Chapter 3

Cystic Fibrosis Carrier Screening: Policies and Practices

Prospects of routine cystic fibrosis CF carrier screening polarize people. Everyone agrees that persons with a family history of CF should have the opportunity to avail themselves of CF mutation analysis, yet controversy swirls around using the same test in the general population. This polarization is illustrated in the written comments of two survey participants.

> NO to widespread screening! Must be close to 100 percent detection for all CF mutations before it can even be considered.
>
> **Let's go with screening!** I can't believe we are not halfday through a pilot program by mid 1991.

As described in the full OTA report (18), proponents of a measured approach to CF carrier screening express concern about several issues that might be raised if CF carrier screening becomes routine, such as the use of genetic information by insurance companies to set rates or deny coverage, and concerns that market pressures will drive widespread use of tests before the potential for discrimination or stigmatization by other individuals or institutions is assessed. Also expressed are questions about the adequacy of quality assurance for DNA diagnostic facilities, personnel, and the tests themselves. Still others also wonder whether the current number of health care professionals in genetics can handle a swell of CF carrier screening cases, let alone cases of other genetic conditions arising from increased knowledge from the Human Genome Project. Finally, the extraordinary tensions in the United States about abortion affect discussions about CF carrier testing and screening.

In summer 1991, OTA asked genetic counselors and nurses in genetics to provide data regarding their experiences concerning CF carrier screening as a means to judge the validity of these concerns. The questionnaire was designed to gather data on the frequency of DNA analysis for CF carrier status and trends over time, clinic policies regarding CF carrier screening, counseling and clinical practices regarding CF carrier testing and screening, and sources influencing the development of, and policies and procedures related to, CF mutation analysis. Survey participants were also asked their opinions about who should conduct carrier screening, in what settings, and on what target population(s). Respondents were encouraged to rank the most important issues to be addressed before embarking on a large-scale screening program.

The data in this chapter are specific to CF carrier screening. Data regarding third-party reimbursement for DNA-based tests are presented in chapter 2, along with general demographic data concerning the survey respondents and their clientele and clinical settings.

POLICIES AND PRACTICES, SUMMER 1991

Survey participants were asked to consider three issues. First, what is their opinion or the policy of their institution about the appropriateness of CF carrier screening at this time? Second, what are the current logistics of providing DNA-based tests for CF carrier status-i.e., once a decision had been made to offer CF mutation analysis, which mutations are analyzed, and how are those individuals to be tested identified or contacted? Third, survey participants were asked to estimate whether requests for DNA-based tests for CF had changed since the tests' development in 1989.

Policies on Cystic Fibrosis Carrier Screening

Currently, it is standard practice to offer CF carrier tests to individuals who have a positive family history of CF (6,16,18). An unaffected sibling of an individual with CF has a 2 in 3 likelihood of being a CF carrier. A consanguineous uncle or aunt of an individual with CF has a 1 in 2 likelihood of being a carrier. A first cousin of an individual with CF has a 1 in 4 likelihood of being a carrier (table 3-l).

As of the summer of 1991, most genetic counselors and nurses in genetics did not offer unsolicited CF mutation assays to individuals with a negative family history. A large majority of survey respondents use medical journals and other professional sources to obtain information regarding new advances in human genetics (table 3-2), and the American Society of Human Genetics (ASHG) and the National Institutes of Health (NIH) published policy documents in 1990 discouraging CF carrier

Table 3-I—A Priori Carrier Risks for Cystic Fibrosis

Negative family history	
Caucasian	1 in 25 (4%)
African American	1 in 60 to 65 (1.5 to 1.7%)
Asian American	1 in 150 (0.7%)
Hispanic American	1 in 46 (2.2%)
Positive family history	
Parent of child with CF	1 in 1 (100%)
Sibling with CF	2 in 3 (67%)
Aunt or uncle with CF[a]	1 in 3 (33%)
First cousin with CF	1 in 4 (25%)
Niece/nephew with CF	1 in *2 (50%)*

[a]Consanguineous.

SOURCE: Office of Technology Assessment, 1992.

Table 3-2-Sources of Information About New Advances in Human Genetics

Human genetics	Percent indicating yes
Medical journals	96
Professional colleagues	94
National inferences	83
American Society of Human Genetics	82
National Society of Genetic Counselors	80
State or regional conferences	71
Grand rounds	44
Lay press	37
Continuing education courses	35
Literature from biotechnology companies or commercial firms	35
Other	8

SOURCE: Office of Technology Assessment, 1992.

screening (6,16).[1] Seventy-six percent of respondents stated that they were familiar with the 1990 ASHG statement. Thirty-five percent were familiar with the NIH statement.

OTA's survey of genetic counselors and nurses revealed that 53 percent of respondents believe that CF carrier tests should only be offered to individuals with a positive family history of CF and not to those with a negative family history. Twenty-one percent felt that CF carrier tests should be offered to individuals with no family history. The most frequently cited reasons for making tests available to individuals regardless of family history were to reduce anxiety or increase patient autonomy. In the words of one counselor, ''DNA screening is a personal issue, different in every case. What one person or family feels may be quite different from that of another person or family in any given genetic disorder with any given family history.' Twenty-six percent of respondents were uncertain as to whether they should provide CF carrier screening where family history is negative.

When asked about their likelihood of introducing the topic of CF carrier tests during a counseling session, 82 percent of respondents stated that they would seldom, if ever, do so to all patients or families (table 3-3). Seventy-three percent would seldom, if ever, discuss it with pregnant women seeking prenatal diagnosis unless there was a family history of CF in which case, 90 percent would almost always bring it up during counseling.

When asked whether their institution or clinic had a specific policy regarding CF carrier screening, 33 percent of genetic counselors and nurses responded in the affirmative. Of those responses, 70 percent stated that it is the policy of their clinic or organization to offer CF carrier tests only to those with a positive family history (table 3-4).

The overall lack of policies for CF carrier screening apparently stems from the fact that, in general, explicit and official policies for clinical practices were not routine at the majority of facilities. When asked whether their group or unit had

Table 3-3-Likelihood of Introducing the Topic of DNA Testing for Cystic Fibrosis

Patient population	Predominant response	(Percent)
All patients/families	Seldom if ever	(82)
Pregnant women seeking prenatal diagnosis	Seldom if ever	(73)
Coupies/individuals with a family history of CF	Almost always	(90)
Caucasian coupies/individuals with a negative history of CF	Seldom if ever	(65)
individuals/famiiies who inquire about CF	Almost always	(80)
Selected coupies/individuals	Seldom if ever	(72)

SOURCE: Office of Technology Assessment, 1992.

Table 3-4-Specific Policies Regarding DNA Testing for Cystic Fibrosis

Policy	Percent
Offer to all regardless of family history	14
Offer only to those with a positive family history	70
Provide to those with no family history upon request if informed consent is obtained	16

SOURCE: Office of Technology Assessment, 1992,

[1] In 1992, ASHG's leadership issued a revised statement that CF mutation analysis ' 'is not recommended' fOr those without a family history of CF, but it has not yet been published (1,18).

official policies and procedures for other issues in genetics, 21 percent reported they have policies regarding DNA storage, 42 percent have policies in place concerning prenatal diagnosis for sex selection, 37 percent have policies regarding cases of nonpaternity, and 28 percent adhere to policies regarding confidentiality and Huntington disease testing.

Criteria for Cystic Fibrosis Carrier Screening

Sixty-five percent of survey participants felt strongly that there is an optimum rate of detection that should be reached before they would feel comfortable offering CF carrier screening, as compared to 14 percent who felt there is not and 21 percent who were uncertain. Of those who felt there is an optimum rate of detection, nearly half (46 percent) said that 95 percent test sensitivity should be required before proceeding with widespread screening. Twenty-five percent believe test sensitivity should be even higher, with 4 percent stating that it should be 100 percent (figure 3-l).

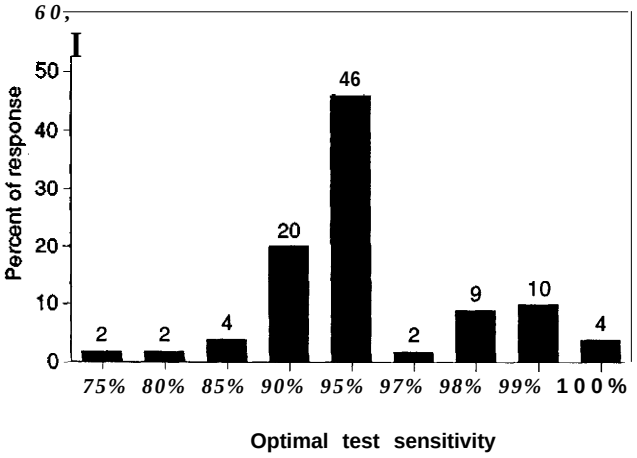
However, survey respondents ranked the availability of adequate counseling and an adequate system of referral for individuals who test positive as slightly more important criteria for CF carrier screening than test sensitivity (table 3-5). Guarantee of informed consent also was mentioned as necessary for implementation of large-scale CF carrier screening.

Perhaps the point on which there was greatest consensus among the respondents is on the issue of autonomy and choice in screening. There are no mandatory genetic screening programs of adult populations in the United States. Ninety-nine percent of survey participants responded that CF carrier screening should be voluntary and never mandatory.

Practices Regarding DNA-Based Cystic Fibrosis Carrier Tests

When asked about the frequency of requests for DNA testing or screening for CF carrier status during the 6-month period from January to June 1991, most respondents reported occasional requests (figure 3-2). When asked to compare this time period with the previous 2 years, nearly half indicated a small increase in the number of requests and a quarter noted a large increase in requests (figure 3-3). The survey did not distinguish whether the requests were carrier tests for individuals known to be at risk by virtue of family history or carrier screens for individuals with no known family history of CF

Figure 3-l—Opinions on Optimal Rate of Detection



SOURCE: Office of Technology Assessment, 1992.

Table 3-5-Minimal Criteria for Cystic Fibrosis Carrier Screening Protocol

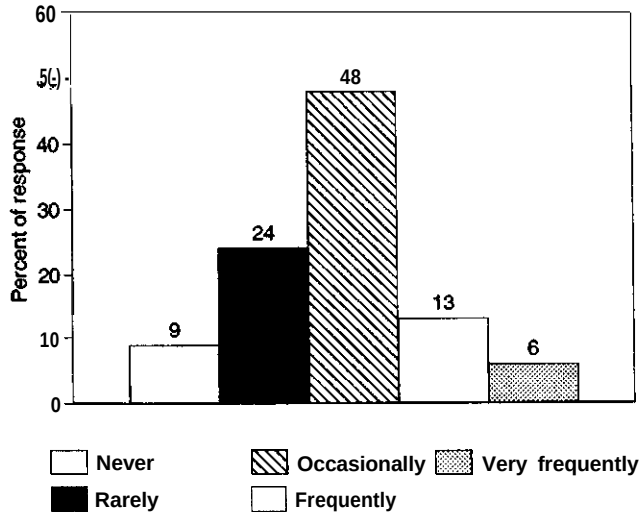
Question: What do you feel should be the minimum criteria for CF carrier screening protocol)?	
Criteria	**Percent[a]**
Provision of adequate counseling	40
Adequate system of referral in place	37
Improved test sensitivity.	35
Guarantee of informed consent	32
Availability of educational materials.	18
Only offer to families with a positive history of CF	15
Must be voluntary.	14
Reasonable cost or payment	12
Protection of confidentiality	12

[a]Percentages do not add to 100; respondents could reply with multiple answers.

SOURCE: Office of Technology Assessment, 1992.

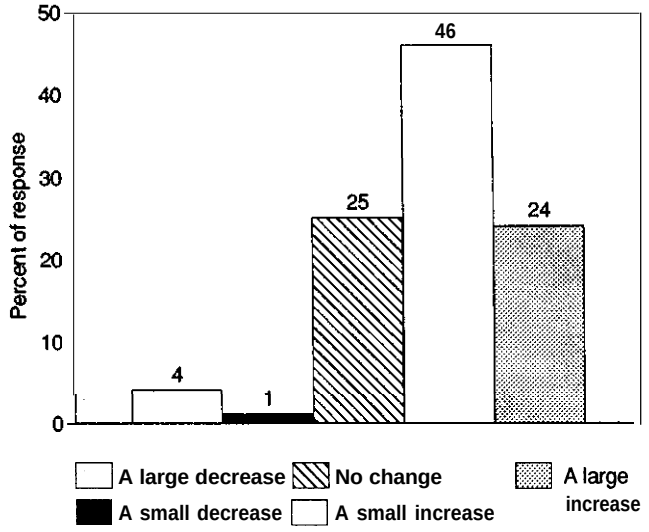
Although 55 percent of survey participants responded that a CF treatment center exists at their institution, 86 percent reported that they do not provide genetic counseling through that facility. Several respondents noted that this is the choice of the CF treatment provider, not necessarily the genetics unit. Because OTA did not survey CF treatment centers, it is not known to what extent CF families are informed of, offered, or request carrier testing. The data do show, however, that most families who have a child with CF are not routinely

Figure 3-2—Frequency of Requests for Cystic Fibrosis Carrier Screening/Testing, January-June 1991



SOURCE: Office of Technology Assessment, 1992.

Figure 3-3-Comparison of Requests for cystic Fibrosis Carrier Screening/Testing Between January-June 1991 and Past 2 Years



SOURCE: Office of Technology Assessment, 1992.

seen in genetics service settings, and few counselors have routine contact with CF families.

Encouraging known carriers to notify consanguineous relatives (e.g., siblings and first cousins) provides economic and pragmatic benefits because it can detect a larger percentage of at-risk couples (18); testing those known to be at higher risk because of family history is more effective than screening those with unknown risk. In reality, complex psychological factors enter when family members of individuals with CF contemplate screening, and it cannot be assumed that all will want to be tested.

For this type of carrier identification to work, those providing health care and counseling to CF families will have to actively participate in referrals of relatives to genetics centers, an uncommon practice, according to OTA's data. Fewer than 10 percent of respondents reported contacting previously identified CF families with whom they had had contact about the availability of CF mutation analysis.

For those respondents whose institutions are engaged in CF carrier testing or screening, direct DNA mutation analysis is the most common approach (table 3-6). In the recent past, the sensitivity of the carrier test was limited to the DF508 mutation. All respondents involved in analyzing CF carrier status assay for the DF508 mutation. But roughly 74 percent indicated that they also test for at least one other mutation, most commonly four others, G551D, R553X, G542X, and N1303K (table 3-7). At the time the survey was done, the mutation that accounts for 60 percent of CF mutations in Jewish persons of

Table 3-6—Types of Genetic Analyses Provided for Cystic Fibrosis Screening/Testing

Procedure	Percent response
Direct mutation analysis	67
Prenatal DNA analysis	63
DNA linkage analysis	61
DNA haplotyping	56
Staging of studies	37
DNA banking	31
Fetal intestinal enzyme analysis	28

SOURCE: Office of Technology Assessment, 1992.

Table 3-7-Cystic Fibrosis Mutations Routinely Analyzed

Mutation	Percent response
DF508	100
G551D	77
R553X	76
G542X	71
N1303K	70
Other	79

SOURCE: Office of Technology Assessment, 1992.

Central and Eastern European descent (Ashkenazic Jews), W1282X, had not been found.[2]

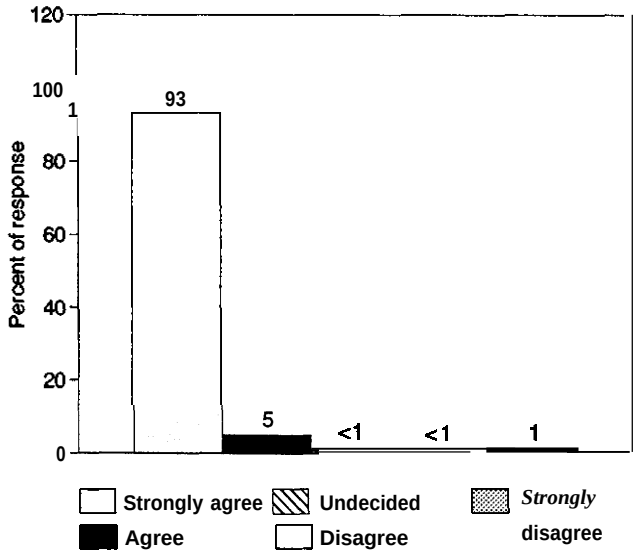
Respondents report an almost even split between commercial and university-based laboratories as the facility performing their CF mutation assays (45 percent and 48 percent, respectively). Most centers send the sample offsite (76 percent), frequently to a laboratory greater than 150 miles away.

Finally, although the need for professional and public education was cited as critical for the implementation of widespread carrier screening, few genetic counselors and nurses in genetics reported spending professional time engaged in either activity. For those respondents who do, an average of 3 hours per week devoted to educating health professionals and 1 hour per week on educating the general public was reported (ch. 2). For CF carrier screening, specifically, 8 percent of genetic counselors and nurses had developed, or were in the process of developing, educational materials relevant to DNA tests for CF mutation.

PREFERRED STRATEGIES AND PROTOCOLS

The importance of informed consent, careful presentation of counseling, and confidentiality have long been recognized as essential components of genetic testing and screening (9). Respondents strongly agreed that genetic counseling should precede DNA tests for CF carrier status regardless of family history (figures 3-4 and 3-5). Geneticists, perhaps more than any other medical specialty, have advocated a nondirective approach to counseling and have a strong commitment to patient autonomy (3). Further, a history of concern exists about the delivery of genetic information by health professionals used to a more directive approach (7). This concern has been played out in the debate over maternal serum alpha-fetoprotein (MSAFP) screening and is a factor in the reluctance of the clinical genetics community to rush toward widespread screening for any disease (18). For example, as part of the debates surrounding MSAFP and CF carrier screening, concern has been voiced about informed consent—in particular, that tests would be available to primary care practitioners who might incorporate

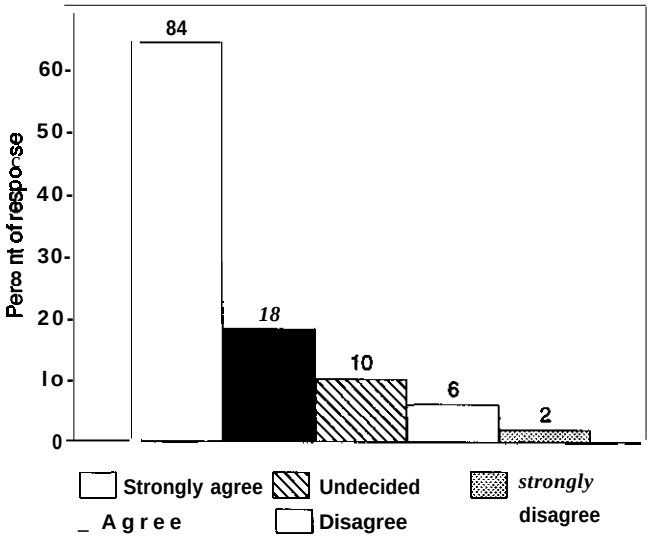
Figure 3-4-Opinions Regarding Genetic Counseling of Individuals with a Positive Family History



Genetic counseling should precede DNA testing for CF when there is a positive family history.

SOURCE: Office of Technology Assessment, 1992.

Figure 3-5-Opinions Regarding Genetic Counseling of Individuals with a Negative Family History



Genetic counseling should precede DNA testing for CF when there is a negative family history.

SOURCE: Office of Technology Assessment, 1992.

2 When the survey was fielded, test sensitivity was 75 to 85 percent, depending on race and ethnicity. Today, most commercial and university laboratories examine DF508 and 6 to 12 additional mutations, and taken together these mutations comprise 85 to 90 percent of CF mutations in U.S. Caucasians (95 percent in Ashkenazic Jews).

the assay into their practice without considering the informed consent requirements usually adhered to in genetics practices. Seventy-nine percent strongly agree that informed consent prior to CF carrier screening is a necessity (figure 3-6).

In addition to informed consent, prescreening education for clients is imperative. Information regarding an individual's a priori risk, types of tests available, and uncertainties in risk assessment based on screening results are important for potential screenees to understand. When asked if educational materials can provide adequate information about CF carrier screening, 44 percent disagreed or strongly disagreed with that concept (figure 3-7).

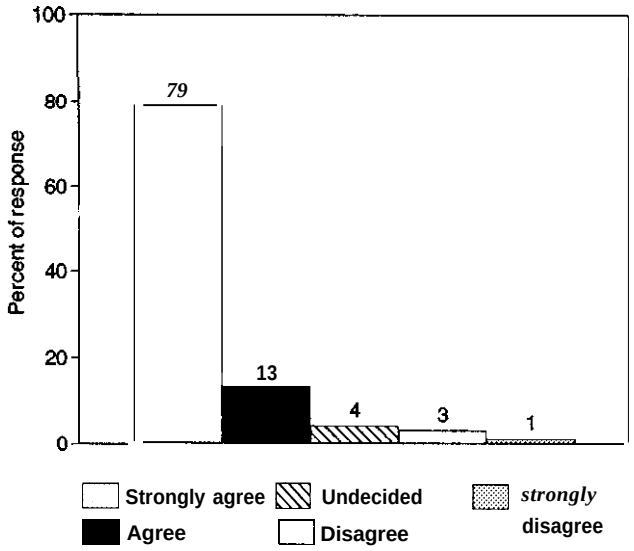
Who Should Provide Cystic Fibrosis Carrier Screening?

Concern about the complex nature of some genetic information and the need in some cases for post-test counseling leads many human genetics professionals to advocate restricting CF carrier screening primarily to the human genetics community. Pretest education, felt many respondents, can be offered by a wide range of professionals (figure 3-8), but organizing CF carrier screening should be provided by genetic specialists (table 3-8). Nearly 82 percent of the respondents surveyed by OTA said the human genetics community should be the primary organizer of CF carrier screening programs (table 3-8). Also mentioned were State or local health departments (59 percent) and primary caregivers (27 percent). Over 89 percent believed CF population screening should be provided in genetics centers, but 59 percent thought CF carrier screening could also be provided in the primary care setting or organized, community-wide programs (53 percent) (table 3-9). Concern about the sometimes difficult nature of communicating risk information regarding CF even for experienced genetic centers—has led some in the clinical genetics community to caution against rapid movement to routine CF carrier screening (2). In the words of one respondent:

> Counseling should not be left to hurried family practitioners or OB's [obstetrician/gynecologists], who routinely spend less than 15 minutes with each patient.

As noted in chapter 2, most counselors and nurses spend little to no time on professional education or general public education in schools and communities. Thus, the majority of people will rely on their primary care provider for preliminary, if not most,

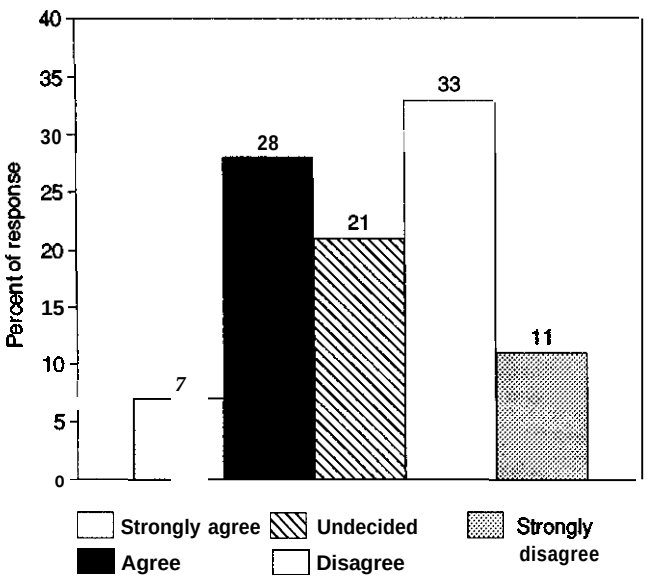
Figure 3-6—Opinions Regarding the Need for Informed Consent Prior to Cystic Fibrosis Carrier Screening



Informed consent prior to CF carrier screening is a necessity.

SOURCE: Office of Technology Assessment, 1992.

Figure 3-7-Opinions Regarding the Use of Educational Materials as a Source of Information About Cystic Fibrosis Carrier Screening



Educational materials (culturally sensitive and understandable) can provide adequate information about CF carrier screening.

SOURCE: Office of Technology Assessment, 1992.

Table 3-8—Preferred Organizations for Implementation of Voluntary Cystic Fibrosis Carrier Screening

Organization	Yes	No[a]
	(percent)	
Human genetics community	82	15
State or local health department	59	39
Voluntary health organizations	30	67
Primary caregivers	27	71
Medical societies	17	81
Federal Government	15	82

[a]3 percent gave no response

SOURCE: Office of Technology Assessment, 1992.

Table 3-9-Preferred Sites for Cystic Fibrosis Carrier Screening Programs

Site	Yes	No[a]
	(percent)	
Genetics centers	89	7
Primary care setting	59	37
Community-wide	53	43
Public health department	48	49
Public schools	14	83
Workplace	9	87

[a]3.5 percent gave no response

SOURCE: Office of Technology Assessment, 1992.

genetic information (18), and many survey respondents said primary care providers and public health departments should play an active role in educating the public about DNA tests for CF carrier status (figure 3-8). Health care provider and community-wide genetics education will become increasingly important, as will the interaction of genetic specialists with other health professionals and the public.

Who Should Pay for Cystic Fibrosis Carrier Screening?

When asked who should pay for screening, 80 percent of respondents ranked third parties as the primary source of payment (table 3-10). Self pay was ranked second, and employers ranked last. Additionally, some participants noted that if screening ever became mandatory, as in many State newborn screening programs, the State or Federal Government should be responsible for payment.

Strategies for Screening Various Populations

Two key considerations in deciding how routine CF carrier screening is best implemented are the clinical settings in which it will take place and the target populations. Delineation of a target group (or groups) determines other elements such as location, educational approach and tools, time, format, types of counseling, facilities, and publicity.

The NIH statement on CF carrier screening emphasized the importance of preconceptional screening (16). Most pilot projects in the United Kingdom are directed at preconceptional populations (18). One program in Canada targets high school students (11).

Newborn Screening

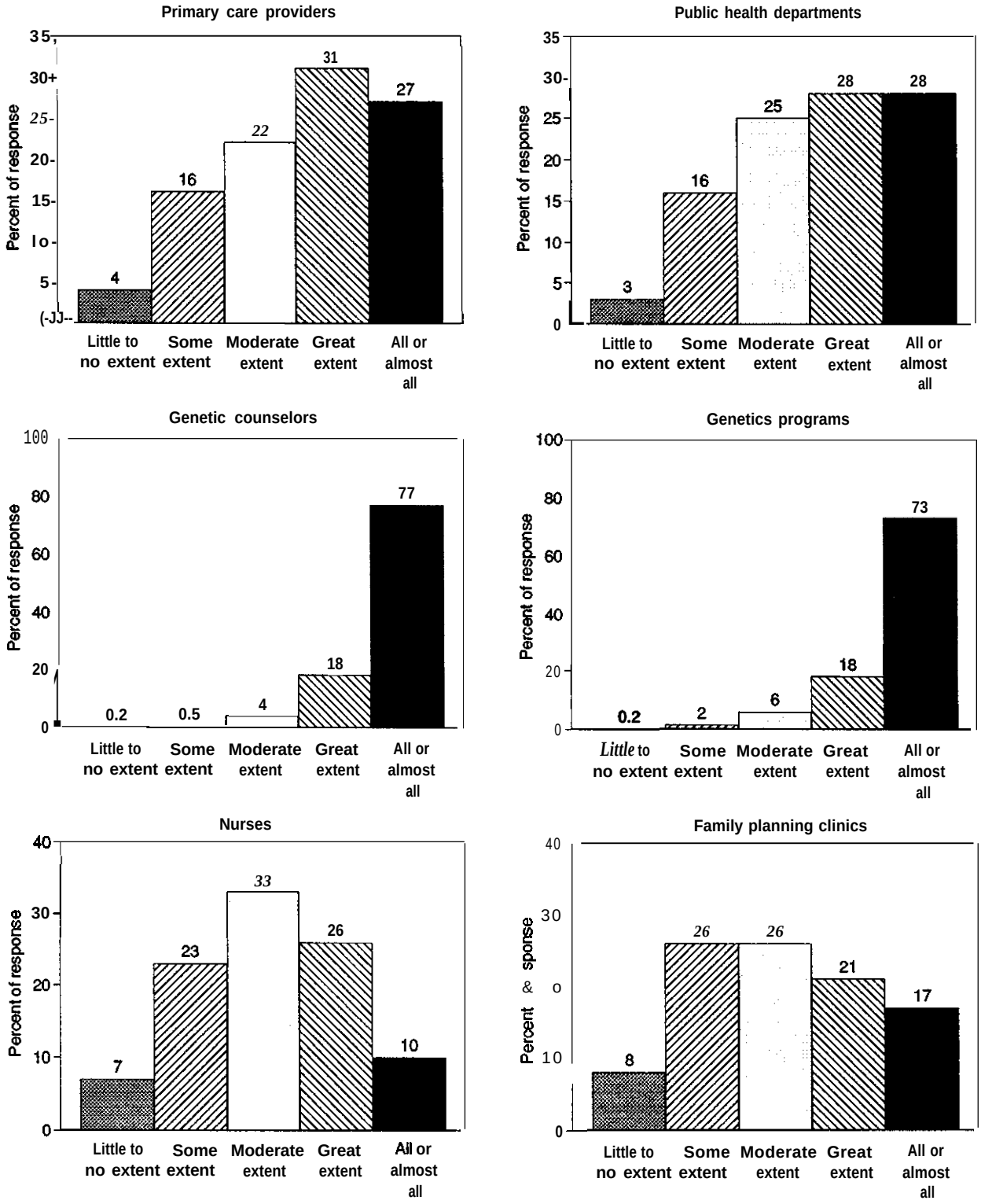
Numerous newborn screening programs exist for genetic disorders such as sickle cell anemia and phenylketonuria. These are programs intended to screen for the presence of disease, although some can also detect the carrier status of the newborn. Using the immunoreactive trypsin assay, Wisconsin has performed statewide neonatal screening for CF disease since 1985, and primary care physicians have been cooperative in referring screened patients to designated CF centers for followup (14). But even newborn screening for CF disease is not without controversy. Evidence of heightened anxiety and disrupted maternal-infant bonding have been reported in cases of false-positive diagnoses (4).

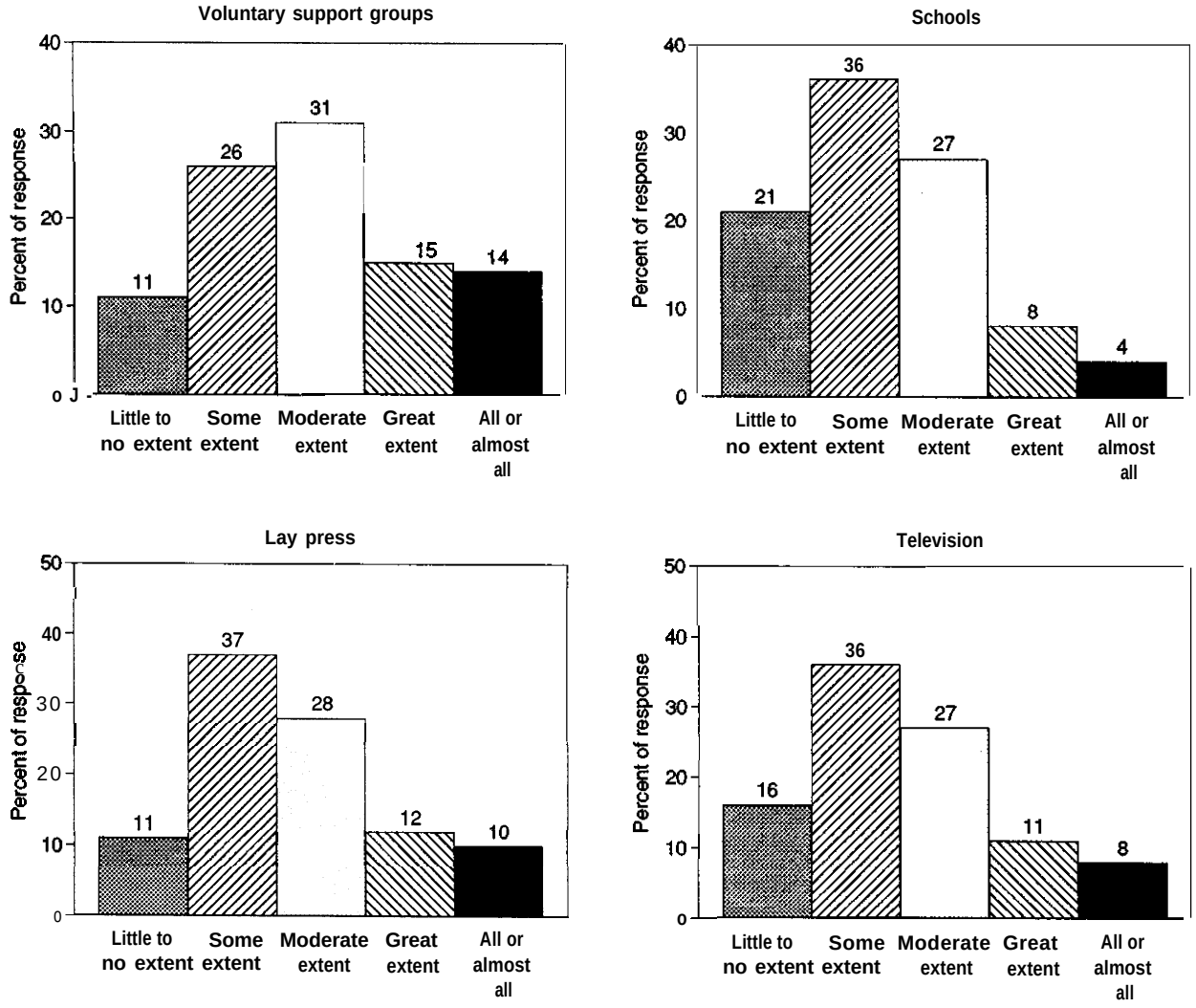
For at least two reasons, many believe that newborn screening is an inappropriate and inefficient mechanism for carrier detection. First, newborns determined to be carriers must be tracked through their reproductive years to ensure they are aware of their carrier status. Second, detection of newborn carriers might unnecessarily raise the anxiety level of parents. Thus, newborn screening for CF carrier status is not generally viewed as acceptable (15). This survey revealed that 33 percent of genetic counselors and nurses in genetics believed the newborn population would be an appropriate target group for widespread CF carrier screening (table 3-11).

Adolescent Preconceptional Screening

Some geneticists advocate carrier screening at the high-school level (11). A recent nationwide survey of American attitudes about, and knowledge of, genetic tests showed better knowledge and more positive attitudes in younger populations (17). Studies of pregnant women known to be carriers of a hemoglobinopathy gene have shown that age is a predictor of postcounseling knowledge-younger women (and adolescents as young as 12 years old) are more likely to understand genetic information (13). While not routinely done in the United States,

Figure 3-8-Extent to Which Various Groups Should Be Involved with Cystic Fibrosis Pretest Education





SOURCE: Office of Technology Assessment, 1992.

high-school screening programs have been conducted in Montreal, Canada for some time. For any disease where screening is done in childhood or adolescence, however, the benefits of such screening, including savings in resources or anxiety, must be balanced against the potential problems, such as the possibility that an adolescent will be falsely assigned to a low-risk group because of poor test sensitivity (thereby obviating further screening), or the possibility of psychosocial harm to the child as a result of identified carrier status (9).

Adolescents were not considered an appropriate target by the genetic counselors and nurses surveyed by OTA (table 3-11). Less than one-fifth felt individuals ages 13 to 18 years should be screened; only 6 percent responded that children ages 2 through 12 years should be screened.

Adults—Preconceptional or Prenatal?

One debate surrounding CF carrier screening focuses on whether the goals are best accomplished by targeting preconceptional adults or pregnant women. These approaches are not necessarily mutually exclusive. Many believe, however, that the receipt of troubling information during pregnancy is not desirable, and that it would be better for

Table 3-10-Who Should Pay for Cystic Fibrosis Carrier Screening?

Rank order
1. Third parties
2. Self pay
3. State, city, or county
4. Federal Government
5. Employers

SOURCE: Office of Technology Assessment, 1992.

Table 3-1 l—Target Populations for Cystic Fibrosis Carrier Screening

Population	Yes	No[a]
	(percent)	
Adults in reproductive years.	88	8
Prenatal	75	22
Pregnant women or "couples"	66	31
Newborns	33	63
Children ages 13 to 18	19	78
Children ages 2 to 12	6	91
Adults in post reproductive years.	3	94

a3percent had no response in each category.

SOURCE: Office of Technology Assessment, 1992.

individuals to know their risks before getting pregnant (12). Others argue that individuals not facing a pregnancy are not motivated to seek or use information on their carrier status, but will wait until they are either planning a family or starting a family before viewing such information as useful (5).

CF carrier screening offered as part of primary health care rather than prenatal care is likely to encourage preconceptional CF carrier screening. For most individuals, however, the first real opportunity for carrier screening takes place postconception (8). In the future, the primary responsibility for providing CF carrier screening might reside with the obstetrician, as has happened with MSAFP screening. Sixty-six percent of respondents to OTA's survey identified pregnant women or couples as the appropriate target population for CF carrier screening, yet 88 percent more generally identified adults in their reproductive years as the appropriate target group (table 3-11). While most respondents state that the *ideal* target population for carrier screening is the preconceptional adult, in reality, the first target population is likely to be the prenatal population because it has been the traditional entry point into genetic services for many people and comprises the largest population served by genetics centers (table 3-12).

Table 3-1 2—Frequency of Patients Seen by Major Areas of Clinical Practice

Area	Predominant response
Prenatal genetics.	Very often
Pediatric genetics	Sometimes
Adult genetics	Sometimes
Teratogen exposure	Sometimes
Reproductive loss	Sometimes
Specialty disease(s) clinics	Sometimes
Newborn screening	Seldom if ever
MSAFP screening followup.	Often
Carrier screening	Sometimes

SOURCE: Office of Technology Assessment, 1992.

PROFESSIONAL CAPACITY

Another issue in considering widespread carrier screening for CF is whether there are enough adequately trained health professionals to handle the volume of tests. One study estimated that a minimumof651,000 counseling hours would be required annually if the maximum estimate of 6 to 8 million preconceptional couples are screened for CF carrier status (19). Considering the current number of practicing genetic counselors in the United States today, this translates to 17 weeks per year from each genetic counselor to serve solely CF-related clients. On the other hand, another estimate suggests the supply of genetic specialists could absorb routine carrier screening for CF sickle cell anemia, hemophilia, and Duchenne muscular dystrophy, assuming that obstetricians or other primary care physicians perform the screening on pregnant women, with referral of those with positive results to genetics professionals (10).

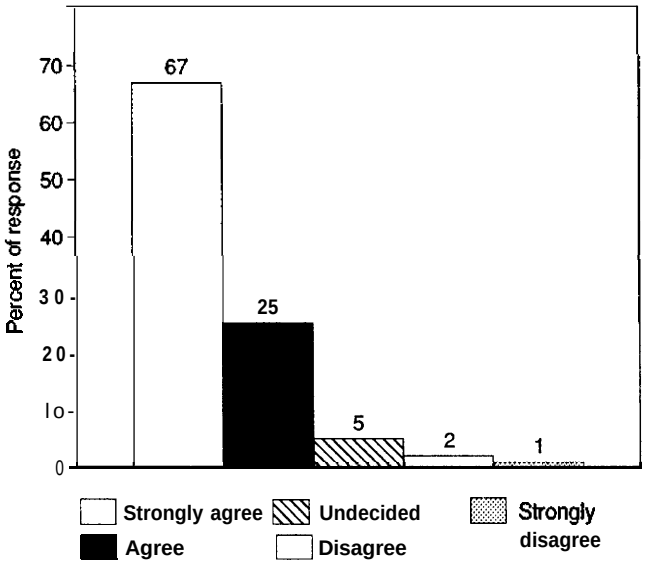
The counselors and nurses surveyed by OTA estimate pretest counseling time for CF carrier status would range from about 45 minutes to over 1 hour, depending on family history (table 3-13). It is unclear to what extent increased demand for CF carrier screening would strain the current system. Current estimates undercount the number of health care professionals who practice genetic counseling and assume that counseling would always be provided in a clinical genetics setting by board-certified or board-eligible counselors. Such estimates also ignore the role that aggressive public education can play in improving pretest knowledge. Improvements in public education could result in dramatically less time required in formal counseling, as could reliance on health professionals not formally trained in genetics.

Table 3-1 3-Time Required for Genetic Counseling for Various Conditions

Condition	Time (minutes/visit)	Number visits
Prenatal counseling for advanced maternal age	54	1
Positive family history for neural tube defects	57	1
Elevated MSAFP screen	55	1
Couple with newly diagnosed (Tri21) Down syndrome child	78	2
Couple with 14/21 translocation Down syndrome child	73	2
Carrier testing for Duchenne muscular dystrophy	75	2
Newly diagnosed case of neurofibromatosis	70	2
Newly diagnosed CF family	59	2
Carrier testing for CF with a positive family history	70	2
Carrier testing for CF with a negative family history	44	1

SOURCE: Office of Technology Assessment, 1992.

Figure 3-9—Opinions Regarding the Need for More Genetic Counselors



A need for more genetic counselors exists.

SOURCE: Office of Technology Assessment, 1992.

Table 3-14-Strategies for Implementation of Widespread Cystic Fibrosis Carrier Screening

Question: What strategies have you considered implementing if widespread screening for CF becomes a reality?[a]

Strategy	Percent
Plan professional education activities	40
Develop videotapes for patient education	21
Conduct public education	15
Arrange for group counseling sessions	14
Administrative changes in clinics to handle patient load.	13

a237 of the 43 I respondents gave no response.

SOURCE: Office of Technology Assessment, 1992.

Two-thirds of respondents strongly agreed that a need for more genetic counselors exists (figure 3-9). A few respondents raised the possibility of training ''single-gene' counselors to assist in the increased workload, although others expressed concern about this prospect, as taking a family history can reveal other genetic conditions that might not be detected by an individual trained to handle one genetic disorder (18). Still other respondents mentioned the need for more professional education of health care providers who might be in the position of administering such tests, and many survey participants noted that all groups of health care providers should be involved after appropriate training and education. Noted one genetic counselor, "Once screening is close to 100 percent sensitive, doctors and nurses could easily be trained to provide the necessary counseling.

When asked what strategies would be considered to alleviate the projected increase in workload should widespread CF carrier screening occur, 55 percent gave either no response or reported that they had not yet developed any. Of those who had considered or developed strategies, 40 percent said they would plan professional education activities to educate other health professionals, 21 percent would develop videotapes for patient education, 15 percent said they would conduct public education, and 14 percent reported they would arrange for group counseling sessions (table 3-14).

ISSUES TO BE ADDRESSED BEFORE IMPLEMENTATION

When OTA undertook this survey, privately funded pilot projects were under way, but federally funded pilot studies to evaluate CF mutation analysis in the general population had not yet begun, although NIH had begun a grant competition for such projects (18).[3] Thus, OTA asked survey respond-

[3] In October 1991, NIH launched a 3-year research initiative on clinical assessments of alternative approaches to genetic education, testing, and counseling related to CF mutation analysis (18).

Table 3-15-Issues that Need to be Addressed by Pilot Programs in Cystic Fibrosis Carrier Screening

Rank order
1. Access to genetic counseling
2. Education of the public
3. Payment/cost
4. Sensitivity of the test
5. Protection of confidentiality
6. Quality control and assurance
7. Identification of a target group
8. Availability of reproductive options

SOURCE: Office of Technology Assessment, 1992.

ents what issues they viewed as important before widespread screening is embraced. Specifically, survey participants were asked at the conclusion of the questionnaire to list by priority the important issues to be addressed by pilot studies in CF carrier screening.

Interestingly, the sensitivity of the test, which was often cited as the reason not to proceed with screening, was ranked fourth (table 3-15). Access to genetic counseling was listed as the most important issue to be addressed. But with vast geographic inequities in availability of genetic services it is not clear how access could be considered as anything other than a variable in following pretest and post-test consumer behavior. Education of the public was ranked as second in level of importance for evaluation by pilot programs. Payment and cost issues were ranked third.

SUMMARY

A majority (53 percent) of genetic counselors and nurses in genetics do not offer unsolicited CF carrier screening. They are also unlikely to be providing genetic counseling and DNA tests to families followed in CF clinics and have not yet made efforts to contact CF families seen previously to offer carrier testing to family members. Those who advocate CF carrier tests for use beyond affected families argue that individuals should be routinely informed about the assays so they can decide for themselves whether to be voluntarily screened. This population was a minority (21 percent) of respondents.

If carrier screening is to become routine, 99 percent of respondents believe it should be voluntary, and a majority prefer it be offered to preconceptional adults. Given the clientele found in most clinical genetics settings, it is likely that CF carrier screening will be offered as part of family planning or reproductive health, and the medical specialty most likely to offer the test will be obstetrics. This perceived tension over the technology's control likely contributes to the opinions of some in the clinical genetics community that widespread CF carrier screening is premature until greater genetics education of professionals is in place. With regard to CF carrier screening, concern exists that layers of uncertainty will inhibit informed consent, adequate pretest education, and post-test counseling and that, ultimately, more harm than good might be done. Yet respondents recognize the critical role that could be played in pretest education by other health care professionals and some indicated that should the momentum toward CF carrier screening accelerate, they would make efforts to increase their public and professional education activities.

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