Chapter 4
Summary

For years, experts theorized about confronting the potential consequences of increased knowledge of human genetics. In the early 1990s, the cystic fibrosis CF mutation test moved the debate from the theoretical to the practical. OTA concludes that the value of the CF carrier testis the information it provides. No one can estimate in common terms what it means to an individual to possess information about his or her genetic status, especially when the value concerns reproductive decisionmaking. On a larger scale, the potential for widespread CF carrier screening raises legal, ethical, economic, and political considerations.

This survey of genetic counselors and nurses working in genetics, conducted in the summer of 1991, reflects the tensions and concerns surrounding dissemination of CF mutation analysis. The survey was conducted to better understand the environment in which the average genetic counselor or nurse in genetics works, to describe the infrastructure and tools available to these professionals, to assess the state of practice in the provision of CF carrier screening, and to evaluate their attitudes regarding CF carrier screening.

In summer 1991, most genetic counselors and nurses in genetics did not offer unsolicited CF carrier screening and expressed concerns about access to health insurance, quality control, public education, discrimination, stigmatization, and the adequacy of trained personnel as reasons why they did not. 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, although agreement exists that such individuals should be offered CF mutation analysis.

Reasons why survey respondents do not offer CF mutation analysis are varied, but professional guidelines exert some influence. The 1990 policy statement of the American Society of Human Genetics (ASHG) stated that CF carrier screening is ***"NOT*** yet the standard of care," and a majority of survey respondents were aware of that statement. Several stated that it alone was the reason for their refusal to offer CF carrier screening. In mid-1992, after extended discussion, ASHG's leadership approved a revised statement that CF mutation analysis 'is not recommended' for those without a family history of CF Some argue that the subtle change in language of the new statement reflects an evolution of debate within the society-that some believe CF carrier screening *may now be offered* to individuals without a family history of CF although it might not be the "standard of care. " Others argue that ASHG's position is unchanged. The effect of the new statement remains to be seen.

Concern about test sensitivity was another barrier that respondents said should be addressed before routine CF carrier screening. Two-thirds of participants felt that an optimum frequency of detection should be reached before they would feel comfortable offering CF carrier screening to the general population, although nearly a quarter of respondents were uncertain about whether an optimum was necessary. Of those who felt there is an optimal frequency of detection, nearly half felt that 95 percent test sensitivity should be required before proceeding with widespread CF carrier screening. Twenty-five percent believed test sensitivity should be greater than 95 percent, with 4 percent stating that it should be 100 percent. At the time this survey was conducted, test sensitivity was approximately 80 percent. It has increased to 85 to 90 percent as of summer 1992, so opinions might have changed.

Two other factors ranked slightly more important than test sensitivity as criteria to consider before implementing routine CF carrier screening: the availability of adequate counseling and an adequate system of referral for individuals who test positive. Genetic counseling can be labor intensive. Survey respondents indicated that they spend most of their work week seeing or talking with clients; patient loads are frequently heavy. Respondents said that, given the potentially complex or emotional nature of some genetic information, professionals trained in human genetics are essential to insure high quality care and informed consent. Guarantee of informed consent also was mentioned as necessary for implementation of large-scale carrier screening.

Those who advocate CF carrier tests for use beyond affected families are no less concerned about

informed consent and quality of services. Proponents argue that the tests are sensitive enough for current use and that individuals should be routinely informed about the assays so they can decide for themselves whether to be voluntarily screened. These voices believe that failing to inform patients now about the availability of CF carrier assays denies people the opportunity to make personal choices about their reproductive futures. In this survey population, however, advocates of routine CF carrier screening were in the minority.

Perhaps the point on which there was greatest consensus among 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.

Given the existing tensions surrounding CF mutation analysis in the general population, who should serve as gatekeeper of this new technology? *Survey* respondents strongly believe that CF carrier screening should be organized by and provided by the human genetics community. This assumes, however, that large numbers of Americans will learn of their CF carrier status through interaction with the genetic services system.

Based on the client populations reported in this survey, routine CF carrier screening will likely integrate into medicine in the reproductive context first. The prenatal population has been the traditional entry point into genetic services for many people; OTA's survey found 70 percent of the genetics clientele are adults of reproductive age, reinforcing the notion of prenatal diagnosis as an entry point for primary genetics service.

Preconceptional individuals are the ideal population for CF carrier screening, according to survey respondents, but for most individuals the first real opportunity for carrier screening takes place post-conception. Thus, despite survey respondents' desire that information about the availability of assays such as CF mutation analysis should come from genetic specialists, the primary responsibility for providing CF carrier screening is likely to reside with obstetricians, at least initially, and especially if reimbursement for CF mutation analysis and its attendant counseling become part of routine obstetric care. Such a scenario would mirror that which has occurred with maternal serum alpha-fetoprotein screening to detect fetuses with neural tube or abdominal wall defects or Down syndrome-a prospect that concerns some, but not others.

OTA's survey of genetic counselors and nurses also reports some consumers experience difficulties in obtaining or retaining health care coverage after genetic tests, though the large majority were not for carrier status, but were for genetic illness. Nevertheless, because genetic-based predictive testing promises to have a profound impact on clinical medicine-and because access to medical care is inextricably linked to private health insurance in this country—such cases underscore certain policy dilemmas arising from the increased availability of genetic assays.

Critics of widespread CF carrier screening question whether the present genetic counseling system in the United States can handle the swell of cases if CF carrier screening becomes routine. Currently, about 1,000 master' s-level genetic counselors practice in the United States, and an additional 100 nurses in genetics provide similar services. OTA's survey of genetic counselors and nurses in genetics indicates that respondents believe routine CF carrier screening will strain the present genetic services delivery system. Respondents estimated that, on average, 1 hour would be needed to obtain a three-generational family history and to discuss CF carrier screening and genetic risks.

Skeptics of a personnel shortage assert that counseling about CF carrier assays is likely to take place in the general obstetric/prenatal context, however, and believe 1 hour exaggerates the amount of time that suffices for all prenatal tests, let alone only CF carrier screening. Furthermore, counseling related to CF carrier screening is likely to extend beyond genetics professionals to include other physicians and allied health professionals. For example, an unknown number of social workers, psychologists, and other public health professionals perform genetic counseling, often to minority and underserved populations.

ultimately, the issue of adequate services and professional capacity could turn on the extent to which patients receive genetic services through specialized clinical settings, as they largely do now, versus access through primary care, community health, and public health settings. Overall, OTA cannot conclude whether increased numbers of genetic specialists are necessary, but clearly in-

creased genetics education for all health care professionals is desirable. Routine carrier screening for CF-and tests yet to be developed for other genetic conditions-will require adequate training and education of individuals in the broader health care delivery system. Some survey respondents recognize the critical role other health care professionals will play in pretest education and indicated that should the momentum toward CF carrier screening accelerate, they would make efforts to increase their public and professional education activities.

Although genetic counselors and nurses in genetics work in a variety of settings, they are concentrated in metropolitan medical centers on the West coast or in the Northeast. States with large rural populations are less likely to be served. The clientele served, in the aggregate, tend to be representative of the national averages for racial and ethnic populations, although no effort was made by OTA **to** match racial and ethnic data with regions, cities, or localities. This diversity presents great opportunity in terms of professional and public education, yet few counselors report an emphasis on these activities in their weekly routine because patient services comprise two-thirds of their time.

One of the tasks of genetic specialists, however, is to provide the educational and counseling services necessary to successful implementation of new technologies. Diagnostic tools, such as DNA tests, can provide powerful information. Increasingly, genetic counselors and nurses working in genetics will be at the front line on the issues raised by assimilating DNA technologies into clinical practice.