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Tay-Sachs Screening: Social and Psychological Impact

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Mass screening of healthy persons for genetic conditions has been sometimes accompanied by adverse social consequences [1]. These are due to the participants' lack of information, as well as to such organizational defects as inadequate prescreening education and failure in counseling, follow-up, and treatment. Fears of such unhappy results were sufficient to cause one community not to undertake mass Tay-Sachs screening [2].

The conditions which engender such fears should be studied so as to eliminate them. Preventive medicine cannot be effective in the face of public ignorance, and if in the course of educating potential beneficiaries, some are made anxious, steps should be taken to reduce the unease rather than not provide the service. It was to observe the presence and to weigh the consequences of such hazards in the Baltimore-Washington Tay-Sachs screening program that this study was undertaken.

MATERIALS AND METHODS

The populations and the conditions for data collection have been described previously [3]. Participants in Tay-Sachs screening programs were asked questions designed to assess the personal impact of the screening experience and to test their sense of fulfillment of the program's promise.

Two populations were studied. One (retrospective) consisted of 128 carriers and their noncarrier spouses, 109 noncarrier couples, 52 persons whose initial test results were inconclusive and their spouses, and 57 men whose wives were not tested because of pregnancy. All of these were tested in mass screening sessions. The second group (prospective) consisted of 75 persons tested at the Johns Hopkins Hospital; of these, three were carriers.

RESULTS

Attitudes Toward Self

Responses of carrier couples to test results. The respondents were asked how they felt on learning the results of the test. All of the noncarriers expressed

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TABLE 1

IMPACT OF TEST RESULTS ON FEELINGS OF PARTICIPANTS

	Upset*	Not Upset	Other or Do Not Know	Total
Carriers	64 (48.8)	63	4	131
Carrier spouses	55 (42.0)	70	6	131
Noncarriers	0	438	10	448
Total	119 (16.6)	571	20	710

* Nos. in parentheses = percentages.

relief, but just under one-half of the carriers and about two-fifths of their spouses reported a sense of anxiety upon learning the news (table 1).

The impact on the partners of a mating depended upon which one was the carrier. Both partners were likely to be upset when the husband was the carrier, but when the wife was the carrier, she was more likely to suffer alone (table 2). Carriers and spouses under 30 were more likely to be upset ($P < .05$), but there was no relationship to education, the number of present or desired children, attitudes toward premarital testing, or whether or not they had discussed the carrier state with their sibs.

TABLE 2

CONCERN EXPRESSED BY COUPLES DEPENDING UPON WHICH WAS THE CARRIER

	No. COUPLES*		
	Husband Carrier	Wife Carrier	Total
Both upset	18 (34.0)	13 (16.6)	31
Husband upset/wife not	9 (17.0)	15 (19.2)	24
Wife upset/husband not	9 (17.0)	25 (32.1)	34
Neither upset	17 (32.0)	25 (32.1)	42
Total	53 (100.0)	78 (100.0)	131

* Nos. in parentheses = percentages.

Four kinds of concern were expressed by the carriers and their spouses; some manifested more than one. (1) About half of the carriers expressed a sense of shocked surprise at having been "singled out" or at having been found to have some imperfection. (2) Carriers and spouses were equally dismayed by the realization that children might inherit the gene. (3) Anxiety was displayed by those ignorant of the meaning of the carrier state, but this unease was soon allayed by instruction. (4) The most threatened were women who were pregnant at the time of testing or men whose wives were pregnant. This distress, too, was quickly eased

when it was learned that only one partner was a carrier or when it was made clear that the baby could not be harmed.

Responses of inconclusives. The partners of inconclusive couples were also asked how they felt about their indeterminate status or that of their spouse. Of the 52 persons with initially inconclusive results, 33 (or 61%) were upset; of the 52 spouses, only two were upset. Here the anxiety, a consequence of uncertainty, was resolved by the results of the second and definitive test, whichever its direction.

Personal reactions to screening. All of the participants were asked if they were glad or sorry they were tested (table 3). One couple said they were sorry they had been tested, and 18 were ambivalent or indifferent of which five were carriers and five were carrier spouses.

When asked why they were glad, the noncarriers expressed relief and peace of mind. The carriers were relieved too, but in the knowledge that they would not have a Tay-Sachs baby and that their children could be tested.

When asked if and how their lives changed, 77 (10%) said yes (table 3), suggesting that the screening had relieved them of reproductive constraints. When asked if they would be willing to do it all over again, only 6 (1%) would not (table 3).

Attitudes to Family and Others

Carrier couples were asked what effects the testing had had on family attitudes (table 3). It is evident that exposure to the test posed no threat to the integrity of the family.

TABLE 3

INFLUENCE OF SCREENING TEST RESULTS ON ATTITUDES

QUESTION	Yes	No	Do Not Know	Total
	CARRIERS ONLY			
Change attitude toward each other?	0	262	0	262
Had adverse effect on sex life?	2	259	1	262
Change attitude on birth control?	2	258	2	262
Influenced family planning?	0	262	0	262
	ALL PARTICIPANTS			
Are you glad you were tested?	690	2	18	710
Life changed?	77	626	7	710
Would you be screened again?	700	6	4	710

The carrier couples were also asked whether it would have made any difference if they had known of their carrier condition or that of their spouse before marriage. About one-fourth of the carriers said yes, but only 6% of the spouses. Most

of those who said yes suggested that the uncertainty could have been resolved by testing the other partner before marriage. One said he would not have married, one would have adopted a child, and one would have had no children. When asked if they would have insisted on their partner being tested before marriage, 80% said yes and a further 14% would have "recommended" that the partner be tested.

The carrier couples were asked whether they thought it important to keep the knowledge of the carrier state a secret. Ninety-four percent thought it unnecessary to keep it a secret and were prepared to discuss it freely with anyone.

Carrier couples were asked whether they had discussed the carrier condition with parents, first cousins, aunts and uncles, or friends (table 4). The carriers and their spouses were equally likely to discuss the matter with their parents and friends, but the carriers were more likely than their noncarrier spouses to discuss the matter with cousins ($P < .01$) and uncles and aunts ($P < .05$).

TABLE 4

WITH WHOM WAS CARRIER STATE DISCUSSED?

Persons	Yes	No	No Response/ Do Not Know	Total
Parents	219 (83.6)	29 (11.1)	14 (5.3)	262 (100.0)
First cousins	132 (50.3)	117 (44.7)	13 (5.0)	262 (100.0)
Aunts, uncles	120 (45.9)	132 (50.3)	10 (3.8)	262 (100.0)
Others	187 (71.4)	64 (24.4)	11 (4.2)	262 (100.0)

NOTE.—Nos. in parentheses = percentages.

All of the participants were asked whether or not they had discussed Tay-Sachs disease with their sibs; approximately 80% of the carriers did so and so did 64% of their spouses and 55% of the noncarriers. Furthermore, those carriers who knew the correct probability for a sib to be a carrier were significantly more likely to communicate with brothers and sisters.

All of the participants were asked if they thought Tay-Sachs disease is something to discuss with their children. Only two carriers and two carrier spouses said no.

The carrier couples were asked whether they thought their children should be tested. Only one said no. When asked at what time of life they thought the test should be done, 90% of the respondents recommended premarital testing, one-half of these suggesting an age before the child is old enough even to contemplate marriage. Only 3% recommended that the testing be done after marriage, and only 7% suggested leaving the matter of timing to a physician.

Attitudes Toward Screening

Satisfaction with the screening program. In answer to being asked how they had been notified of the results of the test, 92% said they were notified by letter, 5% (mainly carriers) by telephone, and a few could not remember. When asked if they

were satisfied with the means of notification, over 90% of the respondents said yes, but 12% of the carriers and their spouses indicated dissatisfaction compared to only 4% of the noncarriers ($P < .001$). When asked the sources of the dissatisfaction, some replied that a letter was too impersonal, and others thought the interval between test and notification was too long. The dissatisfied participants were evidently scattered throughout the couples since both members of about 65% of the couples were satisfied, and both were dissatisfied in only 1%.

The carriers were asked whether they preferred to be notified by letter or by telephone. About three-quarters did not care, but the rest wanted to be notified by telephone or by some other personal means. Some suggested that when one member of a couple was found to be a carrier and the other not, both should be notified simultaneously.

Ninety-four percent of the participants said that the program had lived up to their expectations. The reasons given by those who said it did not were varied, but were principally technical and consisted of those things which can and do go wrong in any large exercise.

When asked if sufficient information about the program's purposes and about Tay-Sachs disease had been provided, approximately 75% responded affirmatively. Of the remaining 25%, four-fifths said that the information about the disease had been insufficient.

When asked what changes they would suggest for further testing, 40% of the participants had some suggestions; among these, approximately one-half favored additional education and one-half procedural changes.

Finally, when asked for any comments they cared to make about the whole program, 151 (23%) of the participants had something to say. These remarks offered nothing which had not been said already; among those who were dissatisfied, the carriers and their spouses were overrepresented ($P < .01$).

General attitudes toward screening for Tay-Sachs and other diseases. When asked whether they believed that most people think screening for Tay-Sachs carriers is a good idea, 91.8% of the respondents said yes. A majority of the negative responses came from carriers or spouses ($P < .01$). When asked whether screening for other diseases was, in their opinion, a good idea, all but one said yes. When asked whether they would participate in screening programs for other disorders, five said they would not and 87 (12%) said they would consider it carefully first. Carriers were overrepresented in this latter group.

The carrier couples were asked if they would be willing to participate in research studies of various kinds entailing, for example: (1) blood tests: only 4.3% indicated they would not; (2) tests of mental abilities: 13.7% were reluctant; (3) interview studies: 90% were amenable to this. When the carrier couples were asked why they would be willing to participate in such investigations, more than half gave "social" reasons, that is, reasons reflecting some responsibility to help in the discovery of knowledge which might be put to good use. A further one-third gave personal reasons having to do with the probability that the investigation would lead to something useful for themselves. The carriers were more likely to cite personal

reasons than social ones in contrast to their spouses who were more inclined to cite the latter.

When all respondents were asked if they would object to being approached for such studies, 5% said they would object, and about 11% thought that such an approach would represent an invasion of their privacy. When all participants were asked whether or not they believed that all Jews should be screened, only 10.4% said no, but an additional 21% qualified their responses (table 5). The differences in opinions between carrier couples and the noncarrier groups shown in the table are significant ($P < .001$).

TABLE 5

QUALIFICATIONS ON REASONS FOR TESTING

	Only Those Who Choose to Be Tested	Restrict Testing to Child-Bearing Years	Only Ashkenazi Jews	Others	Total
Carriers	11	16	2	1	30
Carrier spouses	13	20	1	3	37
Noncarriers	9	9	18	5	41
Inconclusives	4	5	17	4	30
Husbands	2	1	8	2	13
Total	39	51	46	15	151

Finally, the prospective participants were asked if they thought there ought to be laws requiring screening for Tay-Sachs disease or for other disorders. Their answers are given in table 6.

TABLE 6

SHOULD THERE BE LAWS REQUIRING SCREENING?

	Yes	No	Ambivalent or Do Not Know	Total
For Tay-Sachs	46 (44.2)	45 (43.3)	13 (12.5)	104 (100.0)
For other diseases	52 (50.0)	37 (35.6)	15 (14.4)	104 (100.0)

NOTE.—Nos. in parentheses = percentages.

DISCUSSION

All but a few of the participants said that the program had lived up to their expectations. The vast majority thought that all Jews should be tested, and the enthusiasm of the prospective subjects was such that half thought that screening

for Tay-Sachs disease and other conditions should be required by law. In addition, nearly everyone was glad to have been tested, would do it again, and would participate in other such programs. Personal and family life was not compromised; as for having more children, many expressed a sense of relief from the constraints of fear. Further, the carrier state is not regarded as a stigma, since almost everyone had discussed the matter freely with family and friends. In addition, the carriers saw the implication for their relatives since nearly all of them discussed the issues with parents, sibs, and collaterals. Those carriers whose knowledge of genetics was greatest were the most likely to do so, but 55% of the noncarriers also discussed the issues of screening with their sibs. Finally, while one-fourth of the subjects responded to an invitation to make suggestions, none pointed to any intractable defects in the organization of the screening.

This apparent success and satisfaction should not distract our attention from the unease expressed by some participants or from some unexpected attitudes to screening. A little less than half of the carriers and their spouses expressed some degree of shock, surprise, anger, or anxiety upon learning that one or other of them was a carrier. This unease, soon quenched in some, abated more slowly in others, but was insupportable in none. At the same time it is clear that while this anxiety seems simply to have heightened the awareness of many, it left others with a sense of caution about screening since they are overrepresented among those whose answers to many of the questions were qualified in various ways and among those who made suggestions for changes. On the other hand, since all but a handful of these same people expressed a sense of benefit in being tested, they seem not to think that any permanent harm was done.

Why should these people have been so concerned? The reasons for those whose results were inconclusive, who were pregnant, or who learned they were carriers before knowing the status of their spouses, are clear enough, but what about those who were surprised at having been singled out or hurt to learn of some imperfection in themselves and those who worried about their children. In both of these latter groups, it is possible that lack of information played a part in engendering the feelings. First, the carrier state is not actually much of a blemish and carriers might feel less singled out and less worried about their children's heritage if they knew that all of us were heterozygous for some lethal genes. Secondly, a better comprehension of the odds might have softened the blow.

The nearly unanimous support for premarital testing and the belief of half of the prospective subjects that screening for Tay-Sachs and other conditions should be required by law were unexpected views which need investigation. Apart from the inequity of laws requiring selective testing of one segment of society, there is the undesirability of any law at all for such a thing. As for the premarital testing, how is such information to be used? The inference is that as a result some matings might not occur, but whether or not mating according to genotype is desirable or even possible is unknown.

An impression which has been substantiated in this and other studies is that

many physicians are not yet aware of the possibilities of genetic screening [3–5]. If they could be taught to see it as a part of their daily routine, it might be that the hazards of mass screening could be resolved by lodging the responsibility for testing with physicians offering primary medical care. For example, the unease expressed by the carriers and their spouses might be reduced if the testing could be done under the supervision of a trusted medical advisor who is well known to the individual being screened, fully sympathetic to the aims of preventive medicine, and knowledgeable in genetics. In the mass screening context, no single individual is responsible for all aspects of the service, and there is opportunity for irregularities no matter how careful the arrangements or how attentive the personnel [1]. In contrast, in the traditional setting of the doctor's office, the physician might supply the information which prompts the patient to be tested, take the blood sample and send it to a state laboratory, notify the subjects in ways appropriate to the results, and provide counseling. If these conditions were to prevail, the impact of testing might be reduced to that of any other routine medical procedure.

Evaluations of screening programs are necessary to assure ourselves that screening is harmless, but there is another important reason for doing them. As the relationship of genetic knowledge to medicine has become apparent to the public, the application of such knowledge in some contexts has been attacked as unethical. No one can deny that transgressions have occurred, but many choices will have to be made on ethical grounds in the future. If we could approach those choices with the knowledge of what an informed public finds acceptable and desirable, options would be open which ignorance would deny. It is reassuring, for example, to know that the participants in this study see no reason to keep their carrier state a secret; that is, they did not feel stigmatized. This is not to say that confidentiality should be breached—the information belongs to the carrier who may reveal it if he wishes—but that a sense of labeling on the part of the participant cannot be advanced as a reason for not doing this particular test. Thus, the results of this investigation suggest that it will be only through careful evaluation of current practices that we will see whether genetic screening can be employed, ethically and harmlessly, to advance the aims of preventive medicine.

SUMMARY

Participants in two Tay-Sachs screening programs were generally satisfied with the organization of the tests and the results. There was no evidence of adverse impact on reproductive plans or interpersonal relations, and the respondents professed to believe in the value of screening. While the carriers discussed their condition freely with others and were no less favorable to the idea of screening than the noncarriers, about one-half of their number expressed discomfort in being told they were heterozygotes. These feelings were allayed by counseling, but there was evidence of some residual unease. It is suggested that this anxiety would be less prominent and more easily reduced if screening were done under conditions of ordinary primary medical care rather than outside the conventional system.

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