

Appendix B

Survey Instrument

As part of the 1992 assessment *Cystic Fibrosis and DNA Tests: Implications of Carrier Screening*, OTA surveyed the summer 1991 memberships of the International Society of Nurses in Genetics and the National Society of Genetic Counselors. The items for the two questionnaires were identical, and the following is a reproduction of the survey instrument.

Office of Technology Assessment
 United States Congress
 Washington, D.C. 20510-8025

**SURVEY OF GENETIC COUNSELING ATTITUDES AND PRACTICES
 REGARDING CYSTIC FIBROSIS SCREENING**

Genetic Counselor Demographics

1. **Sex:** a. female b. ___ male
2. **Age:** ___ years
3. **Race:** a. Asian d. ___ Hispanic
 b. - Black Native American
 c. - Caucasian f " - Other: _____
4. **Marital status:**
 a. married never married
 b. - widowed d. - divorced/separated
5. **In what State do you work? State a. _____ ZIP code b. _____**
6. **Degrees held:** Year granted:
 a. MA/MS - Genetic counseling _____
 b. - RN/BSN _____
 c. - MSN _____
 d. - MPH _____
 e. ___ MSW _____
 f. ___ Ph. D.: _____
 g. ___ M.D. _____
 h. ___ J.D. _____
 i. - currently in degree program: _____ (type)
7. **How many years of clinical practice as a genetic counselor do you have? _____**
8. **Certification status (American Board of Medical Genetics)**
 a. Board certified (CIRCLE Year): 1982, 1984, 1987, 1990
 b. - Board eligible
 not necessary for position
 d: none
9. **Are you fluent in any language other than English?**
 a. no b. yes, I speak English and (specify other): _____
10. **Present employment status:**
 a. full time
 b. part time: _____ hours/week
 c. - not working

11. Which **best** describes your work setting(s)? Designate a primary (1) and secondary (2) setting, if applicable.

- a. private hospital/medical facility
- b. university medical center
- c. free standing clinic
- d. Health Maintenance Organization (HMO)
- e. private group practice
- f. solo private practice
- g. private industry (specify type): _____
- h. State laboratory (specify type): _____
- i. regional laboratory (specify type): _____
- j. commercial laboratory
- k. Public Health department (State, county, or city)
- L. State government agency
- m. Federal government agency
- n. voluntary health organization
- o. educational institution (K-12)
- p. higher educational institution (undergraduate or graduate)
- q. other: _____

12. On average, how many **hours** a week are you involved in:

- a. direct patient contact (counseling patients)
- b. indirect patient activities (review of literature or records, coordinating referrals)
- performing administrative/managerial tasks
- d: educating health professionals, medical students, GC trainees
- educating the general public, schools, undergraduates
- f“ performing laboratory work
- g. research
- h. marketing/business
- i. other: _____

13. What sources of information about new advances in the field of human genetics do you rely on? (check all that apply)

- a. professional colleagues
- b. medical journals
- grand rounds
- d : State or regional conferences
- national conferences
- f “ American Society of Human Genetics
- g . : National Society of Genetic Counselors
- h. continuing education courses
- i . literature from biotechnology/commercial firms
- j . lay press
- k. other: _____

14. In your current position, how frequently were you asked about DNA testing/screening for CF during the 6-month period from January - June, 1991? Please consider this in the context of your total clinical practice.

- a. never
- b. rarely
- occasionally
- d: frequently
- e . very frequently

15. If you were asked about DNA testing/screening for CF please estimate the number of requests per month (January - June, 1991)? _____ (per month)
16. Compared to 2 years ago, would you say the number of requests made between January - June, 1991 represents:
- a. _____ a large decrease
 - b. _____ a small decrease
 - _____ no change
 - d : _____ a small increase
 - e . _____ a large increase
17. If you noted an increase, when did you note this? (month/year) _____
18. In your current position are you engaged in providing genetic counseling?
- a. _____ yes b. _____ no

If NO, skip the CLINICAL PRACTICE QUESTIONS and GO TO QUESTION #46

THE NEXT SERIES OF QUESTIONS ARE TO BE ANSWERED BY THOSE INDIVIDUALS WHO CURRENTLY PROVIDE GENETIC COUNSELING SERVICES

(All others please skip to question #46.)

19. Which best describes the primary service area in which you work?
- a. _____ rural
 - b. _____ suburban
 - c. _____ metropolitan/urban
 - d. _____ statewide
 - e. _____ regional (more than one State)
 - f. _____ national
 - g. _____ other: _____
20. Current level of staffing (including yourself) in your counseling unit/program (please indicate number).
- | | # |
|---------------------------|-------|
| a. M.D. geneticists | _____ |
| b. Ph.D. geneticists | _____ |
| c. M.D./Ph.D. geneticists | _____ |
| d. genetic counselors | _____ |
| e. secretaries | _____ |
| f. other: _____ | _____ |

21. Indicate the frequency of patients seen by you for each major area of clinical practice.

1 =seldom if ever; 2=sometimes; 3=often (i.e., majority); 4=very often; 5=all or almost all

- a. ___ prenatal genetics
- b. ___ pediatric genetics
- ___ adult genetics
- d : - teratogen exposure
- reproductive loss
- f “ - specialty disease(s) clinics (please specify): _____
- g. ___ newborn screening
- h. ___ MSAFP screening follow-up
- i. ___ carrier screening (specify disease): _____

22. Does your institution participate in collecting the CORN data set?

- a. ___ yes b. ___ no c. ___ don't know

23. For each of the following categories, indicate the number (or best estimate) of genetics clients/patients served in 1990, either DIRECTLY (i.e., counselor to client relationship; one-on-one genetic counseling) or INDIRECTLY (i.e., involvement such as consultant to primary care physician regarding a patient, telephone consultation).

	<u>TYPE OF PATIENT CONTACT</u>		
	Direct	Indirect	Total
All patients seen in 1990			
a. by your unit:	_____	_____	_____
b. by you individually:	_____	_____	_____
CF patients/families seen in 1990			
c. by your institution:	_____	_____	_____
d. by you individually:	_____	_____	_____

24. With respect to your clinical practice, estimate the percent (%) of your patients who are:

A. RACE/ETHNICITY	Percent (%)
a. Asian/Pacific Islander	_____
b. Black	_____
c. Caucasian	_____
d. Native American	_____
e. Spanish surname	_____
f. unable to estimate	_____
B. AGE DISTRIBUTION	
g. neonatal	_____
h. infants	_____
i. children	_____
j. adolescents	_____
k. adults - reproductive age	_____
l. adults - post reproductive age	_____
m. unable to estimate	_____

24. (cont.) With respect to your clinical practice, estimate the percent (%) of your patients who are:

C. LANGUAGE	Percent (%)
n. English speaking	_____
o. Non-English speaking	_____
p. unable to estimate	_____

25. Do your patients have health care coverage?
- a. seldom if ever (0-15% of patients seen)
 - b. sometimes (about 16-50% of patients)
 - c. often (about 51 -74% of patients)
 - d. very often (about 75-69% of patients)
 - e. always or almost always (90-100% of patients)

26. Please estimate the percent of patients by category of coverage.

<u>Coverage Category</u>	<u>Percent(%. </u>
a. commercial insurance	_____
b. Blue Cross/Blue Shield	_____
c. HMO or managed care plan	_____
d. Medicaid	_____
e. Medicare	_____
f. CHAMPUS	_____
g. self pay	_____
h. no insurance	_____
i. indigent	_____
j. unknown	_____

27. For individuals with insurance coverage, what has been your experience with reimbursement of fees for service in each of the following areas? Also, please indicate the average fee amount charged for each service.

1 =seldom if ever covered; 2 =sometimes covered; 3 = often covered;
4 very often covered; 5=almost a/ways covered; 6=uncertain

- a. ___ general genetic counseling: Fee \$ _____
- b. ___ genetic counseling for cystic fibrosis with positive family history: Fee \$ _____
- c. ___ genetic counseling for cystic fibrosis with negative family history: Fee \$ _____
- d. ___ routine metabolic screen: Fee \$ _____
- e. ___ routine cytogenetic analysis: Fee \$ _____
- f. — DNA analysis for cystic fibrosis: Fee \$ _____

28. Have you had any experience with a patient's insurance claims for DNA testing being rejected?
 a. ___ no experience b. yes. Please provide details:

29. Have any of your patients experienced difficulties in obtaining or retaining health insurance coverage as a result of genetic testing?
 a. ___ no experience b. ___ yes. Please provide details:

30. Consider the following reasons for referral for genetic counseling. Please estimate to the best of your ability, the average NUMBER of patients you see per month, total amount of direct COUNSELOR TIME spent (in minutes), and the average number of VISITS needed to provide genetic counseling to individuals and/or families for each of the following scenarios. (Answer for cases appropriate to your practice.)

	<u>AVG</u> <u># Pts</u>	<u>Time/</u> <u>visit</u>	<u>AVG</u> <u># visits</u>
a. prenatal counseling for advanced maternal age	_____	_____	_____
b. positive family history for neural tube defects concerns for current pregnancy	_____	_____	_____
c. Elevated MSAFP screen	_____	_____	_____
d. Couple with newly diagnosed (Tri 21) Down's Syndrome child	_____	_____	_____
e. Couple with 14/21 translocation Down's Syndrome child	_____	_____	_____
f. Carrier testing for DMD	_____	_____	_____
g. Newly diagnosed case of neurofibromatosis	_____	_____	_____
h. Newly diagnosed CF family	_____	_____	_____
i. Carrier testing for CF with a positive family history	_____	_____	_____
j. Carrier testing for CF with a negative family history	_____	_____	_____

31. If you have not been involved with counseling for CF based on your experience, how much direct counselor time (minutes) would you estimate would be needed to:

- a. obtain 3 generational family pedigree: _____ (minutes)
 b. discuss carrier testing and recurrence risks: _____ (minutes)

32. How would this estimate compare to the direct patient time spent with your typical patient load?

- a. ___ more time b. less time c. about the same

33. How frequently do you use each of the following formats to provide genetic counseling?

1 =seldom if ever; 2=sometimes; 3=often; 4=very often; 5=almost always

- a. individual counseling session(s)
- b . group counseling
- videotape alone
- d : videotape with counseling
- written educational materials
- f “ slide-tape
- g . interactive computer

34. Where is the closest CF treatment center to your institution?

- a. at my institution
- b . less than or equal to 50 miles
- greater than 50 miles
- d : not aware of one

35. Do you personally provide genetic counseling through the CF treatment center in your area?

- a. no
- b. yes

If yes, please provide the following information for 1990.

- 1) total # new patients seen by the CF center _____
- 2) total # return patients seen by the CF center _____
- 3) #referrals for genetic counseling _____
- 4) # requests for information on DNA testing _____
- 5) # undergoing actual DNA testing _____ individuals _____ families

36. Do you or your group/unit have a specific policy regarding DNA testing for CF

- a. no, we do not.
- b. yes; if yes, what is it?

37. Are individuals/families seeking DNA testing for CF asked to sign an informed consent?

- a. no
- b. yes

38. Do you or your group/unit have official policies and procedures for other issues in genetics?
(check all that apply)

- a. DNA storage
- b . prenatal diagnosis for sex selection
- non-paternity
- d : confidentiality and Huntington's disease testing
- e . other: _____

39. For each of the following patient groups, indicate how often, if at all, you introduce the topic of DNA testing for CF

1=seldom if ever; 2=sometimes; 3=often; 4=very often; 5=almost always

- a. all patients/families
- b. pregnant women seeking prenatal diagnosis
couples/individuals with a positive family history for CF
- d : Caucasian couples/individuals with negative family history for CF
- f . individual/families who inquire about CF
- e . selected couples/individuals; how selected:

40. Have you made an effort to contact old genetics families as appropriate regarding the availability of CF testing?

a. yes, by (check all that apply):

- 1) telephone
- 2) letters/mass mailing
- 3) at future visits
- 4) other: _____

b. no, because (check all that apply):

- 1) not enough time; too busy
- 2) no mechanism for rapid chart retrieval
- 3) requires chart by chart analysis
- 4) plan to do so in future, as time permits
- 5) other: _____

41. During the last 12 months:

a. Have you referred any patients for DNA testing for CF

- 1) no
- 2) yes: how many individuals: _____ # samples _____

b. Have you referred any patients/families for DNA testing for other disorders?

- 1) no
 - 2) yes: how many individuals: _____ # samples _____
- If yes, for which conditions:

42. At your institution, is DNA testing for CF

- a. performed at onsite/inhouse lab
- b . sent offsite to lab less than or equal to 50 miles away
- c . sent offsite to lab between 50 miles and 150 miles away
- d. sent offsite to lab greater than 150 miles away

43. Type of laboratory used for CF testing:

- a. private/commercial
- b . private hospital
- university hospital
- d : regional laboratory
- e . other: _____

44. If you are-or have been-involved with CF testing, does the laboratory you use provide (check all that apply):
- a. direct mutation analysis
 - b. DNA linkage analysis
 - DNA haplotyping
 - d : staging of studies depending on case
 - prenatal DNA analysis
 - f “ fetal intestinal enzyme analysis
 - g . DNA banking
45. For direct mutation analysis of CF what mutations does the laboratory you use include? (Please list or give number):

THE FOLLOWING QUESTIONS ARE TO BE ANSWERED BY ALL RESPONDENTS

46. Are you familiar with the following statements concerning CF screening published by:
- a. 1990 ASHG ad hoc CF Screening Committee: no — yes
 - b. 1990 NIH panel: no yes —
- if yes to either one of the above how have you incorporated this into clinical practice?
47. At this time do you think it is appropriate to provide CF screening in cases where family history is negative?
- a. no b. yes c. uncertain
- if yes, why?
48. Do you feel there is a optimum rate of detection at which widespread CF carrier screening should proceed?
- a. yes, specify: _____ % rate of detection
 - b. n o
 - c. no opinion
49. Are you familiar with the NSGC brochure “Genetic Testing for Cystic Fibrosis: A Handbook for Professionals”?
- a. no b. yes
50. Have you developed any educational materials relevant to DNA testing specifically for CF
- a. no b. yes (Please send a copy.)
51. Have you been tested for CF carrier status?
- a. no b. yes
52. If you have been tested for CF carrier status, why were you tested?
- a. research subject
 - b. wanted to know
 - positive family history
 - d : family planning
 - e . other: _____

53. How was your test covered?
- a. by my insurance
 - b. professional courtesy
 - c. self pay
 - d. research subject
54. To what extent, if at all, should each of the following groups be involved with educating the public about DNA testing for CF if it becomes standard practice?

*1=to little or no extent; 2=to some extent; 3=to a moderate extent;
4=to a great extent; 5=to a very great extent; 6=no opinion*

- a. primary care providers
 - b. public health departments
 - c. genetic counselors
 - d. genetics programs
 - e. nurses
 - f. family planning clinics
 - g. voluntary support groups
 - h. schools
 - i. lay press
 - j. television
 - k. other: _____
55. If widespread CF carrier screening begins, it should be:
- a. mandatory
 - b. voluntary
56. If widespread CF carrier screening begins, what target populations should be screened? (check all that apply)
- a. prenatal
 - b. newborns
 - c. children ages 2-12
 - d. children ages 13-18
 - e. adults in reproductive years
 - f. adults post reproductive years
 - g. pregnant women or "couples"
57. If CF carrier screening is voluntary, who should organize the screening programs? (check all that apply)
- a. voluntary health organizations
 - b. State or local health department
 - c. Federal Government
 - d. medical societies
 - e. the human genetics community
 - f. primary care givers
 - g. others (specify): _____
58. If CF carrier screening is mandatory, who should organize the screening programs? (check all that apply)
- a. voluntary health organizations
 - b. State or local health department
 - c. Federal Government
 - d. medical societies
 - e. the human genetics community
 - f. primary care givers
 - g. others (specify): _____

59. Where should CF population screening programs be provided? (check all that apply)
- a. in public schools
 - b. in public health departments
 - c. in organized, community-wide programs
 - d. in the primary care setting i.e., physicians
 - e. in genetic centers/programs
 - f. in the workplace
 - g. other (specify): _____
60. Who should pay for screening? (Please rank, but be realistic.)
- a. self pay by patient
 - b. third party payment
 - c. employers
 - d. State/city or county
 - e. Federal government
 - f. other (specify): _____
61. Do you agree or disagree with the following statements?
1=strongly agree; 2=agree; 3=undecided; 4=disagree; 5=strongly disagree
- a. genetic counseling should precede DNA testing for CF when there is a positive family history.
 - b. genetic counseling should precede DNA testing for CF when there is a negative family history.
 - c. educational materials (culturally sensitive and understandable) can provide adequate information about CF screening.
 - d. a need for more genetic counselors exists.
 - e. informed consent prior to CF screening is a necessity.
62. In your opinion, what are the important issues that need to be addressed by pilot programs in CF screening? List in order of priority:
- 1.
 - 2.
 - 3.
 - 4.
63. What strategies have you considered implementing if widespread screening for CF becomes a reality?
64. What do you feel the minimum criteria for CF carrier screening should be (protocol)?

Thank you very much for your cooperation in answering our Questions! On the back of this survey, please feel free to give us as any other options, concerns, or suggestions that you feel our questions did not address. These comments will be anonymous, but may be incorporated in our report to Congress.