prenatal diagnosis for their second pregnancy. Both stated now that they would interrupt their gestation in the case of an affected fetus, which suggests that even without having a DMD child motherhood itself is an experience that may change former opinions or decisions.

Subjective Opinion About Selective Abortion

Although geneticists are fighting to legalize abortion in cases of severe genetic and congenital diseases, abortion is still considered illegal in Brazil except in cases of rape and threat to the mother's life (The current law dates from the 1940s.) However, abortion is a frequent option throughout all Brazilian social classes as a consequence of several related factors such as lack of knowledge on how to use contraceptive methods or inability to afford them, high adolescent pregnancy rate, promiscuity, or great offer of low-quality abortion services (resulting in frequent abortion deaths). It is also important to point out that the public health care system is extremely inefficient and there is no support for handicapped persons and their families. Thus, this social situation sometimes forces them to take options difficult to understand in any developed country.

As expected, the requests for DNA tests for DMD are significantly associated with the individual acceptance of selective abortion. Interestingly, although forbidden by law more than 80% of the general Brazilian population and about 70% of the Brazilian Catholics stated in a recent survey that they would favor abortion in case of life risk for the mother or the fetus. This is concordant with our experience because only 36% of our counselees are against it. Even so, some of these counselees do seek DNA carrier detection tests, arguing that in the case of a positive result, they would refrain from procreation. These observations suggest that individuals are separating issues such as family planning from religious beliefs.

Reproductive Decisions

Because deciding about reproduction is a complex issue in DMD families, it should be kept in mind that (a) the sense of responsibility toward an affected boy is different for mothers than for sisters; (b) that particularly in cases where the affected brother died recently, the sister's willingness to have a child equally affected could be interpreted as a wish to "replace" the brother and/or what he meant for her; (c) the possibility to know the carrier status in advance brings in a different aspect of responsibility in the case of an affected child being born; and (d) because of this knowledge an affected child would have the right to blame his parents for his disease, particularly if no effective treatment will be found in the next couple of years.

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