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TAY-SACHS DISEASE: TO SCREEN OR NOT TO SCREEN

To a Jewish family awaiting a physician's health report on their newborn there are no words more devastating than "your child has Tay-Sachs[1] disease." Because this congenital disorder[2] is fatal in the early years of a child's life coupled with the statistical studies showing that it strikes primarily Jewish families of East European heritage an increasing awareness and concern has been voiced[3] by the Jewish community with regard to the detection and cure of this genetic disease. Lately,[4] the medical and halakhic aspects of screening large segments of the Jewish population for the carrier state of Tay-Sachs disease and the performance of amniocentesis for the pre-natal detection have been discussed and debated. This essay will deal with the moral, ethical, social, psychological and religious indications and contraindications to amniocentesis for the pre-natal diagnosis of Tay-Sachs disease.

I

Tay-Sachs disease is characterized by weakness beginning at about six months of age, progressive mental and motor deterioration, blindness, paralysis, dementia, seizures and death usually by three years of age. It has been estimated that 1 in 30 Ashkenazi Jews[5] and 1 in 300 non-Jews is a carrier of the Tay-Sachs gene. If a Jew (1 in 30 risk) marries another Jew (1 in 30 risk), the chance that both husband and wife are carriers is 1 in 900. Therefore 1 in 900 Jewish couples is at risk for having children

with Tay-Sachs disease. For such a couple, each pregnancy has a 25 percent chance of producing a child with this disease. Hence, the incidence of Tay-Sachs disease in the Jewish population (assuming there is no inter-marriage with non-Jews) is 1 in 3600 births.[6]

Intrauterine diagnosis of Tay-Sachs in an unborn fetus is now possible by a procedure called transabdominal amniocentesis.[7] After a couple have been identified as Tay-Sachs carriers through simple blood tests, amniocentesis allows a "monitored" pregnancy of the couple so that beginning with the fourth month of a pregnancy a small amount of fluid within the mother's womb can be carefully withdrawn. The fetal cells of this fluid are grown in a laboratory and tested for the presence or absence of hexosaminidase A, the lack of which is the cause of Tay-Sachs.

The majority of the results indicate that the fetus is normal. This, of course, presents no halakhic difficulties. But what if the test reveals the fetus has the disease and that the child is doomed to die? What is done? Usually the parents are advised of their options, one of which is a therapeutic abortion. Orthodox Judaism is opposed to this solution; it does not favor eliminating Tay-Sachs disease by "selective termination of affected pregnancies" even though Reform and Conservative rabbis might sanction such a procedure. As to rabbinical support for Tay-Sachs screening programs, such support is usually limited to detecting the carrier state and does not include the performance of amniocentesis with the sole aim of abortion, if the fetus is found to have Tay-Sachs disease.

II

There are reasons other than religious ones why we should move cautiously before undertaking mass screening programs.[8] A group of physicians[9] specifically discouraged unmarried people from being tested because of the possible social and psychological problems that carrier identification might create. They are:

> Should a known carrier refuse to marry a mate who has not been tested? Should two carriers break up an engagement or a marriage

if they learn they are both carriers as a result of a screening program? Should a young person inquire about the Tay-Sachs status of a member of the opposite sex prior to dating?

When does a person who knows he is a carrier inform his fiancee?

Should one sacrifice primary prevention of Tay-Sachs disease by mate selection to avoid psychosocial consequences?

Is this method of disease prevention the most attractive aspect of genetic screening for recessives?

As a result of arguments advanced by a leading physician involved in studying Tay-Sachs disease, Dr. Kuhr,[10] an advisory committee of doctors in Dayton, Ohio decided against organizing a mass screening program. The major reason given was the potential psychic burden on those young people discovered to be heterozygotes.[11] Dr. Kuhr further pointed out a study of the behavior of physicians and clients in a voluntary program of testing for the Tay-Sachs gene in which mention is made of a 15 percent anxiety reaction to the discovery of the carrier state.[11a]

One must remember that 29 of 30 people tested for the carrier state are found to be free of the Tay-Sachs gene. It is certainly desirable for these 29 of each 30 tested to have peace of mind. Is the anxiety of the 30th person in learning he or she is a carrier sufficiently great to warrant not testing at all? Obviously not! One cannot, however, minimize the possible psychosocial trauma to such an individual.

The social stigma of being a carrier of the Tay-Sachs gene is not fully appreciated. Misinformed people may look at carriers in the same manner as patients with epilepsy and leprosy were looked at half a century ago, i.e., as individuals afflicted with a "taboo" disease, to be shunned and ostracized from normal social contact. If the experience of sickle cell screening will be repeated then discrimination against carriers of Tay-Sachs may also occur in a variety of areas. Individuals found to have sickle cell trait have been dismissed from their jobs, refused employment, and charged higher premiums or rejected by insurance companies when seeking life, accident, and health insurance. The United States Air Force does not train black recruits with sickle cell trait to become pilots. Job seeking is thus made more difficult for those people who possess the sickle cell trait.

Is this fate also to be suffered by people who, after screening, are found to be carriers of the Tay-Sachs gene? Total confidentiality in screening might avoid such problems and should be an essential part of all Tay-Sachs screening programs.

The selection of target groups to be screened for the Tay-Sachs gene, the planning, organization and implementation of such screening events, the educational activities that must precede screening, the laboratory aspects of the testing, and the referral of people for counseling are beyond the scope of this essay. Suffice it to say that compliance with screening for Tay-Sachs disease is dependent not only on the motivation of the client, his or her perception of the susceptibility to and seriousness of the inherited condition, the possible consequences of noncompliance and the potential benefits from participation, but also on the moral, ethical and religious background of the Jewish people who are the clients at risk.

If the purpose of Tay-Sachs screening is to provide eligible clients with genetic counseling about reproductive and mating options, few would argue against screening. If the purpose, however, is to introduce couples at risk to the benefits of pre-natal diagnosis by amniocentesis with the specific intent of recommending abortion of affected fetuses, then screening should best not be performed. Jewish religious teachings must be considered if cooperation from the rabbinate and compliance from the clients are to be obtained in any screening program. A plan for the screening of heterozygotes for Tay-Sachs disease in Israel[12] specifically includes the rabbinate, although that portion of the plan which dictates abortion for homozygote fetuses is obviously not acceptable to either of the Chief Rabbis.

III

Some bitter lessons were learned from the laws passed in regard to sickle cell screening. Georgia has a law which requires sickle cell screening of all newborns unless the parents object on religious grounds. In California, all black people admitted to hospitals must be screened by law. In Illinois, mandatory premarital screening for sickle cell trait was enacted "if the phy-

sician indicates that it is needed." In New York, mandatory pre-marital testing only of blacks is required. Many other states passed mandatory screening laws but few, if any, enforced them.

Most of the above laws were challenged on constitutional grounds. Obligatory screening of any age group for any genetic disease seems to be an unconstitutional invasion on the rights of the individual. As a result, many of the laws pertaining to sickle cell screening have been or are being repealed or amended. In some states, such as Virginia and Kentucky, pre-marital screening is now voluntary, the mandatory provision of the original law having been amended.

In the famous Jacobsen decision in 1905, the United States Supreme Court upheld the law of mandatory smallpox vaccination on the basis of the needs of society to protect its citizens from smallpox. However, neither sickle cell disease nor Tay-Sachs disease are contagious illnesses and, therefore, do not constitute dangers to others. Although there may be a financial burden on society to care for such patients, mandatory screening laws appear to be unconstitutional when weighed against the right of individual privacy.

Discriminatory screening along racial or ethnic lines is also unconstitutional. To pass a law mandating sickle cell screening only for blacks or Tay-Sachs screening only for Jews is clearly discriminatory and under-inclusive. Screening must be offered to all, although not all ethnic or racial groups need participate in the screening if they so choose.

If any law is to be passed in regard to Tay-Sachs disease, it must indicate that screening is completely voluntary and that the results will remain confidential. Not only is the preservation of confidentiality an essential component of the doctor-patient relationship and a patient's constitutional right, but a repetition of discriminatory practices which occurred in the sickle cell screening experience must be avoided. Finally, any proposed law concerning Tay-Sachs screening should NOT require abortion, sterilization or prohibition of marriage but must preserve the fundamental rights of marriage or procreation.

IV

Rabbi Moses Feinstein was asked about the advisability of screening for Tay-Sachs disease, and, if proper, at what age should the test be performed. He was further asked whether screening should be performed as part of a publicized screening program or only as a private test. His written responsum of 1973[13] states:

> . . . it is advisable for one preparing to be married, to have himself tested. It is also proper to publicize the fact, via newspapers and other media, that such a test is available. It is clear and certain that absolute secrecy must be maintained to prevent anyone from learning the result of such a test performed on another. The physician must not reveal these results to anyone . . . these tests must be performed in private, and, consequently, it is not proper to schedule these tests in large groups as, for example, in Yeshivas, schools, or other similar situations . . .

Rabbi Feinstein also points out that most young people are quite sensitive to nervous tension or psychological stress and, therefore, young men (below age 20) or women (below age 18) not yet contemplating marriage should not be screened for Tay-Sachs disease. Finally, Rabbi Feinstein holds that abortion of a defective fetus after amniocentesis is prohibited.

Rabbi J. David Bleich, writing in *TRADITION*,[14] indicates that the elimination of Tay-Sachs disease is, of course, a goal to which all concerned individuals subscribe. The Jewish legal ramifications of the testing program appear in another article by Rabbi Bleich in the summer 1972 issue of the Hebrew periodical *Or ha-Mizrach*. He points out that

> . . . the obligation with regard to procreation is not suspended simply because of the statistical probability that some children of the union may be deformed or abnormal. While the couple may quite properly be counseled with regard to the risks of having a Tay-Sachs child, it should be stressed that failure to bear natural children is not a *halakhically* (Jewish legal) viable alternative.
>
> Of at least equal if not graver concern is the proposal that fetal monitoring be performed with a view toward termination of the pregnancy if the fetus be identified as a victim of Tay-Sachs disease.

> The fear that a child may be born physically malformed or mentally deficient does not in itself justify recourse to abortion . . . Since the sole available medical remedy following diagnosis of severe genetic defects is abortion of the fetus, which is not sanctioned by Halakhah (Jewish law) in such instances, amniocentesis, under these conditions, does not serve as an aid in treatment of the patient and is not *halakhically* permissible . . .

Rabbi Bleich concludes that screening programs for the detection of carriers of Tay-Sachs disease "are certainly to be encouraged." He suggests that the most propitious time for such screening is childhood or early adolescence, since early awareness of a carrier state, particularly as part of a mass screening program, is advantageous.

At its 70th anniversary biennial convention during the Thanksgiving weekend of 1974, the Union of Orthodox Jewish Congregations of America adopted a resolution concerning Tay-Sachs screening which essentially echoes the opinion of Rabbi Bleich. The Union suggests that

> the Orthodox community can extend support to programs of genetic screening *only* when competent Halakhic guidance is provided for all participants.

The Union called upon its constituent synagogues to work for such programs in every Jewish community but emphasized that all Tay-Sachs screening programs must be accompanied by adequate and competent rabbinic counseling.

The Association of Orthodox Jewish Scientists issued the following statement in 1973 outlining its position in regard to Tay-Sachs screening:

> We endorse voluntary screening of young adults of an age in which marriage has become a serious consideration but before definite marital commitments have been made. The screening of younger individuals, years before marriage, yields no immediate benefits and might result in a longer period of anxiety in carriers than is warranted. We feel that all screening must be linked to both genetic and religious personal counseling. Emotionally immature individuals may be traumatized psychologically if they learn of their carrier state, and these individuals must be provided with the opportunity for additional professional psychological support. Genetic counseling must be in consonance with Torah principles.

> The Association is unalterably opposed to amniocentesis, whose natural and logical consequence is abortion. It likewise feels that screening of married couples or those whose marriage is imminent and who are not committed to disruption of their mutual marital commitments, were both partners to be discovered to be Tay-Sachs carriers, is unwise, again because virtually the only consequences would be abortion, or a childless marriage. We are also concerned that any program be absolutely voluntary, and that the nature of any educatonal drive be informational rather than coercive. There must also be absolute assurance that the confidentiality of all carriers will be safeguarded.

The various objections to amniocentesis and abortion in Jewish law are predicated on considerations surrounding the fetus. Extreme emotional stress in the mother leading to suicidal intent might constitute one of the situations in which abortion might be sanctioned by even the most Orthodox Rabbi. If a woman who suffered a nervous breakdown following the birth (or death) of a child with Tay-Sachs disease becomes pregnant again, and is so distraught with the knowledge that she may be carrying another child with the fatal disease that she threatens suicide, Jewish law could allow amniocentesis. If this procedure reveals an unaffected fetus, the pregnancy continues to term. If the result of the amniocentesis indicates a homozygous fetus with Tay-Sachs disease, rabbinic consultation should be obtained regarding the decision of whether or not to abort. No general rule of permissiveness or prohibition can be enunciated in regard to abortion in Jewish law. Each case must be individualized and evaluated on the basis of its merits taking into consideration all the prevailing medical, psychological, social and religious circumstances.

V

A research group on ethical, social and legal issues in genetic counseling and genetic engineering of the Institute of Society, Ethics and the Life Sciences, recently proposed a set of principles[15] for guiding the operation of genetic screening programs to focus attention on the problems of stigmatization, confidentiality, breaches of individual rights to privacy, and freedom of choice in childbearing. Some of the principles include

> the need for well planned program objectives, involvement of the communities immediately affected by the screening, absence of compulsion, provision of counseling services, an understanding of the relation of screening to realizable or potential therapies, and well formulated procedures for protecting the rights of individual and family privacy.

This research group pointed out that widespread genetic screening may produce ethical, psychologic and sociomedical problems for which physicians and the public may be unprepared.

In the past several years, screening for sickle cell trait has become very widespread. There are some,[16] however, who believe that

> screening for sickle cell trait *per se* now seems unjustified, except for genetic counseling purposes, which, as many would now agree, should be undertaken only at the specific request of high-risk individuals who are able to grasp the implications. However, even judiciously conducted screening and genetic counseling may still produce psychosocial side effects, which are yet to be fully assessed.

Efforts to educate, test and counsel for the sickle cell gene have shown features that can arouse anxiety, fear and apprehension; generate guilt, resentment and frustration; inhibit the development of personal self-esteem and racial pride; destroy families; and reactivate neglect of the sickle cell problem.[17] There have been no systematic studies to date of these factors in relation to either sickle cell disease or Tay-Sachs disease.

The birth of a child with a serious congenital deformity, or mental deficiency, or a lethal metabolic error, such as Tay-Sachs disease, is a terrible shock to any parent. The personal decisions involved are very difficult: whether to marry; whether to have children; whether to have another child; whether to adopt a child. For the general Jewish population, not only must medical, genetic and psychological factors be considered in any given case, but the religious attitude of Judaism toward such matters as abortion, contraception, amniocentesis, genetic screening, and procreation, to name but a few, must be taken into account. Hence, Rabbinic consultation and advice should be sought concomitant with the medical-genetic counseling. One can inform and educate the Jewish community about Tay-Sachs screening,

but such a program will only truly achieve its goal when the community wants it and is willing to work for it.[18]

It seems desirable to screen a limited population that would be followed closely to observe all the psychological and social implications as well as the genetic accomplishments of the screening. In this way, the scientific and religious approaches might coincide, and any ultimate conclusion would be based on fact instead of speculation. There is no reason to be stampeded by those professional enthusiasts whose interests are founded on mass screening.[19]

NOTES

1. Tay-Sachs is named after a British ophthalmologist Warren Tay who, in 1881, described degeneration of the macular region of the eye in a one-year old child, and an American neurologist, Bernard Sachs, who six years later, published the clinical and pathological findings.

2. The "cherry-red spot" in the macula of the eye is the clinical sign most frequently equated with Tay-Sachs disease.

3. Recently the First International Conference on Tay-Sachs Disease: Screening and Prevention was held.

4. See Carmody, P. J., Rattazzi, M. C., & Davidson, R. G., "Tay Sachs Disease — the Use of Tears for the Detection of Heterozygotes," *New Engl. J. Med.* 289: 1072-1074, 1973; Kaback, M. M., & Zeiger, R. S., "Heterozygote Detection in Tay-Sachs Disease: A Prototype Community Screening Program for the Prevention of Recessive Genetic Disorders," *Adv. Exp. Med. Biol.* 19: 613-632, 1972; O'Brien, J. S., Okada, S., Fillerup, D. L., Veath, M. L., Adornato, B., Brenner, P. H. & Leroy, J. G., "Tay-Sachs Disease: Prenatal Diagnosis," *Science* 172: 61-64, 1971; Navon, R, & Padeh, B., "Prenatal Diagnosis of Tay-Sachs Genotypes," *Brit. Med. J.* 4: 17-20, 1971; Schneck, L. Adachi, M., & Volk, B. W., "The Fetal Aspects of Tay-Sachs Disease," *Pediatrics* 49: 342-351, 1972; Saifer, A., Schneck, L., Perle, G., Valenti, C. & Volk, B. W., "Caveats of Ante-natal Diagnosis of Tay-Sachs Disease," *Amer. J. Obst. Gynec.* 115: 553-555, 1973; Nadler, H. L., "Prenatal Diagnosis of Inborn Defects: A Status Report," *Hosp. Pract.* 10: 41-51, 1975; Kardon, N. B., Chernay, P. R., Hsu, L. Y., Martin, J. L., & Hirschhorn, K., "Pitfalls in Prenatal Diagnosis Resulting from Chromosomal Mosaicism," *J. Ped.* 80: 297-299, 1972; Milunsky, A., "Prenatal Diagnosis of Tay-Sachs Disease," *Lancet* 2: 1442, 1973; Rosner, F. "Amniocentesis in Tay-Sachs Disease," *J.A.M.A.* 228: 829, 1974; Jackson, L. G., Glazerman, L. R., Faust, H. S. & Nimoityn, P., "Screening for Carriers of Tay-Sachs Disease," *J.A.M.A.* 229: 640, 1974; Pearlmutter, F. A., "Tay-Sachs Disease," *J.A.M.A.* 230: 38, 1974.

5. The inheritance of Tay-Sachs disease follows laws of Mendelian genetics. The transmission appears to be autosomal recessive since both parents of patients are clinically normal, sex ratios are equal, and both parents have enzyme levels that are intermediate between those of patients and normal controls. Thus a child who inherits one recessive gene from only one parent is a carrier or has the trait but is clinically completely normal. Only a child who inherits two Tay-Sachs genes, one from each parent, will have the disease. If two carriers marry, there is a 25 percent chance with each pregnancy that the child will have the disease and a 50 percent chance that the child will be a carrier like the parents. There is also a 25 percent chance that the child will be totally free of the disease as well as the carrier state. If a carrier marries a non-carrier, none of the children can have the fatal disease but half the children will be carriers, like the one carrier parent.

6. In view of the high rate of intermarriage in the United States the incidence of Tay-Sachs is probably much less than 1 in 3600. One expert has calculated that 30 children with Tay-Sachs disease are born annually in North America. See O'Brien, J. S. "Tay-Sachs Disease: From Enzyme to Prevention," *Federation Proc.* 32: 191-199 as to whether there is an increase in Tay-Sachs disease. Shaw, R. F., & Smith, A. P., "Is Tay-Sachs Disease Increasing?" *Nature* 224: 1214-1215, 1969, and Myrianthopoulos, N. C., Naylor, A .F., & Aronson, S. M., "Tay-Sachs Disease is Probably Not Increasing," *Nature* 227: 609, 1970.

7. See Schwartz, H., "Amniocentesis in Tay-Sachs Disease," *J.A.M.A.* 231: 1229, 1975; Roberts, E. J., "Amniocentesis in Tay-Sachs Disease," *J.A.M.A.* 231: 1230-1231, 1975; Orleans, J. "Amniocentesis in Tay-Sachs Disease," *J.A.M.A.* 231: 1231, 1975.

8. See Beck, E., Blaichman, S., Scriver, C. R., & Clow, C. L., "Doubtful Benefits of Tay-Sachs Screening," *New Engl. J. Med.* 292: 371, 1975; Schneck, L., Saifer, A., & Volk, B. W., "Benefits of Tay-Sachs Screening," *New Eng. J. Med.* 292: 758, 1975; Jackson, L. G., Nimoityn, P., Faust, H. S., & Glazerman, L. R., "Benefits of Tay-Sachs Screening," *New Eng. J.* 292: 758-759, 1975; Kaback, M. M., "Heterozygote Screening — A Social Challenge," *New Engl. J. Med.* 289: 1090-1091, 1973; Evans, P. "Testing for Tay-Sachs Carriers," *Brit. Med. J.* 3: 408, 1973; Evans, P. "Testing for Tay-Sachs Heterozygotes," *Lancet* 2: 391, 1973; Edwards, J. H., "Testing for Tay-Sachs Heterozygotes," *Lancet* 2: 1143, 1973; Evans, P. R., Ellis, R. B., & Masson, P. K., "Testing for Tay-Sachs Heterozygotes," *Lancet* 2: 1143-1144, 1973.

9. Kaback, M. M., Zeiger, R. S., Reynolds, L. W. et al. "Tay-Sachs Disease: A Model for the Control of Recessive Genetic Disorders," *Proceedings of the Fourth International Conference. Vienna, Austria.* Sept. 2-8, 1973. *Amsterdam. Excerpts Medica,* 1974, pp. 248-262.

10. Kuhr, M. D., "Doubtful Benefits of Tay-Sachs Screening," *New Engl. J. Med.* 292: 371, 1975, and "Tay-Sachs Screening," *New Engl. J. Med.* 292: 1300, 1975.

11. Dr. Kuhr's opinion was disputed, however, by others in the medical community.

11a. Beck, E., Blaichman, S., Scriver, C. R., & Clow, C. L., "Advocacy and Com-

pliance in Genetic Screening. Behavior of Physicians & Clients in a Voluntary Program of Testing for the Tay-Sachs Gene," *New Eng. J. Med.* 291: 1166-1170, 1974.

12. Padeh, B., "A Screening Program for Tay-Sachs Disease in Israel," *Israel J. Med. Sci.* 9: 1330-1334, 1973.

13. Copy kindly provided by Rabbi Feinstein's son-in-law, Rabbi Dr. Moses D. Tendler.

14. 13: 145-148, 1972.

15. Lappe, M., Gustafson, J. M. & Roblin, R., "Ethical & Social Issues in Screening for Genetic Disease," *New Engl. J. Med.* 286: 1129-1132, 1972.

16. Mamman, I., "Screening for Sickling," *Lancet* 1: 1030, 1975.

17. Whitten, C. F., & Fischhoff, J., "Psychosocial Effects of Sickle Cell Disease," *Arch. Int. Med.* 133: 681-689, 1974.

18. Jackson, L. G., "Heterozygote Detection for Autosomal Recessive Genetic Diseases. Community Aspects of the Tay-Sachs Experience," *Clin. Ped.* 13: 307-309, 1974.

19. The following articles are of interest:

Seligmann, J., "Jewish Diseases," *Newsweek.* New York, May 26, 1975, p. 57.

Edelson, E., "The Jewish Disease," *New York Daily News,* Nov. 9, 1971.

Ellis, R. B., Ikonne, J. V., Patrick, A. D., Stephens, R., & Willcox, P., "Prenatal Diagnosis of Tay-Sachs Disease," *Lancet* 2: 1144-1145, 1973.

Rosner, F., "Screening for Tay-Sachs Disease," *Lancet* 1: 359, 1974.

O'Brien, J. S., "Diagnosis of Tay-Sachs," *Nature* 224: 1038, 1969.

Ohman, R., Ekelund, H., & Svennerholm, L., "The Diagnosis of Tay-Sachs Disease," *Acta Pediatr. Scand.* 60: 399-406, 1971.

To further inform the public the following pamphlets have been published: "Tay-Sachs. The Killer is Cornered," National Tay-Sachs & Allied Diseases Assoc. Inc., New York; "What You Should Know About Tay-Sachs Disease and How to Protect Against this Fatal Illness," New York State Dept. of Health; "Tay-Sachs Facts. What Every Family Should Know About This Fatal Childhood Illness," Calif. Tay-Sachs Disease Prevention Program; "Tay-Sachs Disease and Birth Defects Prevention," National Foundation — March of Dimes. White Plains, New York; "Operation Gene Screen," Albert Einstein College of Medicine, New York and National Genetics Foundation.