Carrier Screening for Cystic Fibrosis: A Pilot Study of the Attitudes of Pregnant Women

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ABSTRACT

We surveyed the attitudes of a consecutive sample of 306 pregnant Caucasian women toward carrier screening for cystic fibrosis. Of the 214 respondents, 98% said that screening should be offered before pregnancy, and 69% said they would accept carrier screening during pregnancy. Twenty-nine percent of the respondents indicated a willingness to terminate a pregnancy if the fetus were found to have cystic fibrosis. We conclude that carrier screening is of interest to pregnant women, although interest in terminating a pregnancy because of screening results may be limited. (*Am J Public Health.* 1992;82:723–725)

Introduction

The identification of the gene responsible for cystic fibrosis (CF) offers the capability of performing carrier screening for CF in the general population. Cystic fibrosis is an autosomal recessive disease and is the most common lethal genetic disease in Caucasians. The incidence at birth is approximately 1 per 2500 Caucasian infants. The carrier (heterozygote) frequency is approximately 1 in 20 to 25 Caucasians in North America.[1] Carriers for CF are asymptomatic; however, individuals may be interested in knowing their genetic status in order to make informed reproductive decisions.

Carrier screening for CF in the general population is controversial.[2–5] The initiation of carrier screening in the general population has been inhibited by the relatively low sensitivity of the current technology for screening[6–8]; to date, only 80% to 85% of the CF mutations have been identified in most populations.[9]

If carrier screening for CF is offered to the general population, a primary interest group is likely to be pregnant women. A body of literature exists that describes and analyzes the attitudes and behavior of women toward screening and prenatal diagnosis in a variety of circumstances.[10–18] However, CF has its own distinctive features as a disease, including its morbidity and mortality, risk levels, and the population group at risk. This pilot study was undertaken to explore the interest in carrier screening for CF in pregnant women without a family history of CF and to determine the attitudes of such women toward the potential use of this information in making reproductive decisions.

Methods

Subjects were recruited from the obstetric clinics at a large urban county hospital in Cleveland and at a suburban health maintenance organization in northeastern Ohio. A consecutive sample of 306 pregnant Caucasian women at less than 18 weeks' gestation was recruited to participate in the study. The participants' baseline knowledge of CF was assessed by their responses to three questions; the women were then asked to read educational materials and complete a 40-item questionnaire at home. The participants were encouraged to complete the questionnaire with the father of the baby, when appropriate. Respondent confidentiality was maintained. Pretest responses were available from 84 women who did not return the questionnaire.

Part 1 of the questionnaire assessed demographic information, attitudes about the present pregnancy, and history of previous pregnancies. Part 2 of the questionnaire consisted of information about CF, the carrier state, and reproductive options for carriers. The information was written at a 10th-grade reading level. The information was reviewed for accuracy and balance by parents of persons with CF and by medical specialists. Part 3 of the questionnaire consisted of a test of factual knowledge about CF, followed by items designed to ascertain attitudes about testing and reproductive decisions. Most questions were designed to elicit yes or no responses on a 4-point scale: (1) Yes, definitely; (2) Yes, probably; (3) No, probably not; (4) No, definitely not.

Data were analyzed with the Statistical Package for the Social Sciences/PC+. Analysis for this project consisted primarily of univariate and bivariate analysis, including cross-tabulation and correlational procedures.

Results

Of the 306 questionnaires distributed, 214 were returned in usable form, for a

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TABLE 1—Demographic Characteristics of Respondents (n = 214)

Age, y	27.0 ± 5.7 (range 15–42)
Education,	%
High school or less	42
Technical school	3
Some college	22
College graduate	25
Advanced degree	7
Employment	
Homemakers	31
Laid off	1
Unemployed	7
Students	7
Full-time employment	40
Part-time employment	14
Religion	
No affiliation	28
Catholic	40
Jewish	1
Pentecostal/Southern Baptist	7
Other (Protestant denom.)	24
Marital Status	
Married	81
Parity	
0	42
1	36
2	14
3+	8
Previous abortions (n = 145)	
Yes	25
No	75

TABLE 2—Posteducation Knowledge of Cystic Fibrosis (CF) (n = 214)

Question	Correct Response (% Who Answered Correctly)
1. CF is a disease of the ____?	Lungs and digestion (100)
2. Children with CF are usually ____?	Not retarded (99)
3. Most people with CF will die by ____?	30 years of age (93)
4. Carriers for CF show no signs of the disease	True (96)
5. Chance of CF in a child of two carriers is ____?	1 in 4 (93)
6. Chance *you* are a carrier for CF?	1 in 20 (53)
7. Brothers and sisters of carriers are likely to be carriers.	True (87)

TABLE 3—Participants' Interest in Carrier Screening for Cystic Fibrosis (n = 214)

Question	Responses, %			
	Yes (Total)[a]	Yes, definitely	No (Total)[b]	No, definitely not
1. Is it important to know carrier status before marriage?	75	33	25	6
2. Should carrier screening be offered before pregnancy?	98	69	2	1
3. Would *you* have taken the test before pregnancy?	84	58	16	2
4. Would *father* have taken the test before pregnancy?	79	48	21	4
5. Would *you* have the test done now during pregnancy?	69	40	31	14
6. Would *father* have the test done now during pregnancy?	63	34	37	13

[a]Yes (Total) = "Yes, definitely" + "Yes, probably."
[b]No (Total) = "No, definitely not" + "No, probably not."

response rate of 70%. There were no variables with more than 2% missing data. The demographic characteristics of the study population (Table 1) demonstrate participation by women with a broad range of demographic and socioeconomic characteristics.

Seventy-eight percent of the respondents had heard of CF, and 14% of those had known someone with the disease, although none had siblings or children with CF. Of those who had heard of CF, 47% were aware that it was a disease of the lungs. There was no significant difference in baseline knowledge between responders and nonresponders (n = 84).

Responses to the knowledge questions (Table 2) demonstrate that the participants read the information and that the information was effective in transmitting factual knowledge of CF. It is notable that 11% of the women responded incorrectly that the risk of two carriers' producing an affected child is 100%. Respondents also were likely to underestimate their personal risk of being a carrier: 43% responded that they were at a 1 in 400 risk of being a carrier (the lowest risk level offered as a choice). For each of these two questions those answering incorrectly were less likely to have attained a high level of formal education than were those who answered correctly ($P \leq .01$).

The results shown in Table 3 demonstrate that the participants had a strong interest in being tested for CF carrier status before pregnancy (84%) and during pregnancy (69%). Those who had heard of CF ($P \leq .01$), those who knew someone with CF ($P \leq .01$), those who answered correctly on the nature of CF on the pretest ($P \leq .05$), and those who attended religious services frequently ($P \leq .01$) were *less* likely to want carrier testing during pregnancy. Age, education, and employment were not predictive of the desire to be tested, and those who underestimated their personal risk of carrier status were not less likely to want testing.

With respect to reproductive choices (Table 4), the majority of women (67%) said they would be interested in prenatal diagnosis if they were at risk for having a child with CF; however, a minority (29%) expressed a willingness to terminate a pregnancy if the fetus were found to have CF. Choices about abortion were not related to age, education, employment, pretest knowledge of CF, or perceived risk. Our sample contained a high proportion of Catholic women; however, Catholics were not less likely to choose abortion, nor were those who said they had a religious affiliation versus those who did not.

To estimate the proportion of affected pregnancies that would have been identified and terminated in our sample population, we analyzed the data to estimate the sample's use of testing for each stage of a screening protocol. These results (Table 5) suggest that approximately 24% of affected pregnancies among the women offered screening would have been identified and terminated as a result of a carrier screening program. This estimate assumes a 100% test sensitivity.

With respect to issues of confidentiality and free choice, 99% of the respondents indicated that they would be willing

TABLE 4—Participants' Interest in Using Cystic Fibrosis (CF) Screening to Make Reproductive Choices (n = 214)

Question	Responses, %			
	Yes (Total)[a]	Yes, definitely	No (Total)[b]	No, definitely not
1. Is carrier status a good reason for two carriers not to marry?	9	0	91	43
2. If *you* and the *father* were both carriers, would you want the baby tested in the womb?	67	44	33	16
3. If you found your child would have CF, would you have an abortion?	29	9	71	42

[a]Yes (Total) = "Yes, definitely" + "Yes, probably."
[b]No (Total) = "No, definitely not" + "No, probably not."

TABLE 5—Estimated Sequential Use of Services

A. Proportion of pregnant women accepting screening	69%
B. Proportion of (A) with father accepting screening	86%
C. Proportion of (A+B) choosing prenatal diagnosis	86%
D. Proportion of (A+B+C) choosing termination	47%
Complete utilization = A × B × C × D = 24%	

to share their test results with family members. A large majority (80%) felt that carrier testing should not be required by law, and 60% said that physicians should not be required to divulge the results of CF testing to family members against the wishes of the patient.

Discussion

The results demonstrate a substantial interest in carrier screening in this sample and support a design for a program that is voluntary and confidential. Although the majority of the women were interested in knowing their carrier status, most indicated a reluctance to use the information to prevent the birth of an affected child. Of particular interest is our finding that those who were more familiar with CF tended to be less interested in carrier screening. The hypothetical nature of this study limits our ability to make confident predictions of actual behavior in a screening program, and attitudes may vary with time, clinical approach, and test validity. □

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