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# Teaching about Cystic Fibrosis Carrier Screening by Using Written and Video Information

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## Summary

We performed two studies using only written and video materials to educate people about cystic fibrosis (CF) and carrier screening. Participants were randomized to receive written or video materials. All received a brief questionnaire. Subjects in group I ($n$ = 238) were (1) individuals in steady relationships and their partners, (2) ≥18 years old, and (3) not pregnant. Those who accepted free screening and were not demonstrable carriers were sent a letter explaining their results and another questionnaire. Subjects in group II ($n$ = 108) were parents seeking well child care in a university clinic. The main outcome measures were ability to answer questions correctly about (1) health status of CF carriers and people with CF, (2) the possibility of false-negative results, and (3) for those who had screening, the implications of their own results. Written and video materials were equally effective in conveying information. Prior to screening, subjects answered an average of 86% of questions correctly. Subjects with less formal education answered fewer questions correctly; 60% of those with less than a high school education had adequate knowledge of the health consequences of having CF or being a carrier, compared with ≥94% of college graduates. Performance improved after screening. Where neither partner was a demonstrable carrier, 88% knew their own and their partner's test results, and 90% indicated that their risk of having a child with CF was not zero. Written and video educational materials can be used without face-to-face counseling to inform most people about carrier screening and their test results. These materials may be less effective for those with lower educational backgrounds.

## Introduction

Carrier screening for cystic fibrosis (CF) illustrates many of the problems that can be associated with offering carrier screening to the general public. Even though CF is a common genetic disorder among white families and its treatment is the target of many fund raising efforts, relatively few people in the general population know much about CF (Decruyenaere et al. 1992; Magnay et al. 1992). Among those who have heard of CF, some may have inaccurate impressions of the disease since the prognosis for individuals who have this multisystem disorder primarily affecting the lungs and digestive tract has improved dramatically in the past 40 years. Individuals in the general population know even less about the strengths and limitations of carrier screening (Committee on Assessing Genetic Risks 1994). Since almost 500 different mutations that can cause CF have been described to date, general population carrier screening for CF cannot detect all carriers (Tsui 1994). The usual practice of screening for the six most common mutations detects ≤85% of all carriers who have a Northern European background, so approximately one in six of all carriers are missed (U.S. Congress, Office of Technology Assessment 1992). Even when 32 mutations are sought, only 90% of carriers of this ethnic origin are detected (Integrated Genetics 1994). The carrier detection rate is much lower in individuals from most other ethnic backgrounds.

The impossibility of detecting all carriers has led to several concerns. First, people who are not found to have the specific mutations sought may believe, inaccurately, that they have no risk at all of having an affected child. These individuals may be surprised or angry if they later have a child with CF, raising the specter of litigation. A second concern arises from the difficulty experienced by many in understanding probabilistic information. Specifically, couples in whom one partner is shown to be a carrier and the other is not have a residual risk of ~1/640, in the absence of any family history in the latter partner. Such couples may be distressed, a situation aggravated by the lack of clear options for prenatal diagnosis (Wald 1991; Asch et al. 1993).

Despite the population's general lack of knowledge about CF, its inheritance, and the limitations of carrier screening, several factors make it unlikely that prospective parents will be offered comprehensive counseling about carrier screening for CF by genetic professionals.

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First, this technology is heavily marketed to primary care providers, in the hope that they will order these tests for their patients (Genetic test from Eastman [1994]) Second, the move to managed care and capitation may give primary care providers incentives not to refer patients to genetic professionals who might provide more complete information. Third, given the number of genetic tests currently and soon to be available, there simply are not enough genetic professionals available to provide traditional counseling and testing if many people are interested in being tested (Wilfond and Fost 1990, 1992)

To address these problems, we devised written and video educational materials discussing CF carrier screening to be used without face-to-face counseling. If effective in conveying information, such materials could be used in the primary care setting to help individuals make informed decisions about the use of such tests. We report here our findings regarding (1) the efficacy of these materials in educating individuals who generally lack a family history of CF about carrier screening; and (2) the ability of those who accept screening and are not found to be carriers to understand the implications of their test results without face-to-face counseling.

## Subjects, Material, and Methods

### *Subjects and Protocols*

These educational materials were administered to two groups of subjects.

*Group I.*—This group consisted of individuals interested in having CF carrier screening who were at least 18 years of age, who were not pregnant at the time of the study, and who stated that they had a steady partner. A sign with accompanying letters was used to offer screening in the following sites: (1) a public health clinic in which women receive primary care; (2) a hospital infertility clinic; (3) a hospital based obstetrics and gynecology clinic that serves pregnant and nonpregnant women; (4) a private obstetrics and gynecology practice that also serves many nonpregnant women; (5) a hospital based pediatric continuity clinic; (6) a private pediatric practice; (7) Vanderbilt employee health service; (8) a CF Walk-A-Thon; (9) an after-hours walk-in site at a public high school that was made available as a nonhospital site, following advertising in the community and at the CF Walk-A-Thon; and (10) a table set up in a main thoroughfare in the research area of the medical center. The letters available with the signs stated that the study had two parts. Subjects were first invited to review information about CF and carrier screening and to respond to a brief questionnaire during their visit. They were informed that participation in this first stage would entitle them to a free book about child care and the opportunity to have free carrier screening. The packet of educational materials and questionnaire contained a cover sheet seeking their informed consent to participate in this part of the study. The packet also included an informed consent for screening, in which participants were asked to give their names and addresses. In the second part of the study, individuals who were interested in having free carrier screening were required to take a copy of the written information and questionnaire to their partner. Both partners then had to give their informed consent to screening, to submit blood samples obtained by finger stick, and to agree to answer some more questions after they received test results.

*Group II.*—To increase the diversity of the cohort who received our educational materials, we conducted a separate study in which parents whose children were receiving primary care in a resident continuity clinic were asked to read the written information or view the video and respond to a brief questionnaire in exchange for a free book about child care. Unlike those in group I, individuals in this second study were actively solicited to participate by personnel in the clinic. Since individuals in group II did not provide identifying information about themselves, because they were not asked to review a consent form for screening as part of this study, they were not asked to provide written consent. Neither were they asked to invite their partners to view the materials and answer the questions. After these participants reviewed the educational materials and responded to the questionnaire, however, they were informed about the opportunity to obtain free carrier screening by participating in the main study.

*Randomization and educational intervention.*—All subjects in groups I and II who participated at the study sites were randomized by day of recruitment to receive either written or video information. Some subjects may have been aware of the method in use on the day they considered participating. In both studies, the written and video materials were the only patient education about CF and CF carrier screening provided. To this end, health care professionals at the various sites were instructed not to answer any questions the subjects might have, but rather to refer the subjects to the genetic counselors involved in this study.

*Notification of the results of screening.*—Letters with the results of CF carrier screening tests were sent to the couples in group I who chose to have the test. If neither partner was found to be a carrier, the letter explained their residual risk and asked each partner independently to fill out an additional questionnaire that was designed in part to assess their understanding of their results. Their responses are discussed in this paper. If one partner was found to be a carrier, the couple was invited to come to Vanderbilt for free, face-to-face counseling. Because these couples spoke with a genetic counselor

(V.L.H. or J.P.P.) on the telephone, if not in person, their experiences are not included in this paper.

*Assessment of efficacy before screening.*—The efficacy of the educational materials in conveying factual information about the health implications of having CF or of being a carrier and about the possibility of false-negative screening results were assessed using a multiple choice questionnaire. Identical questions were used to assess knowledge about CF after screening. The questions used are set forth in appendix A. Educational efficacy was assessed in three ways. First, the number of questions answered correctly was counted. Second, subjects were judged to have adequate knowledge prior to screening (1) about the health implications of having CF or being a carrier, if they correctly answered one of the questions about the general health and longevity of individuals with CF (question c or d) or the question about the general health of CF carriers (question e) (*adequate information*); and 2) about the limitations of carrier screening, if they indicated that people could still be carriers even if they were told that they probably are not carriers (question f) (*adequate understanding*). Subjects who requested CF carrier screening but who failed to demonstrate adequate information or understanding were sent letters explaining in greater detail the points they had missed and offering the opportunity to opt out of screening. Subjects in both groups also provided information about their gender, age, education, marital status, and their own health and the health of their children, if any; and indicated whether they were working outside the home and whether they had previously heard of CF.

*Assessment of efficacy after screening.*—After they received their test results, subjects who had CF carrier screening and were not found to be carriers were judged to have adequate knowledge about (1) their test results, if they knew their results; and (2) the implications of these results, if they said that their risk of have an affected child was low or very low, but not zero.

*Protection of human subjects.*—These protocols and consent procedures were approved by the institutional review board at Vanderbilt University Medical Center.

### *Educational Material*

We developed a four-page written handout describing the health implications of having CF and of being a carrier, how CF is inherited, the ability of carrier screening to detect most but not all carriers, and the reasons why people might choose or reject carrier testing. These materials were repeatedly revised, on the basis of results of four pilot studies conducted among parents whose children were being seen at the Pediatric Acute Care Clinic at Vanderbilt Children's Hospital and suggestions of an advisory board consisting of community health care professionals, parents of children with CF, and others. The final handout was written at the sixth-grade reading level, as assessed by Grammatik 5 (1994). These materials are contained in appendix B and may be used without permission of the authors or publisher. Once the written materials were finalized, a videotape running ~8 min, containing the same information, was prepared professionally.

### *Statistical Methods*

Continuous variables were compared using analysis of variance, followed by Wilcoxon rank sum tests for the two preplanned comparisons (written randomized vs. written nonrandomized; written randomized vs. video randomized). The number of questions answered (out of five), and the number of questions answered correctly (out of the five possible and among only those questions answered), were analyzed using binomial regression in the generalized linear model framework (McCullagh and Nelder 1989). A response of "don't know" was scored as an incorrect answer. An overall analysis of the three groups would be equivalent to an analysis of variance for normally distributed data. The two preplanned comparisons would be equivalent to a *t*-test for normally distributed data. $\chi^2$ and Fisher's exact test (or extensions) were used for categorical data. Paired data were compared using Wilcoxon signed rank test for continuous variables and McNemar's test for binary variables. Results were analyzed separately for the two study groups as well as for each educational stratum to assess consistency of any patterns found. Logistic regression was used for binary variables (adequate information and adequate understanding) and binary regression (for number of questions answered and number of questions answered correctly) to assess the effect of type of material, after adjusting for educational level (classified into five ordered strata, coded 1 [lowest] through 5 [highest]) and study group, submission of a blood sample, and randomization to a treatment group.

## Results

### *Characteristics of Subjects*

In all, 238 subjects in group I read or viewed the educational materials and returned the initial questionnaire. Table 1 summarizes the number of subjects in group I who enrolled through the various study sites. The overwhelming majority (71%) enrolled in the site frequented by personnel at the medical center. Only four individuals enlisted in group II later elected to seek free carrier screening by enrolling in group I. Their responses were included in those from the pediatric continuity clinic.

When those in group I are combined with the 108 who participated in group II, 162 individuals were randomized to receive the written handout, 75 were ran-

**Table 1**

**Sites Where Subjects in Group I Enrolled in The Study**

| Sites | No. of Subjects[a] |
|---|---|
| Public health | 13 |
| Infertility clinic | 12 |
| University OB-GYN | 6 |
| Private OB-GYN | 6 |
| Continuity pediatrics | 9 |
| Private pediatrics | 2 |
| Employee Health | 6 |
| CF Walk-a-thon | 8 |
| Public walk in at high school | 6 |
| Medical center lobby | 170 |

[a] Includes both the persons who actually obtained information at the site as well as their partners.

domized to view the videotape, and 102 individuals received the written handout as partners. Seven partners received video on the same day as the randomized subject; these individuals were included in the video group, so the video group contains a total of 82 subjects. There is a disparity in group II between the number of participants assigned to written and video materials. Recruitment for group II, by chance, began at the beginning of a brief period of days designated in our randomization table for use of written materials and was so successful that the target of 100 was almost reached prior to a day assigned to video. There was no evidence that clinic personnel were more reluctant to recruit or that parents were less likely to participate on days on which the video materials were used.

Table 2 summarizes the demographic characteristics of the participants in the two groups. There were no significant differences between those randomized to written material and those receiving written material as a partner in group I, or between those randomized to video or written in either of the two groups. Because of the unequal recruitment in the second group, there was a slight difference in age between the two methods when both groups are combined. As anticipated, those in group II had less education than those in group I.

### *Immediate Knowledge of Those Who Received Educational Materials*

The subjects who reviewed the educational materials correctly answered an average of 86% of the questions about the health consequences of having CF and of being a CF carrier. Similarly, 86% of these respondents had adequate information about these effects. Eighty-five percent were aware that a person could still be a carrier even if he or she were not found to have one of the six mutations being sought. Thirteen individuals who failed to demonstrate either adequate information about health consequences or adequate understanding of the limitations of screening expressed interest in free screening. None of these individuals opted out of screening.

There was a significant gradient in understanding and knowledge with educational level. Figure 1 summarizes results for both types of materials for the percentage of the five factual questions about CF answered correctly as well as the percentage of respondents who had adequate information and adequate understanding, separately by educational strata. The responses of those who received the different educational interventions did not differ when adjusted for educational level. There was a significant association of educational level with the abso-

**Table 2**

**Demographic Characteristics of Participants in Groups I and II**

| Demographic Characteristics | Group I; R-W; $n$ = 71 | Group I; R-V; $n$ = 65 | Group I; P-W; $n$ = 102 | Group II; R-W; $n$ = 91 | Group II; R-V; $n$ = 17 |
|---|---|---|---|---|---|
| Mean age (years) | 30 | 32 | 30 | 27 | 29 |
| Gender (percent female) | 61 | 58 | 51 | 91 | 94 |
| Race (percent white) | 99 | 94 | 96 | 62 | 53 |
| Family history of CF (percent) | 13 | 15 | 14 | 1 | 0 |
| Heard of CF previously (percent) | 90 | 82 | 80 | 47 | 24 |
| Prior education (percent): | | | | | |
| Less than high school | 4 | 2 | 4 | 36 | 35 |
| High school | 6 | 5 | 6 | 25 | 35 |
| Some college | 14 | 22 | 22 | 27 | 24 |
| College grad | 24 | 25 | 29 | 8 | 6 |
| Postgraduate degree | 52 | 48 | 39 | 3 | 0 |

NOTE.—R-W = randomized to written materials; P-W = received written materials as partner; and R-V = randomized to video information.

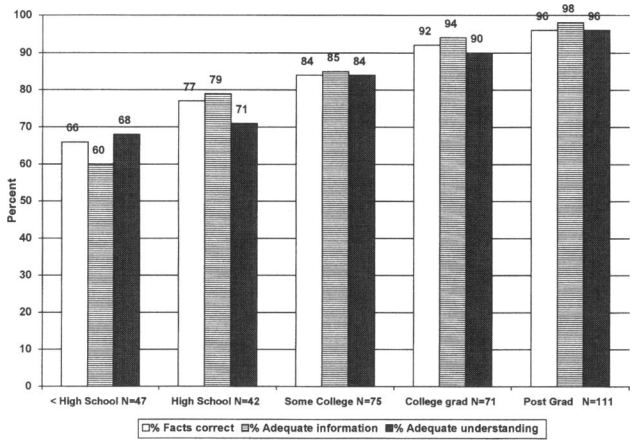


**Figure 1** Efficacy of educational materials, by education background.

lute number of questions answered correctly and percent of questions answered correctly among those attempted (both $P < .0001$), the number of questions attempted out of five ($P < .001$), and the adequacy of information and adequacy of understanding (both $P < .0001$). Nonetheless, 95% of all subjects found the information they received to be easy to understand.

Within each of the five educational levels, we also compared results for all five outcomes (adequate information, adequate understanding, number of questions answered, number answered correctly, and percent answered correctly) between (*a*) those randomized to written material versus those given written material as partners in group I; (*b*) those randomized to written material versus those randomized to video in group I, group II, and both groups combined; and (*c*) those receiving written material versus those receiving video material (in group I and in both groups combined). Two tests (<5% of all those performed) were statistically significant ($P < .05$), consistent with a chance effect. There were no consistent patterns in the significant results.

There was some evidence suggesting that those interested in screening learned more than those who were not interested in screening. This pattern was found in almost all educational strata when a comparison was made between those who accepted screening with those who did not pursue screening and between those in group I (who entered the study because of potential interest in screening) and those in group II.

Overall analysis using logistic or binary regression, as appropriate, found that there was no evidence for an effect of type of material after adjustment for educational level, submission of a sample, study group, or being randomized in the study as opposed to being a partner. All $P$ values were >.10, and most were >.50. When models were limited to those factors significant for a particular outcome, there was still no evidence for a significant effect of type of material.

### *Knowledge of Those Who Were Not Found to Be Carriers about Their Test Results and Their Implications*

Of the 238 people in Group I, 180 (76%) submitted blood samples for free CF carrier screening. One hundred sixty-two of these individuals received test results prior to the cessation of the data collection on September 30, 1994 and so were available for further analysis. Seventeen people were found to be CF carriers or to have CF. (The individuals with CF participated in this study, unbeknownst to us, to obtain free CF carrier screening for their partners.) As noted previously, the responses of these individuals and their partners are not included in the analysis described in this paper. Of the remaining 128 individuals who were in relationships in which neither partner was found to be a carrier, 110 (86%) returned the follow-up questionnaire. There were no statistically significant differences between those returning follow-up questionnaires and those who did not, in terms of (1) their family history of CF; (2) whether they had heard of CF before; (3) such demographic characteristics as age, race, and gender; and (4) the total number and percent of questions answered correctly and the adequacy of their information and understanding as assessed in the first questionnaire. The nonresponders, however, did have slightly lower educational backgrounds than responders ($P < .05$) The follow-up questionnaires were returned on average 3.5 mo after the original questionnaire was received, with a median time for return of 2 mo, and a range of 1 mo to >1 year.

Eighty-eight percent of the respondents correctly identified their own results, and 88% knew their partner's results. The remaining 12% stated incorrectly that they and their partners were not CF carriers. Eighty-nine percent of the respondents understood that while their risk of having an affected child with their partner was quite low (~1/100,000), the test results did not provide a guarantee. When looked at in the aggregate, the ability to answer questions correctly about the possibility of false-negative test results did not change before and after screening. Four individuals had better understanding after they received their test results, but 11 did worse. This difference was not significant ($P > .1$). Eight people believed that since they had not been shown to be carriers, they had no risk at all of having an affected child; three did not know their risk; and one did not answer the question. Eight of these 12 individuals with inadequate understanding had at least a college degree. Ninety-seven percent of the respondents had adequate information about CF on follow-up. Five people who initially had inadequate information improved, while only one who initially had adequate information declined. There was a statistically significant association of the respon-

dents' educational backgrounds with their ability to answer questions about their test results and their consequences.

## Discussion

After receiving our educational materials, the majority of people were able to answer questions correctly about the health effects of being a CF carrier and of having CF and about the possibility of false-negative test results in CF carrier screening. Similar results were obtained by investigators at UCLA using predominantly written materials (Tatsugawa et al. 1994). In our study, recipients of the written information appeared to learn as much as those who viewed the videotape. Based on consideration of the goals of informed consent and the findings of others, we concluded that our material should enable individuals to make informed choices about participating in carrier screening (Katz 1984; Myers et al. 1994). The ability to convey this information, particularly to those who ultimately decided to proceed with CF carrier screening, without relying on face-to-face counseling is reassuring since it may be problematic to rely on generalists to provide the comprehensive, nondirective counseling needed to enable people to make informed choices about tests that may affect their reproductive plans. Primary care providers generally are relatively ill informed about heritable disorders (Hofman et al. 1993). Even if they were knowledgeable, generalists may not devote their full attention to conveying information about genetic risks, because they must provide a wide array of care, of which advising about and testing for genetic disorders is only a small part. Moreover, recent surveys demonstrate that primary care practitioners are more directive in counseling about prenatal diagnosis and abortion than are genetic professionals (Geller et al. 1993).

Some aspects of our study design could tend to overestimate the efficacy of our materials in educating people about CF carrier screening. For example, the use of multiple choice questions for assessment could overestimate the educational efficacy of the written and video materials developed for this project since the ability to select the correct option among several does not necessarily mean that the individual can call the information to mind when needed (Faden and Beauchamp 1986). In addition, the majority of our subjects had at least some college education, which distinguishes them from the general population. A substantial proportion of these more highly educated individuals were employed in health care, while many others had some personal contact with a person who had CF. As a result, even though we did not assess baseline knowledge, it is reasonable to think that many of the respondents may have known something about CF and its inheritance prior to receiving our information.

On the other hand, the fact that these materials were the sole source of information about CF and carrier screening could result in an underestimation of the efficacy that these materials might have in clinical practice. In our studies, health professionals neither recommended that their patients participate nor provided clarification of any concerns or questions their patients might have. We specifically asked providers not to discuss any aspect of the study with their patients, in order to avoid biases that could be introduced by their different levels of expertise about and interest in CF carrier screening. If adopted for use, written or video educational materials would probably be more fully incorporated into the practitioners' care. People might learn more from these materials if they were offered by their health care providers and explained as well.

The fact that few people with lower levels of education were interested even in learning about free CF carrier screening (authors' unpublished information) led us to add a second study, the design of which also tended to result in an underestimation of the efficacy of these materials. To confirm our observation that the educational materials were less effective when used by people with less education, we asked parents who were bringing their children for care in a resident continuity clinic only to review the informational materials and to answer questions. We chose this population because the overwhelming majority of children in this clinic come from families of lower socioeconomic status and less formal education. The less educated individuals in this second study answered fewer questions correctly than did the participants in the main study. Perhaps the parents in the continuity clinic were not as attentive to these materials as the couples in the main study, because (1) they had children with them at the time they were reviewing the materials and answering our questions, and (2) they were not reviewing the educational materials to decide whether to have CF carrier testing.

Prior to screening, we did not ask the subjects about every aspect of CF and its inheritance. We queried the subjects about information regarding the health implications of having CF and of being a CF carrier and the possibility of false-negative test results because we believed these to be the facts most salient to an individual's decisions about undergoing carrier screening. We did not assess, prior to the person's receiving the test, ability to answer questions about the inheritance of the genetic disease at issue, such as whether both parents must be carriers in order to have a child affected with an autosomal recessive disorder. Our decision not to study their knowledge of the genetics of CF prior to their having carrier screening was deliberate. Assessing this knowledge may well be appropriate prior to performing genetic tests focused on the individual, since it is critical that the person who is found to be a carrier understand

that his or her partner must also have the test to get the most information. In our study, CF carrier screening was provided only when both partners agreed to testing. As a result, while the educational materials did contain information about the inheritance of CF, it seemed less pressing to assess couples' knowledge of these facts prior to testing, because the majority of couples in whom neither partner is a demonstrable carrier have a risk of ~1 in 100,000 of having an affected child. Instead, after screening, we provided specific risk information to each couple on the basis of the results of their tests for the six alleles and their family history. Their ability to answer questions about their specific risks was then assessed.

One of the most promising results of our study is demonstrated in the analysis of the follow-up questionnaires. The overwhelming majority of people who accepted CF carrier screening and who were not found to be carriers were able to answer questions correctly about the health implications of having CF or being a CF carrier and about their own reproductive risk well after they received their test results. There has long been concern and some evidence that people would not understand that CF carrier screening cannot detect all the mutations that can cause CF and so would believe that they had no risk of having an affected child if they were not found to have one of the six mutations sought (Caskey et al. 1990; Workshop on Population Screening for the Cystic Fibrosis Gene 1990; Statement of the ASHG on cystic fibrosis carrier screening 1992; U.S. Congress, Office of Technology Assessment 1992; Bekker et al. 1994) This fear appears to have been unfounded with regard to our population, since only 11% of the respondents made this mistake even though most of them never received face-to-face counseling. The fact that ~85% of those who returned follow-up questionnaires had at least a college education, however, suggests caution should be exercised in generalizing our findings regarding long-term retention of information to the general population.

It is good news that a substantial majority of subjects who received only written or video information were able, after receiving their test results, to give the correct answers to questions (1) about CF and carrier screening prior to screening and (2) about their own and their partner's risks of being a carrier and of having an affected child, particularly when compared with observations of the efficacy of face-to-face genetic counseling. Leonard et al. (1972), for example, interviewed 61 families who had received genetic counseling after having children with CF, phenylketonuria, and Down Syndrome, about their knowledge of genetics and concluded that "about of the parents had the kind of comprehension that could make the information helpful to them, whereas in about that understanding was flawed in some way and for about the genetic counseling had served little purpose" (Leonard et al. 1972, p. 436). Many other investigators since then have reported that many people are unable to remember information that was provided them during genetic counseling (Shaw 1977).

Even so, the inability of almost one-half of those with lower educational backgrounds to answer questions correctly prior to screening cannot be ignored. We developed the videotape specifically to reach people who might not be as comfortable with written information. Health educators often rate videotapes as a highly effective educational approach, especially for patients with less formal schooling (Solomon and DeJong 1989; Funnell et al. 1992; Magyari et al. 1994). Despite the fact that the script of our videotape was written at a sixth-grade level, the videotape was no more effective than the written handout in conveying information even among people with less than a high school education. A practical consequence of this finding is that physicians should realize that video is not a panacea for educating patients about genetic tests. Practitioners should consider carefully whether to devote the space and resources to these video educational materials, since written information seems to be as effective as video. Efforts to improve the provision of information about CF carrier screening should not focus exclusively on improving specific written pamphlets and videotapes but rather must include more general strategies to educate the general population and health care providers about genetics. Educational interventions developed for specific screening programs are likely to be far more effective when used by individuals who are more generally informed about genetics and its implications and in a setting that allows at least some time for addressing concerns people may have about screening.

It is unlikely, however, that an educational intervention can be developed that will be effective for everyone who might consider CF carrier screening. The challenge, then, is to decide on an appropriate ethical response to the possibility that some people will want testing even though they do not understand what is involved. In our study, since the subjects' knowledge was assessed before their blood samples were actually tested, those who did not demonstrate adequate knowledge were sent additional information, specifically tailored to the questions they got wrong on the questionnaire, and offered the opportunity to opt out of carrier screening. While such an intervention is feasible in the experimental setting, substantial questions remain about whether efforts should be made in clinical practice to detect which individuals do not appear to understand the implications of screening and to meet their educational needs. Such efforts have been made in experimental settings in the past (Lidz et al. 1984), but pretest assessment of knowledge represents a significant addition to current clinical prac-

tice. Pretest assessments of knowledge also present the dilemma of what to do when individuals fail to meet the criteria for adequate understanding. Requiring that people truly understand carrier screening before they participate means that only fully informed consent will suffice for medical decision making and will effectively deny testing to some people who desire it. While rational and informed decision making is a laudable goal, people do not make choices about their health care, child bearing, and other aspects of their lives in such a considered fashion. On balance, given the personal values at stake in carrier screening, the provider should attempt to ensure that patients understand the potential consequences of genetic testing. Conditioning carrier screening on a demonstration of a certain level of knowledge, however, goes too far.

It is distressing that 10% of those who accepted screening did not know or understand the implications of their test results. The impact of a low-risk couple's inaccurate belief that they cannot have an affected child is unlikely to make a difference in their behavior, however, since their residual risk is actually quite low. It is not clear how much difference people perceive between a risk of 1 in 100,000 and no risk at all. These misunderstandings nonetheless make clear that physicians need to ask their patients whether they have had genetic tests and to explore their understanding of the results.

What may be more important in limiting the impact of this dilemma, however, is the observation that CF carrier screening appears to be of interest primarily to people with higher educational backgrounds and those who have family histories of CF. The educational materials developed for this project appear to be "good enough" for the overwhelming majority of the people who actually use them. Certainly, those who accepted carrier screening and are not found to be carriers appeared to understand the implications of their test results even though they never received face-to-face counseling, a result that is better than has usually been observed in traditional genetic counseling. If CF carrier screening were to be offered to the general population, an acceptable strategy may be to provide these informational materials to all people who are interested and to alert health care providers about the need to provide additional attention to those individuals with less education who have no prior knowledge about CF. Our study also demonstrates that health care practitioners need to continue to be vigilant about the possibility that patients do not understand the results of genetic tests.

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## Appendix A

### Questions Assessing Subjects' Knowledge about CF and Carrier Screening

*Prescreening Questionnaire*

a. CF is a disease that mainly causes problems with the: (Check only one)
( ) Brain and spinal cord
( ) Lungs and digestion
( ) Liver and stomach
( ) Heart and blood vessels
( ) Don't know

b. Are people with CF usually mentally retarded?
( ) Yes
( ) No
( ) Don't know

c. Most people with CF do not live past (Check only one)
( ) Infancy
( ) Childhood
( ) Teenage years
( ) Young adulthood
( ) Don't know

d. Are young adults with CF
( ) Healthier than other young adults?
( ) As healthy as other young adults?
( ) Sicker than other young adults?
( ) Don't know

e. Are young adults who are CF **carriers**
( ) Healthier than young adults who are **not** CF carriers?
( ) As healthy as young adults who are **not** CF carriers?
( ) Sicker than young adults who are **not** CF carriers?
( ) Don't know

f. If your CF carrier test result says "You very likely are **not** a CF carrier," then: (Check only one)
( ) You are not a CF carrier
( ) You could still be a CF carrier
( ) You are a CF carrier
( ) Don't know

*Follow-up Questionnaire Exploring Understanding of Test Results*

a. My CF carrier test result shows that:
( ) I **am** a CF carrier
( ) I **very likely am** a CF carrier
( ) I **very likely am not** a CF carrier

( ) I **am not** a CF carrier
( ) I do not understand the test results
b. My partner's CF carrier test result shows that:
( ) My partner **is** a CF carrier
( ) My partner **very likely** is a CF carrier
( ) My partner **very likely is not** a CF carrier
( ) My partner **is not** a CF carrier
( ) I do not understand my partner's test results
( ) I do not know my partner's results
c. If you and your partner have a pregnancy in the future, what is the chance that you will have a child with CF?
( ) 1 in 4 (25%)
( ) Less than 25% but still high
( ) Possible, but low
( ) Possible, but very unlikely
( ) No risk (neither of us are carriers)
( ) Don't know

## Appendix B

### Information about Cystic Fibrosis (CF) and CF Carrier Testing

*What is Cystic Fibrosis (CF)?*

- Cystic Fibrosis (CF) is a disease. It causes people to have thick mucus in their lungs that makes them cough and causes lung damage over time. People with CF are often sick with lung infections and may need to be hospitalized. People with CF usually die from breathing problems.
- CF also causes poor digestion of food. Children with CF may have problems growing and gaining weight.
- CF is an inherited disease. Other people cannot catch CF from people who have it.
- CF is found mostly in white people. About 1 in every 2,500 white children has CF. CF is much less common in other racial groups. For example, only about 1 out of every 17,000 black children in the U.S. has CF.

*What Is It Like to have CF?*

- Most children with CF have problems from CF by the time they are one year old.
- When they are not sick with lung infections, children with CF can do things other children can do, including going to school and playing sports. CF does not change their intelligence.
- While some women with CF can have children, most men with CF are infertile.

*Is There Treatment for CF?*

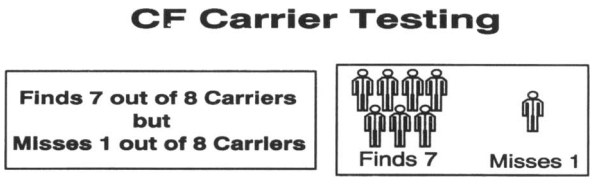
- There is no cure for CF now, but there are medicines and therapy that can help people with CF feel better and live longer. Treatment can take a lot of time each day and is expensive because several medicines are needed every day.
- Even with treatment, some children with CF die very young. Most people with CF live to be adults, but usually die before they are 40. In 1970, only half of all children with CF lived past 14, but now half live to 28 years of age. Treatment for CF is expected to keep getting better and someday there may be a cure.

*How Do You Get CF?*

- People have CF because they got a CF-causing gene from **each** parent. Genes contain information that tells the body how to work.

*What Is CF Carrier Testing?*

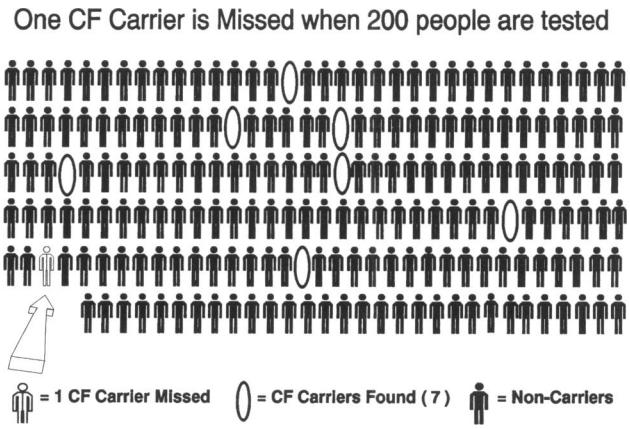
- CF carrier testing tells if a person has a CF-causing gene.
- There are many different types of CF-causing genes. It is **not** possible to test for **all** of them. In this study, we will test for several of the most common types.




- In order to do a CF carrier test, we need samples of your blood and your saliva (spit).

*You Will Get One of Two Test Results:*

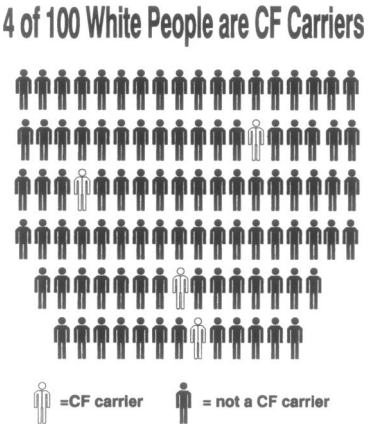
- If we find you have a CF-causing gene, you **are** a CF carrier.
- If we do **not** find that you have a CF-causing gene, you will be told that "You **probably** are **not** a CF carrier." There **still** is a 1 in 200 (%) chance that you **are** a CF carrier. Carrier testing **cannot guarantee** that you are not a CF carrier.



- People who have one CF-causing gene and one CF-preventing gene are called **CF carriers.** CF carriers do **not** have CF and have no personal health problems from their one CF-causing gene.
- If **one** parent is a CF carrier and the other parent is not, then their children almost never have CF.
- If **both** parents are CF carriers, then each of their children has a 1 in 4 chance of having CF.

*Could I Be a CF Carrier?*

- About 4 in 100 white people in the U.S. are CF carriers. This means that most white people (96 out of 100) are not CF carriers and cannot pass a CF-causing gene to their children.




*Why Would a Couple Want CF Carrier Testing?*

- A couple might have CF carrier testing together to find out **their** chance of having a baby with CF. This testing may be more helpful for whites, since CF is more common in whites than in other racial groups. CF carrier testing is also helpful for people who have relatives with CF.
- In most couples, we will not find a CF-causing gene in either person. **Together,** these couples have a very low chance (about 1/100,000) of having a baby with CF.
- Sometimes, we will find that **one** partner in a couple **is** a CF carrier. Each of their children has a low, about 1 in 640, chance of having CF. These couples will be given more information.
- Occasionally, we will find that **both** partners **are** CF carriers. Each of their children has a 1 in 4 chance of having CF. These couples will be offered more information, including information about prenatal diagnosis of CF. If they choose prenatal testing and the baby has CF, then they could stop the pregnancy or continue the pregnancy and start treating the baby right after birth.
- The decision about whether or not to be tested is up to each couple. Some may want this information, but others may not.

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