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OTA ISSUES SURVEY RESULTS ON CYSTIC FIBROSIS CARRIER SCREENING

Since 1989 a test has been available to detect carriers of cystic fibrosis, the most common life shortening recessive disorder affecting Caucasians of European descent. As of summer 1991, a survey by the congressional Office of Technology Assessment (OTA) of 431 genetic counselors and nurses in genetics, found that 53 percent of the respondents did not offer CF carrier screening to individuals without a family history of the disease. Only 21 percent of respondents advocate the use of voluntary carrier screening to individuals with no family history.

The survey is the subject of a background paper issued today that supports OTA's August 1992 assessment *Cystic Fibrosis and DNA Tests: Implications of Carrier Screening*, requested by the House Committee on Science, Space, and Technology, the House Committee on Energy and Commerce, and Representative David R. Obey.

About 2,000 babies with CF are born annually in the United States. Beyond the approximately 30,000 Americans who have CF, as many as 8 million individuals could be carriers for CF. Prior to 1989, the absence or presence of CF in one's family, as well as ethnic and racial background, were the only indicators available to determine risk of being a CF carrier. Today a one-time test can detect 85 to 95 percent of CF carriers, depending on ethnicity.

Those opposed to screening individuals without a family history of CF cite concerns about the test's less than 100 percent detection rate, the need for adequate pre-test education and post-test counseling, and the potential for stigma and discrimination of those identified as carriers. Those who advocate the tests are no less concerned about informed consent and quality of services, but 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.

Two-thirds of respondents to OTA's survey believed that the test sensitivity should be greater before screening is routinely offered, with nearly 30 percent stating that is should be at least 95 percent. At the time of the survey, test sensitivity was approximately 80 percent, but as of summer 1992, it has increased to 85 to 90 percent -- and in some cases 95 percent -- so opinions might have changed. Nearly all surveyed (99 percent) strongly agree that CF carrier screening should be voluntary and never mandatory.

Based on the profile of client populations seen most often by survey respondents, routine CF carrier screening will likely first integrate into medicine in reproductive health care, says OTA. The population seeking prenatal advice most often seek and obtain genetic services; OTA's survey found 70 percent of the genetics clientele are adults of reproductive age.

Individuals who have not yet conceived are the ideal population for screening, according to survey results, but for most individuals the first real consideration of screening takes place after 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. This is especially likely, notes OTA, if reimbursement for CF mutation analysis and its attendant counseling become part of routine obstetric care.

An additional concern raised by those questioned in the survey is the potential strain on genetics professionals should the volume of tests suddenly rise. A minority of survey respondents believe that improved genetics training for all health care professionals is the best method for alleviating this strain. Most cite increasing the number of trained genetic counselors as the best approach.

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 by this group of health professionals, says OTA.

The survey also found a few anecdotal cases of consumer difficulties in obtaining or retaining health care coverage after genetic tests, though the large majority of reported cases were not for carrier status, but were for genetic illness. Eighty percent of respondents believe that third parties should pay for carrier screening.

In addition to the survey results, the background paper describes the environment in which the average genetic counselor or nurse in genetics works, the infrastructure and tools available to these professionals, as well as the state of practice in the provision of CF carrier screening.

Prospects of routine CF carrier screening polarize people, as is clear from the results of the survey. OTA concludes that the value of the CF carrier test is the information it provides.

Copies of the 49-page background paper *Genetic Counseling and Cystic Fibrosis Carrier Screening: Results of a Survey* for congressional use are available by calling 4-9241. Copies for noncongressional use may be ordered from the Superintendent of Documents, U.S. Government Printing Office, Washington, D.C. 20402-9325; phone (202) 783-3238. The GPO stock number is 052-003-01305-3. The price is $3.50.

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