Ethics an Genetic technology: An attitudinal survey

CLARA E. CARRASCO DE MALDONADO

Submitted in Partial Fulfillment of the Requirements for "Genetics and Society: a Dynamic Interaction" - NSF/AAAS Chautauquatype Short Course-March 1978.

INTRODUCTION

Most of the characteristics we human beings possess may be traced to our genetic make-up. Our anatomy, phisiology, even our behaviour towards ourselves and others in our daily lives, may be traced in one way or another to the information contained in our genes. We, as reproductive members in a sexually reproducing society have our share of the genetic information in our species' gene pool. Genes in this pool have dynamic relations with other alleles and with the sorrounding environment. Gene frecuencies in a given population may change as a result of this interaction. This may lead to the appearance or dissappearance of given genotypes.

In spite of the thousands of paired genes in our species, the

packaged genes in our own chromosomes make unique combinations in each and everyone of us. Going back just five generations in an individual's family tree, and if no inbreeding has taken place, one may have up to 110 ancestors all of whom contribute to his or hers genetic make-up. Thus, we all have our own unique set of genes, or genotype.

In each person's genotype deleterious or harmful genes may be present, just as chromosomal anomalies. Deleterious genes may be associated with specific diseases such as Thalassemia, Sickle Cell, or PKU; or may predispose to disease, as in Juvenile Diabetes which may be set off by environmental factors. Chromosomal anomalies may include translocations, inversions, deletions, or duplications, of parts of a chromosome, as well as chromosomal aneuploidies.

Disease carrying genes and chromosomal abnormalities in an individual's genotype may be passed on to their descendants. In fact, 6-9 % of all births may show some kind of genetic disorder (22). According to Borgaonkar and Shah (3) genetic diseases are formally becoming recognized as a major, but still relatively unmeasured, component of the world's heath deficit.

Genetic screening of potential deleterious genes and/or chromosomal abnormalities carriers, is being pursued in America today. One objective of genetic screening is the discovery of individuals possesing particular genotypes. They may be carriers of genes for sex-linked or autosomal recessive traits, or of autosomal dominants which only express themselves after the reproductive years of the person involved are over. The assumption being made is that awareness of such genes may influence their decision about reproducing themselves.

Many genetic problems may arise in connection with marriage and parenthood. A family may be brought to the attention of a geneticist because of an affected child or relative. The case may be referred by a physician who may have too little experience with genetic disorders to advise the family what to expect from it. The geneticist, who may be an M.D. or a physician's genetics associate, will provide professional advice concerning the disorder or characteristic in question, its frecuency, manifestations, if any, and probabilities of transmision to their offsprings (2;27).

A detailed family history or pedigree should be one of the first steps involved in genetic counseling. According to Sly, (2) and García Castro, (12) this very important step should never be ommited. It will provide information necessary in determining the mode of inheritance of genetic disease in that family's particular circumstances. The pedigree may, in some instances, even provide the diagnosis as in a Hurler vs. Hunter syndrome distinction. Multiple abortions in a mother, for example, may indicate the presence of chromosomal anomalies. One out of every ten women who have had multiple abortions have proven to be carriers of chromosomal anomalies (12).

Some 125 to 150 genetic diseases may be detected at present (12,22). However, the number of these which may be detected in prenatal diagnosis has not been made clear. While Friedman in 1971 (11) listed some 30 disorders which could be identified "in utero" in addition to chromosomal aberrations Holtzmann in 1977 (17) stated that figure around 20. Murray (22) indicated that through amniocentesis some 40 diseases could be identified.

Antenatal diagnosis of metabolic disorders such as Tay-Sachs, Cooley's anemia, Neural-tube defects, etc., and chromosomal aneuploidy disorders (ex. Klinefelters' XXY) are now available. In utero tests to obtain reliable data towards this end, include amniocentesis and ultrasonic sonography.

Amniocentesis, which was described by Friedman in 1971 (11) involves obtaining amniotic fluid samples during the 14th to the 16th week of pregnancy. Fetal cells thus obtained are grown in tissue culture. Tests such as karyotypes, including Q, G, R, and C banding, as well as enzymatic analysis for various metabolic disorders, may be perfomed on these cells. This technique was still considered experimental as late as 1975 since the extent of the risks was not yet fully known (27). However, Murray (22) and García Castro (12) agree in that the risk of damage to the fetus is no higher than in a normal pregnancy. Diagnostic error for this test is as low as 1 %.

Ultrasonic sonography is being extensively used in the United States today since it is less risky than X-ray diagnosis, and the test results are much better. Still, it is not yet recommend as routine

procedure for monitoring pregnancies. It has been especially useful in detection of anencephaly, ectopic pregnancies, neural-tube defects, etc., as early as 11 to 13 weeks into a pregnancy (18). An ultrasound examination involves little discomfort. The sound transmitter, which resembles the head of a stethoscope, is rubbed gently over the abdomen. This scanning, performed at 1-2 cm intervals, produces an image of a cross section of the uterus on a television screen. Each image is photographed. When seen together, the physician has a series of photographs of the wall of the uterus, its contents, and the surrounding tissues. A pregnancy may be detected by this method as early as 4 to 5 weeks after a woman conceives. Ultrasound can also assist a phisician in carrying out amniocentesis. Watching the image on the television screen he can guide the needle into the amniotic fluid thus reducing the danger of striking the fetus or of injuring the placenta.

Once the necessary tests have been performed, professional advice concerning the magnitude, implications and alternatives for dealing with the risk of occurrence of the hereditary disorder in the family should be available. Having this knowledge may increase their ability to make a wise decision and enhances their freedom of choice. Follow-up visits throughout the years should be encouraged to compensate for any possible kind of mistake in counseling, and to enhance succesfull communication between the counselor and the family involved.

Puerto Rico, at present, has only one genetic counseling center. It is the Medical Genetics Laboratory's Regional Medical Program of Hereditary Diseases, located at the Children's Hospital in the Puerto Rico Medical Center. The Program's Director, Dr. José Miguel García Castro is also Pediatrics and Medical Genetics professor at the School of Medicine of the University of Puerto Rico, located in the Medical Center. According to his studies (12) deleterious genes exist in the Island's population in relatively high frecuencies. In fact some abnormalities have been reported for the first time in the medical genetics literature, from puertorrican families (13, 14). This may be due, primarily, to the widespread endogamy which took place during the first three to four centuries of the Island's colonization. Only in the last 100 to 150 years has

the Island's population increased considerably (last census in 1970 showed approximately 3 million people in Puerto Rico's 3500 square miles). It's small size, abrupt terrain, and poor communication facilities, favored inbreeding. Socio-cultural division of classes which existed from its discovery in 1492 until the 1800's, favored consanguineous marriages. Nevertheless, the genetic effects of this inbreeding are not well-known and less well understood by the majority of the people. In general, young adults are poorly informed of any inherited diseases which may exist in their own families. In fact, many families fail to recognize the hereditary nature of any such trait.

The present survey was undertaken to gain insight into the views and opinions of a sample of the puertorrican population regarding genetic diseases.

MATERIALS AND METHODS

The survey was conducted in the Ponce Regional College of the University of Puerto Rico, at the begining of the second semester of the 1977-78 school year, from January through March. The questionnaire, "Ethics and Genetic Technology, An Attitudinal Survey", was translated into Spanish to prevent language difficulties. The translation, "Etica y Tecnología Genética, Investigación Sobre Actitudes", was kept as literal as possible so that comparisons of the resulting data could be drawn with the original English version. An answer sheet was designed, and provided with each copy, to facilitate the pooling of the data. (See Appendix A.)

Distribution of the questionnaires was preceded by a short (30-35 min) talk. This was deemed necessary since the students and most of the professionals involved had never taken a Genetics course. Several presentations took place due to scheduling difficulties which prevented everyone coming together at one time. An effort was made towards impartiality during these talks. The author's opinion concerning one or another of the issues involved was deliberately not given at any time during the survey. The

mode of inheritance, external characterizations, risk of occurrence whenever known (as in Huntington's Chorea = 50 percent), as well as an explanation of genetic screening and counseling, tests such as amniocentesis, karyotiping, and electrophoretic blood analysis (as for Sickle-Cell Anemia) were included in the slides viewed during the presentations. A plea towards honesty when answering, was expressedly made. Everyone was asked to return the answer sheets and the questionnaires within 24 to 48 hours.

RESULTS

The views and opinions of 260 people were obtained in the survey. This included both male and female, first and second year college students, professors, and library staff, ranging in age from 16 thru 55 years, from middle class Puertorrican families. The data was analysed as shown on Tables 1 to 7.

Seventy-one percent of those interviewed agree that screening tests should sometimes be mandatory depending on the situation. An average of 75 percent agree that the tests described on questions 10 thru 18 of section II, should be mandatory (Table I).

As to "Society's" role in determining the *type* or kind of disease to be tested for, there is an almost one to one relation between those who agree, and those who disagree, with a possible deciding factor of 9 percent undecided (question number 19 - Table 1). When male versus female answers are analysed, it is the latter who show a tendency towards allowing "Society" to decide for them (question number 19, Tables 2 and 3). However, 62 percent of both males and females disagree in that "Society" is the one to determine *who* should be screened. It is the individual who should determine whether or not he wishes to be screened, according to almost 60 percent of those interviewed (questions number 20 and 21, Table 1). An average of 90 percent agree that it is essential that any inheritable disease which may exist in a family should be the common knowledge of all those who might be affected, particularly before a marriage takes place (questions 24 and 25, Table 1).

Although almost 80 percent indicated that they would consider abortion and sterilization under special circumstances (Table

7a), there's an almost evenly divided opinion as to the rights of parents versus the rights of the fetus (question number 28, Tables 1, 2, and 3). The opinion is also highly divided among those who feel that it is better to have no life at all if it is not of a reasonable quality, and those who either disagree or are undecided (question number 29, Table 1). Here, it is interesting to note, from Table 4, that there is a statistically significant difference, at P (.001), between the arithmetic means of those who agree on sections II and IV questions. It should be taken into consideration that answers to questions 39 and 40 were included in the "t" test although the would-be aborted fetuses were normal.

A negative correlation coefficient was obtained when the life expectancy of the fetuses at birth was compared with those agreeing on abortion under the circumstances expressed on section IV questions (Table 5). However, as Table 6 indicates over one third of those answering indicated a maximum recurrence of only 1 percent.

TABLE 1: Percentage of Agree, Disagree and Undecided answers per question. Total interviewed 260.

Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided	Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided
I	1	53.5	29.6	16.9	II	21	59.6	32.3	8.1
	2	24.2	58.1	17.7		22	67.3	20.4	12.3
	3	38.5	38.1	23.5		23	21.5	66.5	11.9
	4	14.2	61.2	24.6		24	87.0	7.0	6.2
	5	41.2	40.8	18.1		25	92.3	2.3	5.4
	6	82.7	11.5	5.8	III	26	35.4	54.0	10.8
	7	19.6	66.9	13.5		27	29.2	59.6	11.2
II	8	21.2	68.5	10.4		28	45.4	38.1	16.5
	9	70.8	25.4	3.9		29	32.7	44.2	22.0
	10	75.0	14.2	10.8		30	8.5	81.5	10.0
	11	73.8	13.8	11.9		31	22.3	65.4	11.9
	12	70.8	15.8	13.5	IV	32	65.8	25.0	13.1
	13	78.5	10.4	11.2		33	66.2	20.4	13.5
	14	82.3	11.9	5.8		34	31.2	41.5	27.3
	15	66.5	17.7	15.8		35	29.0	49.2	22.0
	16	80.4	12.7	7.0		36	40.4	36.5	23.1
	17	73.1	17.7	9.2		37	16.5	69.0	14.6
	18	70.0	17.7	12.3		38	32.0	43.5	24.6
	19	50.4	40.4	9.2		39	6.2	87.0	6.9
	20	31.2	62.0	7.0		40	9.2	76.2	14.6

TABLE 2: Percentages corresponding to each question answered by 150 females interviewed

Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided	Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided
I	1	60.0	26.0	14.0	II	21	60.7	32.0	7.3
	2	25.3	54.7	20.0		22	70.7	19.3	10.0
	3	40.0	35.3	24.7		23	21.3	70.0	8.7
	4	15.3	59.3	25.3		24	85.3	7.3	7.3
	5	40.7	36.7	22.7		25	92.0	2.0	6.0
	6	82.0	10.7	7.3	III	26	40.0	50.0	10.0
	7	16.0	71.3	12.7		27	28.0	60.0	12.0
II	8	16.7	73.3	10.0		28	44.7	38.0	17.3
	9	74.7	22.0	3.3		29	36.0	40.0	24.0
	10	77.7	14.0	9.3		30	8.0	82.7	9.3
	11	77.3	12.0	10.7		31	20.7	67.3	12.0
	12	72.7	13.3	14.0	IV	32	62.7	26.0	11.3
	13	81.3	10.0	8.7		33	65.3	20.7	14.0
	14	82.7	10.7	6.7		34	28.7	44.7	26.7
	15	66.0	16.7	17.3		35	30.0	44.0	26.0
	16	80.7	11.3	8.0		36	42.0	32.7	25.3
	17	72.0	18.0	10.0		37	14.7	70.7	14.7
	18	68.7	19.3	12.0		38	31.3	40.0	28.7
	19	53.3	37.3	9.3		39	4.7	86.7	8.7
	20	32.0	62.7	5.3		40	9.3	74.7	16.0

TABLE 3: Percentages corresponding to each question answered by 110 males interviewed

Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided	Section	Question Number	Percentage Agree	Percentage Disagree	Percentage Undecided
I	1	44.5	34.5	21.0	II	21	58.2	32.7	9.1
	2	22.7	62.7	14.5		22	62.7	21.8	15.5
	3	36.4	41.8	21.8		23	21.8	61.8	16.4
	4	12.7	63.6	23.6		24	89.1	6.4	4.5
	5	41.8	46.4	11.8		25	92.7	2.7	4.5
	6	83.7	12.7	3.6	III	26	29.1	59.1	11.8
	7	24.5	61.0	14.5		27	30.1	59.1	10.0
II	8	27.3	61.8	10.9		28	46.4	38.2	15.5
	9	65.5	30.0	4.5		29	28.2	50.0	19.1
	10	72.7	14.5	12.7		30	9.1	80.0	10.1
	11	69.1	16.4	13.6		31	24.5	62.7	11.8
	12	68.2	19.1	12.7	IV	32	70.0	23.6	15.5
	13	74.5	11.0	14.5		33	67.3	20.0	12.7
	14	81.8	13.6	4.5		34	34.5	37.3	28.2
	15	67.2	10.5	13.6		35	27.3	56.4	16.4
	16	80.0	14.5	5.5		36	38.2	41.8	20.0
	17	74.5	17.3	8.2		37	19.1	66.4	14.5
	18	71.8	15.5	12.7		38	32.7	48.2	19.1
	19	46.3	44.5	9.1		39	8.2	87.3	4.5
	20	30.0	61.0	9.1		40	9.1	78.2	12.7

TABLE 4: Differences between arithmetic means of those who agree on sections II (# 10-18) and IV (# 32-40) questions

Question Number	X_1	Question Number	X_2
10	194	32	170
11	191	33	171
12	133	34	81
13	204	35	75
14	213	36	104
15	172	37	43
16	198	38	83
17	165	39	16
18	181	40	24

$\bar{X}_1 = 183.4$ $\bar{X}_2 = 85.2$

$t_{.001}(16) = 4.806$

X_1 = Number of persons who agree on section II questions 10-18
X_2 = Number of persons who agree on section IV questions 32-40

TABLE 5: Correlation coefficient for life expectancy of fetus at birth, and number of persons who agree on abortion (section IV questions 32-40)

Question Number	X_1	X_2
32	0	170
33	3	171
34	18	81
35	36	75
36	46	104
37	56	43
38	66	83
39	76	16
40	86	24

Correlation coeficient = $r = -0.88$

X_1 = Fetus' life expectating in years
X_2 = Number of persons who "Agree" on section IV questions

TABLE 6: Maximum recurrence risk indicated by those interviewed

Risk of Recurrence	Percent Answering
100%	12.0
50%	12.7
25%	14.2
10%	9.6
5%	6.2
1%	36.5
No Answer	9.0

TABLE 7a: Percentage of those who would consider abortion and/or sterilization under special circumstances

	Percentage Abortion-Yes Sterilization-Yes	Percentage Abortion-No Sterilization-Yes	Percentage Abortion-Yes Sterilization-No
MALE:	33.8	4.2	3.5
FEMALE:	45.8	5.8	0.8
TOTAL:	79.6	10.0	4.2

TABLE 7b: Percentages of those who would not consider abortion nor sterilization under any circumstances

	Percentage Abortion-No Sterilization-No
MALE:	1.2
FEMALE:	5.0
TOTAL:	6.2

The decision to reproduce or not to, may be a difficult one for responsible people conscious of their genetic make-up. This awareness may arise by learning of an afflicted relative or by having such a child. Having knowledge, before marriage, of a family history of inherited disease proved to be the one question most agreed upon, by 92 percent of those interviewed (question number 25, Table 1). It was followed closely, with 87 percent, by number 24, the essential need to pass on information to relatives who might inherit, or have children, with the genetic condition concerned. Many of those who answered the questionnaire, expressed verbally the view that they would feel "cheated" if this knowledge was purposefully kept from them until too late (i.e. the birth of an affected child). Failure to recognise the hereditary nature of any such trait was also voiced as a major preocupation.

All of these worries are explainable when one considers the probable results of the known, high degree of inbreeding which took place during the european colonization of our Island. Studies conducted by professors from the University of Puerto Rico's History Department, showed a very high degree of endogamous marriages as late as 1750 to 1850 (12). In two towns, Moca—in the West, and Yabucoa—in the East, the marriage registers kept by the Catholic Church were studied. In Moca, a very small town up in the mountains, 52 percent of the marriages that took place from 1750 to 1800 involved cousins. In Yabucoa, approximatelly, the same size as Moca, but in the eastern coastal plains, 34 percent of the marriages involved cousins from 1800 to 1850 (12). These figures are possibly below the real figures since mostly those who could afford the ceremony were the ones who would appear in the Church register. This situation is analogous to the Jewish tradition for endogeneus marriages. Such consaguineous union may raise the genetic allowance for deleterious genes present, in a given population, from 6 or 8, to 10 to 12 genes.

In our island, one possible result of this situation is the presence of a rare mutation causing Anophtalmia (no eyes). Some 8 cases, from "unrelated" families were studied from the South Central

region of Puerto Rico, a few years ago (12). García-Castro, in 1975, described another such rare mutation in the presence of a nictitating membrane in both eyes of a child with Trisomy 18 - (14). When facts such as these are considered, 92 percent agreement on question 25, is not surprising (see Appendix A).

However, knowing before hand that a person is a carrier of a deleterious gene doesn't necessarily forestall marriage and/or having children (15). On the other hand, social stigmatization, restriction of mating freedom, psychological embarrasment, and guilt feelings if an afflicted child is born of two carriers, is the risk undertaken by anyone who is detected as a carrier in a screening program. George Stamatoyannopoulous (15), professor of medicine at the University of Washington's School of Medicine, does not believe that screening and counseling should be prohibited. Restak (24), believes that genetic counseling should be performed yes, but with the utmost care. He feels that giving away too much information on inherited diseases to persons with insufficient biological information "can act like napalm on marriages, family solidarity and sexual identity".

The knowledge gap between counselor and patient should be narrowed, so that "blind" counseling be eliminated as fast as possible. This is the belief of Patricia T. Kelly (19), Ph.D. genetic counselor in the Genetic Advising Program of the Health and Medical Sciences Program at the University of California at Berkerly. She comented on Restak's article and indicated that genetic counselors should provide enough background information on biology, medicine, genetics, and statistics, so that the patients have a context in which to put the information they receive. Epstein (2), since 1973 had stated the desirability of a counselor having both basic and medical sciences training, or in its deffect, of being associated with a medical-geneticist-physician-counselor.

Huntington's disease is one of the most devastating neurological disorders (1, 9, 16). It has taken many lives such as that of folk singer Woodie Guthrie, who died in a state hospital after fifteen years of emotional and physical suffering both for him and his family. Recently an animal model has been proposed for his autosomal dominant genetic "killer" (9). This model may be used to look for some biological marker of the disease. If such a marker is

found, it might then be usefull in the identification of victims before they reproduce, or perhaps even before they are born. This raises the question of wether to abort or not such a fetus. The "normal" life expectancy of a Huntington's victim is approximately 40 years. In the present survey, 49 percent of those interviewed disagreed with the idea of an abortion for a prospective Huntington's victim. However, when faced with a disease, such as Tay Sachs, where "normal" life expectancy (that is before the onset of the disease) is six months, with retrogression of brain function until death at age 3 or 4, 66 percent agreed on abortion. As can be seen from Table 5, a negative correlation coeficient is obtained when life expectancy of fetus at birth is compared with the number of persons who "Agree" on abortion. This indicates that acceptance of abortion diminishes as the life expectancy of a possibly afflicted fetus, increases.

Legal abortion is becoming progressively less restricted in countries around the world (26). In fact, about a third of the world's people now live in countries with non-restrictive abortion laws. In Puerto Rico, abortion still brings social stigmatization, therefore, although, quite possibly practiced widely, it is tried to be kept secret. Both male and female sterilization, more readily than an abortion, might be inferred from the fact that almost a third of those interviewed indicated a maximum recurrence risk of 1 percent (Table 6). Still, it must be indicated that, although, only 10 percent favored sterilization over abortion (Table 7a) these two statements in particular might be considered "leading". That is, the placement and wording of the statements tended to "suggest" the answer.

Question number 41, proved to be one of the most difficult to understand and to answer. It tended to confuse particularly students who were confronted for the first time with such a decision. The risk of recurrence referred to in this question (see Table 6) was explained in the following manner: Suppose you are shown six guns on a table. The first gun has one bullet in the only bullet hole it has (100 percent); the second gun has two bullet holes and one bullet in one of them (50 percent); the third gun has four bullet holes but, again, only one bullet in one of the holes (25 percent);

the fourth gun has ten holes and one bullet in one of them (10 percent). The fifth gun has 20 bullet holes (5 percent) and the sixth gun has 100 bullet holes (1 percent), each one has only one bullet. You are told to choose one of the guns, put it to your temple, and shoot. Which gun would you choose?...The bullet in each case represents the probability of having a child with a genetic disease. With the guns, it's your life which is being dealt with, the risk of recurrence means you are dealing with your unborn child's life...

The questionnaire itself proved to be somewhat confusing and tiring. One of the major difficulties was its lengthyness and that some of the questions were not clearly stated. For example, the meaning of questions 10 to 18 was not very clear. Questions like: "Does answering 'agree' means I'm in favor of the treatment or the test?", "why is the cost of the test included?", were very common. The explanation given was that if the "government" decided to make these test (or any other kind of test, say as for syphilis) mandatory, then it would have to provide the necessary treatment. This, of course, would come out of everyone's tax-payer's money.

Another difficulty arose with the word "Society". "Who, what, where, is this 'Society'?" "Upon whose shoulder will rest the decision as to who is to be screened and who is not, who has to undergo an abortion and/or sterilization?" These last questions went unanswered both by the questionnaire and the author.

Most of those answering were inclined to approve the mandatory nature of the screening tests indicated in questions 10 to 18. This much can be observed in the results of $\bar{X}_1$, in Table 4. They seem to be saying: "If *we* are to decide, if *we* are this so-called 'Society', then it's agreed." However, when confronted with the possible reality of having to face an abortion as a result of such tests, then the percentage of those who "agree" goes considerably low, as can be ascertained from the results of $\bar{X}_2$ in Table 4. That this difference is statistically significant, allows one to infer that when the posibility becomes a reality, the individual's right to determine the fate of his or hers unborn child is not to be relinquised. That this is so, is clearly stated by the fact that 82.7 percent agree on question 6 (see Table 1).

A long term research proyect on genetic abnormality and behavior, at Harvard Medical School, was finally abandoned in May, 1977 (5). This project, conducted by Dr. Bernard D. Davies came under fire when it was known that it entailed the identification of XYY infants, with follow-up observations and therapy. Two opposers of this project, Beckwith and Miller, also from Harvard, criticized it because sensational publicity accorded "criminality" genes might create serious problems in the lives of the children under observation. Also, no control group of "normal" children was studied alongside the XYY ones. Greater public participation was urged in decisions to conduct scientific research. Such participation is evident in, for example, the current controversy over recombinant DNA research (23).

What kind of scientific research should be carried on and where?, how and by whom should these decisions be made? These points pose a very important question: "where should the line be drawn?" The people have a right to be informed of what's going on but, also, should be correctly educated with the real facts about an issue before being asked to make a decision. What would have happened if, for example. Paul Ehrlich's experimentation on syphillys in the 1890's, or Sigmund Freud's studies on human psychoanalysis had been stopped because they were deemed too "controversial"? On the other hand, what would have happened if it would have been determined in utero, that Beethoven would be deaf at age 30, or that Toulousse-Lautrec was going to be a crippled child?...

CONCLUSIONS

1. Enough evidence has accumulated to indicate the desirability of having genetic information readily available. An overwhelming majority of the Puertorricans interviewed agreed mainly of this point.

2. The individual's right of freedom of choice as to treatment (i.e. abortion and/or sterilization or reproduction) was crearly stated.

3. Both abortion and sterilization were highly favored particularly where the fetus' life expectancy was very short.

RECOMENDATIONS

1. More Genetic Counseling Centers should be set-up throughout the Island, where those concerned may go for professional advice.

2. Public education programs as to the nature and effects of genetic diseases may be set-up by the Public Health Department to reach the majority of the people.

BIBLIOGRAPHY

Articles:

1. Arehart-Treichel, J. Biomedical Science. Huntington's - an infections disease? Science News III, 17:267 - April 23, 1977.
2. Bergsma, D. Ed. "Contemporary Genetic Counseling". April 1973. Birth Defects: Original Article Series IX, 4.
3. Borgaonkar, D.S. and Shah, S.A. "Preface". Advances in Human Genetics and Their Impact on Society" in Birth Defects: Original Article Series VIII, 4:1. July 1972.
4. Braestrupp, P. Ed. "Sociobiology". T. Wilson Quaterly 1, 4:108-143. Summer 1977.
5. Cook, P.S. Periodicals ed. "Genetic Research and Social Ills". T. Wilson Quaterly 1, 2:21. Winter 1977.
6. De Vore, Irven. "The New Science of Genetic Self. Interest" Psychology Today 10, 9:42. February 1977.
7. Dawkins, R. "The Remarkable Replicators". Natural History. 86, 2:34. February 1977 (except for "The Selfish Gene").
8. Dobrish, C.M. "The Double Miracle That's Saving High Risk Newborns". Parents Mag. LII, 6:35. June 1977.
9. Frazier, K. Ed. "Huntington's Disease: An animal Model". Science News 110, 17:263. October 23, 1976.
10. Frazier, K. "Tale of 3 disease: A common cause?" Science News III, 17: 263. April 23, 1977.
11. Friedman, T. "Prenatal diagnosis of Genetic Disease" Science. AM. 225: 34-42. 1971.
12. García Castro, J.M. Personal Communication 1977-1978. Director, Regional Medical Program of Hereditary diseases, Profesor of Pedriatrics, University of Puerto Rico, School of Medicine.
13. ——, y Pérez Comas, A. "Non-lethal achondrogenesis (Grebe-Quelce-Salgadotype) in two Puerto Rican sibships". In Press.

14. ——, y Reyes de Torres, L.C. "Nictitating Membrane in Trisomy 18 Syndrome". Am. J. of Ophtalmology 80, 3: Part II: 550-1.
15. Horn, J. "When Genetic Counseling Backfires". In Newsline. Psychology Today 9, 4:21. September 1975.
16. Horn, J.C. Ed. Newsline. "A New Way to Predict Genetic Disease". Psychology Today 11, 7:28-33. December 1977.
17. Holtzman, N.A. "Newborn screening for genetic metabolic diseases". United States Department of Heath, Education, and Welfare DHEW Publ. (HSA) 77-5207. 1977.
18. Laing, F.C. "Ultrasound". Human Nature 1, 3:50. March 1978.
19. Letters. Phychology Today 9, 8:6-12. January, 1976.
20. Murray, D. Periodical's ed. "Gene Splicing: a Public Debate". T. Wilson Quarterly 1, 3:18. Spring 1977.
21. ——, "Abortion Rights. Questions Persist. T. Wils. Quarterly-1, 3:27. Spring 1977.
22. Murray Jr. R. Personal Communication 1977. Professor of Medical Genetics, School of Medicine, Howard University, Washington, D.C.
23. Murphy, C. Periodicals ed. "T. State of Academic Science". T. Wilson Quarterly 1, 5:51. Autumm 1977.
24. Restack, R. "The Danger of Knowing Too Much". Psychology Today. 9, 4:21. September 1975.
25. Rosenfeld, A. "Birth Defects: Not for Babies Only". Sat. Review 5, 5:42. November 26, 1977.
26. Tietze, C. and Lewit, S. "Legal Abortion". Sci. Am. 236, 1: 21. January 1977.

Books:

27. Childs, B. Chairman. "Genetic Screening: Programs, Principles and Research". National Research Council. Committee for the study of Inborn Errors of Metabolism. National Academy of Sciences. 1975.
28. Gardner, E.J. Principles of Genetics. Fifth edition 1975. John Wiley and Sons, Inc.

29. Moody, P.A. Genetics of Man. Second edition 1975. W.W. Norton and Company.
30. Rothwell, N.V. Human Genetics. 1977. Prentice-Hall, Inc.
31. Stern, C. Principles of Human Genetics. Third edition. W.H. Freeman and Company.

APPENDIX A

ETICA Y TECNOLOGIA GENETICA
INVESTIGACION SOBRE ACTITUDES

HOJA DE CONTESTACIONES

Iniciales . . . . . Edad . . . . . Grupo Etnico . . . . . .

Prof. Clara E. Maldonado
Departamento de Biología
Universidad de Puerto Rico

I. 1. (A) (D) (U)
2. (A) (D) (U)
3. (A) (D) (U)
4. (A) (D) (U)
5. (A) (D) (U)
6. (A) (D) (U)
7. (A) (D) (U)

II. 8. (A) (D) (U)
9. (A) (D) (U)
10. (A) (D) (U)
11. (A) (D) (U)
12. (A) (D) (U)
13. (A) (D) (U)
14. (A) (D) (U)
15. (A) (D) (U)
16. (A) (D) (U)
17. (A) (D) (U)
18. (A) (D) (U)
19. (A) (D) (U)
20. (A) (D) (U)
21. (A) (D) (U)
22. (A) (D) (U)
23. (A) (D) (U)
24. (A) (D) (U)
25. (A) (D) (U)

III. 26. (A) (D) (U)
27. (A) (D) (U)
28. (A) (D) (U)
29. (A) (D) (U)
30. (A) (D) (U)
31. (A) (D) (U)

IV. 32 (A) (D) (U)
33. (A) (D) (U)
34. (A) (D) (U)
35. (A) (D) (U)
36. (A) (D) (U)
37. (A) (D) (U)
38. (A) (D) (U)
39. (A) (D) (U)
40. (A) (D) (U)
41. (A) (D) (U)

100% 50% 25%
10% 5% 1%

ETHICS AND GENETIC TECHNOLOGY
AN ATTITUDINAL SURVEY

Age .... Sex: M F Ethnicity: Wh ... Bl ... Sp ... Or ... Other .....
N° of brothers and sisters ..... Father's job .....
Mother's job ..... Estimated Family Income . $ .....
Religious Affiliation ..... Home State .....
Single ..... Married ..... Separated ..... Divorced .....
Setting reared in: (Where Most Time Spent) Rural ..... Urban .....
Highest Level of Education: ..... College Major: .....
Have you ever taken a Biology course? Yes ..... No .....
Have you ever taken a Genetics course? Yes ..... No .....

..... Check here if you would NOT consider *abortion* under any circumstances.

..... Check here if you would NOT consider *sterilization* under any circumstances.

INSTRUCTIONS:

The following statements concern ethical and moral decisions relative to genetic diseases. Circle A if you agree or agree more than disagree with the statement. Circle D if you disagree or disagree more than agree with the statement. Circle U if you have no tendency to either agree or disagree with the statement.

Please answer as honestly as you can.

I Hemophilia is an inherited disease in Which trauma produces excessive bleeding because there is a deficiency of one of the clotting factors. Death usually occurred in childhood until the recent institutuion of therapy consisting of frequent injections of highly concentrated and purified clotting factor. Only males have this problem because the abnormal gene causing the deficiency is on the X chromosome. Before this new therapy was available males usually died before they married and had children. Every daughter of a male with hemophilia will be a carrier of the gene and will pass it on to half her sons. The sons of hemophiliacs are not affected. The treatment of this disease currently costs $10,000 per year.

Below are listed some actions that might be taken with regard to reproduction and treatment of men with this disease.

1. Males with hemophilia should be sterilized so they won't pass on the gene.
   A D U

2. Since the sex of offsspring can be determined by amniocentesis early in pregnancy, all female offsprings of hemophiliacs should be aborted.
   A D U

3. Female offspring of males with hemophilia and all females known to be carriers of the gene for hemophilia should be sterilized.
   A D U

4. The sex of the fetuses of known female carriers of hemophilia should be identified by amniocentesis and all male infants should be aborted. (There is currently no reliable test for hemophilia in the fetus.)
   A D U

5. Since treatment is available no intervention in reproduction should be allowed.

A D U

6. The parents should always decide what action should be taken in any case.

A D U

7. Since the treatment for this disease is so expensive that the state will usually have to pay for it, society (State or Federal government) or insurance companies should decide what action should be taken.

A D U

II With new techniques in biochemistry, cell culture and chromosome analysis, it is often possible to detect inherited conditions soon after birth or in utero before 20 weeks of pregnancy. It may soon be possible to detect some inherited disorders that don't show up until early or even late adulthood.

Genetic screening tests have been used in some cases and might be used in others to detect these conditions. Please indicate which of the following conditions or situations you feel genetic testing should be MANDATORY.

8. Screening tests for inherited disease should never be mandatory.

A D U

9. Screening test for inherited disease should sometimes be mandatory depending on the situation.

A D U

10. If the screening test will detect an inherited metabolic disorder at birth affecting 1 in 15,000

newborns which if untreated may result in mental retardation, but which can be treated by a special diet. (Example – phenylketonuria.)

A D U

11. If the screening test will detect an inherited blood disorder affecting 1 in every 500-600 U.S. Blacks for which there is no effective treatment but whose symptoms (intermittent pain, recurrent infections) physicians feel can be ameliorated and whose lives can be prolonged from 25 to 30 years to 30-40 years. It costs about $4,000 – $5,000 per year to treat. (Example – sickle cell anemia.)

A D U

12. If the screening test will detect a complex inherited disease affecting 1 in every 2,500 whites, causing a debilating disease of lungs and digestive tract. The usual life expectancy is for about 20 years. Doctors don't know whether or not life can be prolonged by medical therapy. Treatment costs about $4,000 - $5,000 per year. (Example – cystic fibrosis.)

A D U

13. If the screening test will detect a nervous disorder that affects the individual from age 30 onwards in which there is gradual loss of control of hands, feet, chewing, swallowing and finally brain function. Persons usually die in mental institutions. No treatment is available. Each child of these individuals has a 50-50 chance of inheriting this condition and they are usually born before the person knows they have the disease. (Example – Huntington's Disease.)

A D U

14. If the screening test will detect sex chromosome

abnormalities at birth which occur in about 1 in 500-600 infants (mostly in males) and there can be *partially* corrected by surgery and/or hormone treatment. (Example – XXY, Klinefelter's syndrome, which in males produces slight breast development, small testes and sterility.)

A D U

15. If the screening test involves amniocentesis (taking out fluid and cells from around the fetus) in women over 38 years and detection of severe chromosomal disorders that are associated with birth defects and mental retardation. These cannot be treated and if found fetuses would be aborted.

A D U

16. If the screening test detects normal people who carry an inherited disease which has a 25 percent, or 1 in 4 chance of being inherited by the children if two carriers marry. Fetuses with the disease can be detected in early pregnancy and aborted if the parents wish. There is no treatment for the disease which leads to mental deterioration and death by 3 to 4 years of age.

A D U

17. If the screening test detected at birth an anormality of fats in the blood that markedly increases the chance of a heart attack in men (and to a lesser extent women) before age 50 and in which the effectiveness of a low fat diet, which would have to be followed throughout life, was unknown. (Example – Type II Hyperlipoproteinemia.)

A D U

18. If the screening test detects normal people who

carry a gene for an inherited blood disease which cannot be detected in early pregnancy by amniocentesis and which cannot be treated. If two carriers marry each of their children will have a 25 percent or 1 in 4 chance on inheriting this disease. (Example – sickle cell anemia.)

A D U

19. Society should determine the type or kind of disease to be tested for in genetic screening.

A D U

20. Society (government, institutions) should determine who should be screened for genetic disease.

A D U

21. Individuals should determine whether or not they wish to be screened.

A D U

22. Every effort should be made to eradicate genetic disease.

A D U

23. It is more important to control diseases that can be treated than to try to control hereditary diseases.

A D U

24. It is essential to pass on genetic information to relatives who might inherit or have children with the condition even if the person who has the disease objects strenously.

A D U

25. Before marrying it is very important to know whether or not they are carriers of or have a family history of an inherited disease.

A D U

III In each case below legitimate rights are in conflict. Indicate your agreement or disagreement in each case with which rights should predominate.

26. The freedom of the individual predominates over his or her responsibility to society.
A D U

27. The rights of the parents predominate over the rights of their children.
A D U

28. The rights of the parents predominate over the rights of the fetus.
A D U

29. It is better to have no life at all if it is not of a reasonable quality.
A D U

30. The rights of parents are more important tham their obligation to future generations.
A D U

31. The rights of persons with normal intelligence predominate over those who are mentally retarded.
A D U

IV Those individuals who are willing to consider abortion under certain circumstances should indicate whether they agree, disagree or are uncertain about choosing *to abort* a fetus who has an inherited condition that would result in the following situations if he or she is born.

32. The child will die at birth or within a few days, no matter what measures are taken. (Example – anencephaly, in which the brain hemispheres don't develop.)
A D U

33. Development is normal for about six months after birth then there is retrogression of brain function and death by 3 years of age. (Example – Tay-Sach's disease.)
A D U

34. After birth there is a life expectancy of 18 or more years with complicated and expensive medical treatment with a normal but restricted life. (Example – cystic fibrosis.)
A D U

35. Life is normal until 35-40 years of age then Huntington's disease begins (a progressive loss of control of arms, legs, chewing, swallowing and brain function with bizarre movements. Death in mental institution).
A D U

36. Life expectancy is realtively normal but there is severe mental retardation requiring guardianship at home or in an institution.
A D U

37. Life expectancy is normal but there is some retardation in which the individual can be trained and work in a sheltered workshop.
A D U

38. Life expectancy is normal with normal intelligence but the individual is markedly incapacitated. (Example – paraplegic without control of urine or bowels as in spina bifida – failure of normal closure of the spinal column.)
A D U

39. Life expectancy and intelligence are normal

but the fetus is a sex not wanted by the parents.

A D U

40. Life expectancy and intelligence are normal but fetus just not wanted by the parents.

A D U

41. The recurrence risk for an hereditary condition is the chance that it will occur a second time if it has occurred once. It is sometimes used to indicate the chance that a condition will occur at all. What is the *maximum* (highest) recurrence risk that you would be willing to take for a *serious* genetic condition in one of your children. (Circle your choice.)

100% 50% (1 in 2) 25% (1 in 4)
10% (1 in 10) 5% (1 in 20) 1% (1 in 100)