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Attitudes toward Genetic Testing among the General Population and Relatives of Patients with a Severe Genetic Disease: A Survey from Finland

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Summary

In the present study we explore the attitudes of the Finnish population toward genetic testing by conducting a questionnaire study of a stratified sample of the population as well as of family members of patients with a severe hereditary disease, aspartylglucosaminuria (AGU). The questionnaire evaluated attitudes toward gene tests in general and also respondents' preparedness to undergo gene tests for predictive testing, carrier detection, prenatal diagnosis, and selective abortion, in theoretical situations. The results of the study indicate that both the Finnish population in general and family members of AGU patients have a favorable attitude toward genetic testing. However, a commonly expressed reason against testing was that test results might lead to discrimination in employment or insurance policies. Based on the responses, we predict that future genetic testing programs will most probably be met with a high acceptance rate by the Finnish population.

Introduction

Modern biotechnology has created previously unforeseen possibilities in medical genetics to determine the genetic make-up of individuals and to predict their future health. Reliable and low-cost gene tests facilitate detection of genetic diseases in the fetus and prediction of the future onset of a variety of diseases prior to any clinical manifestations, as well as determination of the carrier status, in the case of recessively inherited conditions, representing a potential risk for the disease in future generations.

Optimal utilization of the potential of genetic testing depends greatly on the knowledge and attitudes of the population, the consumers of the tests. The main source of information on gene technology, particularly among the older population, has so far been the mass media: TV, newspapers, and magazines. Only the youngest generation has had up-to-date genetics in its primary and/or high school curriculum. Misconceptions and unfounded expectations are therefore very likely to be common among the general population. Some people may harbor fears of all modern biotechnology and therefore turn down any opportunity for genetic testing.

Attitudes toward and expectations of genetic testing among the general population, and particularly among family members of patients with hereditary disease, have rarely been explored. In conjunction with the recent pilot programs on screening for cystic fibrosis (CF) carriers, the experiences and attitudes of the screened individuals have been evaluated (Kaplan et al. 1991; Watson et al. 1992; Mennie et al. 1993; Witt et al. 1993). A high acceptance rate and a low level of distress among the participants were recorded in these studies. The actual participation rate, however, seems to be somewhat lower than hypothetical willingness to take a test (Mitchell et al 1993; Tambor et al. 1994). A recent survey of participants in a CF carrier screening study indicated that the participants' perception of their health was not influenced by the carrier testing, irrespective of the results (Bekker et al. 1994). Earlier experience of carrier screening for Tay-Sachs disease (Clow and Scriver 1977; Zeesman et al. 1984) and thalassemia (Angastiniotis et al. 1986) by using conventional enzyme and protein assays have likewise revealed favorable attitudes toward screening and a low level of psychological distress.

The prospects for large-scale genetic screening programs are particularly favorable in Finland. The population is characterized by genetic homogeneity, accounting for a spectrum of some 30 recessive diseases enriched in the population with one major mutation in each of them being the main cause of the disease (de la Chapelle 1993). Socioeconomic differences are also relatively small in this Scandinavian country, with a uniform level of basic education and a high standard of public health care.

Here we have explored the attitudes of the Finnish population toward gene testing by conducting a

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questionnaire study with a stratified sample of the population. For comparison, the same survey was carried out among family members of patients with aspartylglucosaminuria (AGU), a lysosomal storage disorder resulting in progressive mental retardation. The first signs of the disease present between 1 and 4 years of age, usually as a delay in speech development. As they reach adulthood, the patients are severely mentally retarded. AGU is one of the recessive diseases enriched in the Finnish population with a carrier frequency of 1:40–1:70 (Aula et al. 1982) and among the first of these diseases for which a gene test for carrier detection is available (Syvänen et al. 1992).

Subjects and Methods

A self-report questionnaire (available upon request) was developed to evaluate attitudes toward gene testing in general and preparedness to undergo gene tests for presymptomatic testing, carrier detection, prenatal diagnosis, and selective abortion. A short description of the concept of genes, genetic testing, and the current applications of gene tests was given at the beginning of the questionnaire. It was stated that genetic tests are currently available only for a relatively small number of diseases but that it is anticipated that testing will become possible for many more disorders in the near future. The written information also indicated that every human being is estimated to be a carrier of 5–10 recessive disease genes.

The questionnaire consisted of 50 self-rating Likert-type statements on the following topics: (1) to whom and when gene tests should be offered (see table 1), (2) arguments for and against gene testing (see table 2), (3) preparedness to undergo gene tests in health care or reproduction in theoretical situations (see fig. 1), and (4) causes for concern (see table 3). One of the following multiple choice answers could be given to each attitude statement: (1) totally agree, (2) partially agree, (3) partially disagree, (4) totally disagree or (5) don't know. Finally, questions about personal health and personal experiences of genetic diseases and/or gene tests were asked, to obtain some background information.

The subjects of the study were (1) a stratified sample of the population of Finland, $n = 1{,}169$, and (2) a group of relatives of Finnish AGU patients, $n = 82$. For statistical analysis, a matched sample ($n = 82$) was drawn from the total population sample by selecting a control case for each respondent from the group of relatives. The matching criteria were gender, age (ten-year groups), education, and area of residence. The characteristics of the samples are shown in table 4.

The original population sample ($n = 1{,}169$; ages 15–69 years) was a stratified sample of the general population of Finland. The sample was stratified at both a provincial level and an urban/rural municipality level and met age and gender quotas. The response rate was 59% (530 men, 639 women). The study population is a representative sample of the Finnish population. The nonrespondents did not significantly differ from the respondents in terms of gender, age, level of education, social class, or area of residence.

The data of the population sample was gathered in September and October 1993 by a marketing research company (Taloustutkimus Oy) accustomed to conducting national poll studies. The elected individuals were personally approached by the interviewer, who collected the socio-demographic data and gave the attitude questionnaire to be returned in a prepaid envelope.

The family members of AGU patients were approached through the Finnish AGU parents association. The questionnaire was mailed to 170 relatives of AGU patients who were asked to return it in the prepaid envelope. Ninety-seven responses (57%) were received, including 70 parents, 7 siblings, and 20 other relatives. Fifteen responses were excluded due to inadequate socio-demographic data. Thus the number of subjects left for analysis was 82.

Results

To Whom and When Genetic Tests Should Be Offered

Ten statements offering differing views on the provision of genetic testing were presented (table 1). The alternatives ranged from obligatory tests before the age of fertility to a total ban of gene testing. Options for the timing of tests included the newborn period, when choosing a spouse, family planning, and pregnancy.

In the analysis of the responses, "agree" and "partially agree" are combined, as are "disagree" and "partially disagree." About 90% of respondents in both groups agreed with the statement "Gene testing should be available to anybody who wishes to have information about disease genes he/she carries." However, 10% of the matched sample (17% of the total population) but only 4% of the relatives of AGU patients supported the view that "gene tests should not be performed at all."

Over half of the respondents in both groups were against mandatory testing before childbearing age. The relatives of AGU patients were significantly more in favor of testing of those considering marriage (65% vs. 31%) as well as testing of all pregnant women (86% vs. 67%). Genetic testing of newborns was supported less by the AGU relatives than by the sample of general population (19% vs. 46%).

Reasons For or Against Gene Testing

Nine statements were used to evaluate why gene tests might be considered acceptable or unacceptable (table 2). The individual's right to know his/her own genetic information in order "to be able to influence his/her own health and life" or "the health and life of offspring"

Table 1

Attitudes toward Gene Testing in General

Statement	Proportions (%) of Respondents Agreeing (Totally or Partially) or Disagreeing (Totally or Partially)		
	Population (n = 1,169)	Matched Sample (n = 82)	AGU Relatives (n = 82)
Gene testing should be obligatory for everybody before fertility age:			
Agree	33	36	28
Disagree	60	53	61
Don't know	7	11	11
Gene testing should be performed on newborn babies:			
Agree	37	46	19[a]
Disagree	53	44	67
Don't know	10	10	14
Gene testing should be available to anybody who wishes to have information about disease genes he/she carries:			
Agree	94	94	91
Disagree	5	4	9
Don't know	1	2	0
Gene testing should not be performed at all:			
Agree	17	10	4
Disagree	73	79	95
Don't know	10	11	1
Individuals should be offered a gene test when choosing a spouse:			
Agree	37	31	65[b]
Disagree	50	44	31
Don't know	13	25	4
Individuals should be offered a gene test in family planning:			
Agree	71	65	85
Disagree	24	22	15
Don't know	5	12	0
All pregnant women should be offered a gene test:			
Agree	69	67	86[b]
Disagree	24	26	9
Don't know	6	7	5

[a] $P < .001$; χ^2 test (AGU relatives vs. matched sample of population; agree vs. disagree).
[b] $P < .01$. See note a.

were the most widely accepted reasons for testing. Saving society health care costs was a justification for genetic testing for ~60% of respondents in both groups.

The argument against testing most widely opted for both among the sample of general population (54%) and the relatives of AGU patients (40%) was the possibility of discrimination (in employment or in insurance policies). Forty-two percent of the general population but significantly fewer—only 22%—of the AGU relatives considered gene tests unacceptable because they may lead to an increase in selective abortions. Other public health problems were considered to be more important by 30% of the matched sample, but only by 9% of the AGU relatives.

Preparedness to Undergo Gene Tests

Four groups of questions were used to evaluate the conditions under which the respondents would consider undergoing tests for the following purposes: (*a*) predictive testing for late-onset diseases (fig. 1*A*), (*b*) carrier testing (fig. 1*B*), (*c*) prenatal diagnosis (fig. 1*C*), and (*d*) selective abortion (fig. 1*D*). In the introductory part of the questions, the assumption was made that in addition to monogenic disorders, predisposition to some polygenic diseases—and to some behavioral characteristics such as homosexuality—could in the future be revealed by gene tests.

As can be seen from figure 1*A*, willingness to take a gene test to evaluate predisposition to some common diseases was similar in both test groups. About 70% of respondents would take a test to identify their predisposition to a cardiovascular disease, about 60% for a predisposition to hereditary cancer, and about 50% for a predisposition to schizophrenia.

Preparedness to take a carrier test (fig. 1*B*), to opt for prenatal diagnosis (fig. 1*C*), or to terminate an affected pregnancy (fig. 1*D*) varied for different groups of disorders. The matched sample of general population and the

Table 2

Reasons For or Against Acceptance of Gene Testing

Statement	Proportions (%) of Respondents Agreeing (Totally or Partially) or Disagreeing (Totally or Partially)		
	Population (n = 1,169)	Matched Sample (n = 82)	AGU Relatives (n = 82)
Society would save on the costs of treatment of diseases:			
Agree	62	58	59
Disagree	33	38	38
Don't know	5	4	4
People have the right to know about their genes to be able to affect their own health and lives:			
Agree	89	94	93
Disagree	9	2	7
Don't know	2	4	0
People have the right to know about their genes to be able to affect the health and life of their offspring:			
Agree	84	93	96
Disagree	13	5	4
Don't know	2	2	0
There are bigger public health problems that should be taken care of first:			
Agree	40	30	9[a]
Disagree	51	60	86
Don't know	9	10	5
The natural order should be respected without interference:			
Agree	35	27	17
Disagree	59	66	81
Don't know	6	7	2
Knowledge of test results may lead to discrimination against disease gene carriers (e.g., in employment and insurance policies):			
Agree	59	54	40[b]
Disagree	32	32	49
Don't know	8	14	11
Testing would make abortions more common:			
Agree	47	42	22[c]
Disagree	41	43	72
Don't know	12	15	6

[a] $P < .001$; χ^2 (AGU relatives vs. matched sample of population; agree vs. disagree).
[b] $P < .05$. See note a.
[c] $P < .01$. See note a.

AGU relatives differed slightly in their opinions. The relatives of AGU patients were more willing to take prenatal diagnosis for mental retardation (89% vs. 68%) and for lethal metabolic disease (85% vs. 65%). In both groups, prenatal diagnosis was opted for more frequently than selective abortion.

In regard to different diseases only a few significant differences were noticed in the responses of the matched population sample. They were more willing to choose abortion for lethal metabolic disease than for cardiovascular disorder ($P < .01$) or cancer ($P < .05$), but no significant differences were found in responses for carrier testing and prenatal diagnosis. The AGU relatives were significantly more willing to take tests or choose abortion for mental retardation ($P < .005$) and lethal metabolic disease ($P < .01$) than for other disorders.

In addition to distinct disorders and diseases we also asked about respondents' opinions regarding gene tests for homosexuality and the sex of a fetus. Twenty-two percent of the matched population sample would take prenatal diagnosis for homosexuality and 10% for identification of the sex of the fetus. The figures for the group of AGU relatives were 15% and 9%, respectively. Selective abortion for homosexuality was favored by ~10% and for undesired sex by 2%–3% of respondents in both groups.

Concerns about Genetic Testing

Four statements were presented to evaluate possible causes for concern about gene tests (table 3). The most

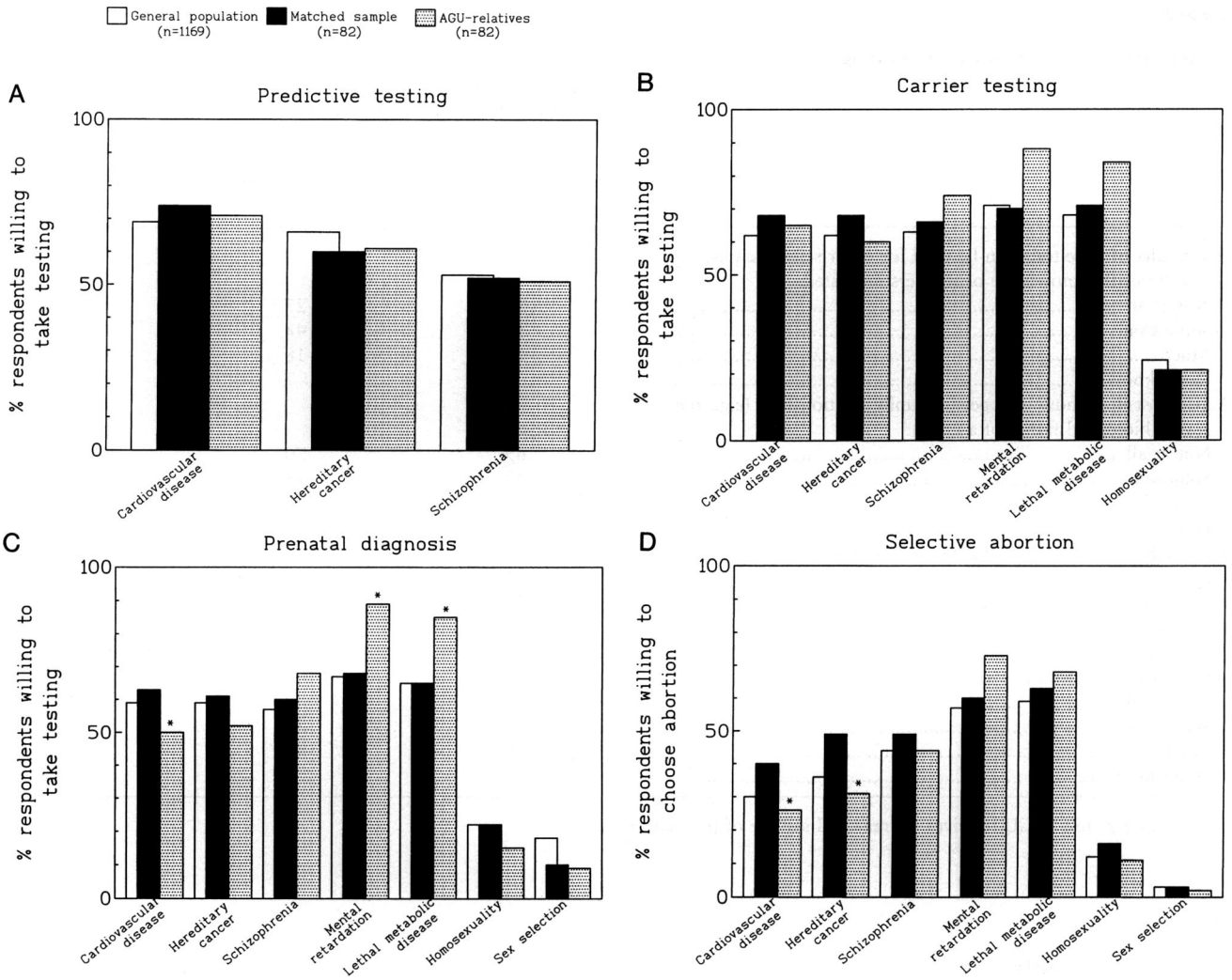

Figure 1 Proportions of respondents willing to take a gene test (or choose abortion) for certain disorders/characteristics in theoretical situations. *A*, predictive testing; *B*, carrier testing; *C*, prenatal diagnosis; and *D*, selective abortion. An asterisk (*) indicates $P < .05$ for difference between AGU relatives and matched sample, χ^2 test.

common cause for concern was that gene testing could lead to "eugenics" (a term not specified further). The possibility that unauthorized groups might obtain information from gene tests also worried the respondents: 38% of the matched population sample (28% of the total population) and 17% of the AGU relatives expressed great concern about this topic. The possibility that results could be used for other scientific purposes was less worrisome. The discovery of unexpected family relationships via gene analyses was a relatively minor cause for concern (72% of the total population and 85% of the relatives did not express any concern).

Other Issues

Ninety percent of the population and 99% of the relatives expressed confidence when asked whether the current health care system in Finland would serve the best interests of consumers in matters of genetic testing. About 90% of respondents were also confident that they would retain their complete individual freedom both to participate in genetic tests and to decide how the results would be applied.

Discussion

The present questionnaire survey indicated that the Finnish population in general, and family members of patients with AGU in particular, have a positive attitude toward genetic testing in risk determination for a variety of genetically determined disorders. The effect of age, gender, and education in responses will be reported separately (authors' unpublished information). Preparedness to undergo gene tests for predictive diagnosis of a late-onset disease, for carrier identification of a recessively inherited disorder, and for prenatal diagnosis was high, especially for disorders causing mental retardation

Table 3

Reasons for Concern about Gene Testing

	Proportions (%) of Respondents Worried about Possible Adverse Effects of Gene Testing		
	Population (n = 1,169)	Matched Sample (n = 82)	AGU Relatives (n = 82)
The results of gene tests can be used for other scientific purposes without the knowledge of the persons tested.			
Not at all	27	27	37[a]
Somewhat	54	48	57
Much	15	18	4
Don't know	4	7	2
A gene test may show unexpected family relationships (e.g., the true biological father of a child).			
Not at all	69	70	85[a]
Somewhat	22	18	11
Much	3	5	0
Don't know	6	7	4
The results may end up in the hands of outsiders.			
Not at all	14	12	22[a]
Somewhat	56	49	58
Much	28	38	17
Don't know	3	1	3
Gene testing may lead to "eugenics."			
Not at all	15	9	14
Somewhat	39	38	49
Much	40	46	32
Don't know	6	7	5

[a] $P < .05$; χ^2 test (AGU relatives vs. matched sample of population).

Table 4

Characteristics of the Groups Surveyed for Attitudes toward Gene Testing

	General Population (n = 1,169) n (%)	Matched Sample (n = 82) n (%)	AGU Relatives (n = 82) n (%)
Sex:			
Females	639 (55)	50 (61)	50 (61)
Males	530 (45)	32 (39)	32 (39)
Age (years):			
15–19	99 (9)	4 (5)	4 (5)
20–29	247 (21)	4 (5)	4 (5)
30–39	263 (22)	21 (26)	21 (26)
40–49	232 (20)	29 (35)	29 (35)
50–59	181 (16)	17 (21)	17 (21)
60+	147 (13)	7 (9)	7 (8)
Education:			
Low (<10 years)	368 (32)	19 (23)	20 (24)
Intermediate	672 (58)	55 (67)	54 (66)
High (university)	127 (11)	8 (10)	8 (10)
Area of residence:			
Southern Finland	259 (22)	31 (38)	31 (38)
Western Finland	295 (25)	2 (2)	2 (2)
Middle Finland	227 (20)	21 (26)	21 (26)
Eastern Finland	215 (18)	18 (22)	18 (22)
Northern Finland	173 (15)	10 (12)	10 (12)

and severe metabolic diseases. The opinions of family members of patients with AGU, a disease resulting in severe progressive mental retardation, and the general population were found to be surprisingly similar in the analysis. The family members would, however, favor even wider application of gene tests.

Certain discrepancies in the responses are probably due to the statements in the questionnaire not being mutually exclusive. The term "genetic testing" was not further specified in the questions dealing with acceptance and concerns, and this also might have led to some discrepancies, since attitudes toward various types of genetic tests were different (as can be noticed in table 1). In our study we did not present any questions dealing with respondents' preparedness to pay for tests or convenience related to testing. In other studies it has been noticed that convenience plays a major role in utilization of testing (Green 1992; Tambor et al. 1994).

Only a little difference was noticed in attitudes of the population sample in regard to different diseases. For example, no significant difference was found between preparedness to choose abortion for lethal metabolic disease versus schizophrenia. This could reflect the heavy social stigma of psychiatric disorders as well as the difficulty of obtaining a realistic view in a hypothetical situation dealing with unfamiliar disorders.

The positive attitudes toward gene testing found in our survey reflect the overall attitude of the population toward health care. Equalized opportunities for high-quality public health care in Finland have resulted in almost 100% participation, particularly in maternity and child care programs. Providing future gene testing through public health care systems will probably also meet with a similar high acceptance rate.

Our study, however, also revealed some sources of concern about gene testing. The possibility for discrimination on the basis of test results and the concept of disturbing the order of nature made some respondents hesitant or negative toward gene tests. Employment and insurance policies were stated as examples for possible situations of discrimination. In addition to public health care, individual private health and pension insurance policies are available in Finland. Respondents may also be concerned about restrictions in public health care, for economic reasons, in the future. Many people also expressed concern that test results might be disclosed to unauthorized persons. Such notions must be carefully considered and taken into account in planning large-scale genetic testing. To maintain the current high level of trust in health care, including medical genetics, informing the public and educating health care personnel must be made high priorities when launching testing programs.

The results of the present study are in line with previous reports on attitudes toward genetic testing. Previous studies, however, deal mostly with one specified disease only, such as Tay-Sachs disease (Clow and Scriver 1977; Zeesman et al. 1984), thalassemia (Rowley 1984), and, more recently, CF (Williamson et al. 1989; Decruyenaere et al. 1992). CF carrier screening seems to be widely accepted, although the willingness to actually participate in the screening programs has been somewhat lower. A majority of respondents in earlier studies have expressed interest in prenatal diagnosis of CF, but the proportion of those considering termination of a pregnancy has been much lower (Decruyenaere et al. 1992). The attitudes of recent parents of healthy babies toward carrier screening programs in general was recently surveyed by Green (1992). Screening was widely supported by the parents, the advantages of testing being considered more important than the disadvantages. The possibility of discrimination on the basis of the test results was found to be the most commonly expressed reason against testing in this study also.

The attitudes of family members of patients with a severe genetic disease have only rarely been evaluated. In the study by Watson et al. (1991) the relatives of CF patients favored carrier testing on a community basis. Ninety percent of those wishing to be tested would have opted for prenatal diagnosis, while only 60% would have considered termination of an affected pregnancy. In a recent study, both adult CF patients and their parents supported carrier screening as well as the option of terminating an affected pregnancy (Conway et al. 1994).

The feasibility and acceptance of genetic testing in the Finnish population will be further evaluated in an ongoing pilot carrier screening program for AGU in the maternity clinics in Helsinki. A rapid and simple PCR-based detection system offers an excellent technical means of detecting carriers of one mutation, the so-called AGU_{Fin} mutation, which is responsible for 98% of disease alleles in Finland, with a carrier frequency of 1:40–1:70 (Syvänen et al. 1992; Hietala et al. 1993). A questionnaire survey of the attitudes and experiences of 2,000 participating mothers will be carried out both soon after the test and after the delivery. This survey should tell us how women make decisions in a real situation and how gene testing influences the attitudes toward the baby and the pregnancy.

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