OTA Report Brief

August 1992

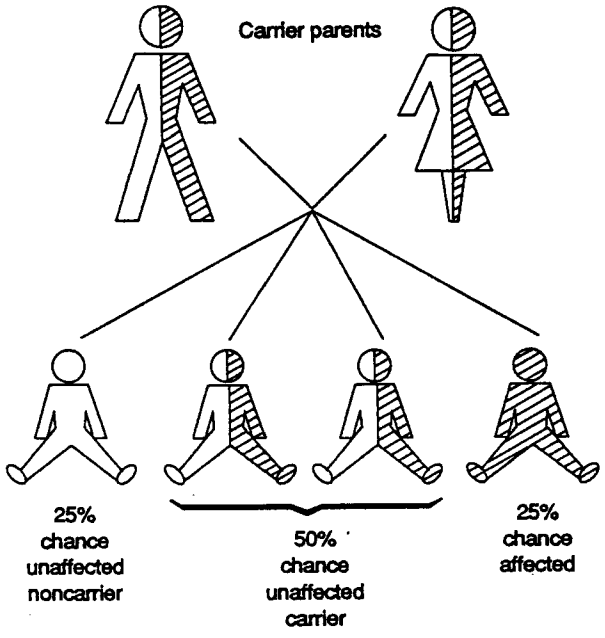
Cystic Fibrosis and DNA Tests: Implications of Carrier Screening

Today, medical technology includes genetic tools that can deliver predictive information with ever-increasing accuracy. One of these tools is a DNA test that can tell people about their risk of having a child with a genetic condition called cystic fibrosis (CF).

CF is the most common, life-shortening, recessive disorder affecting Caucasians of European descent, and it also affects other racial and ethnic groups. Parents of a child with CF do not have CF, but are referred to as CF carriers. They have no symptoms of CF, but with each pregnancy are at 1 in 4 risk of having a child with CF and 1 in 2 risk of the child being a CF carrier (figure 1). Couples are not at risk of having a child with CF if only one or neither partner is a carrier.

Since 1989, clinicians have been able to use DNA tests to screen and identify CF carriers before they have a child with CF. The OTA Report concentrates on these millions of CF carriers, who are, today, largely unidentified (table 1). Consensus exists that individuals who have relatives with CF should be told about the availability of CF carrier tests; the disagreement is whether *everyone* should be informed about the assays, since 4 of 5 babies with CF are born to couples with no previous family history of the condition.

Figure 1—Inheritance of Cystic Fibrosis



SOURCE: Office of Technology Assessment, 1992.

Table 1—Cystic Fibrosis and Cystic Fibrosis Carrier Screening in the United States

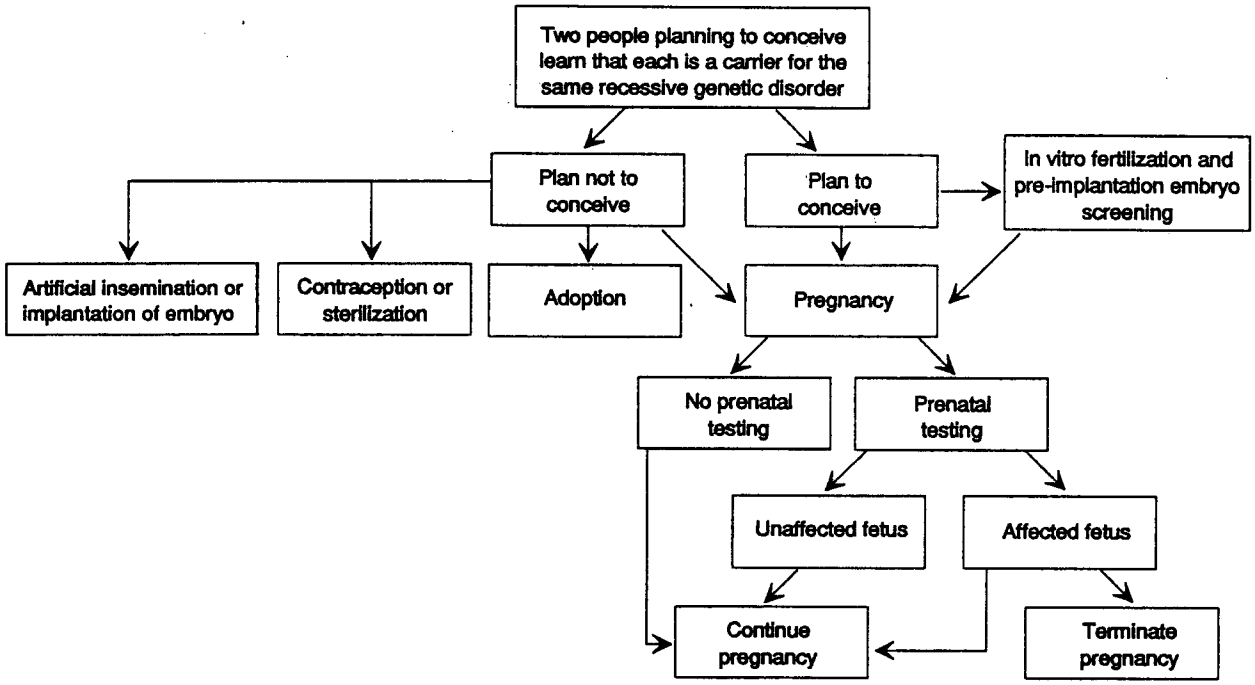
1,700 to 2,000. .	Babies born annually with CF
30,000.	Americans with CF
12 years.	Median age of individuals with CF
28 years.	Median lifespan of individual CF patient
8,000,000.	Number of Americans who might be CF carriers
1 in 25.	Frequency of CF carriers among Caucasian Americans of European descent
1 in 40 to 50. . . .	Frequency of CF carriers among Hispanic Americans
1 in 60 to 65. . . .	Frequency of CF carriers among African Americans
1 in 150.	Frequency of CF carriers among Asian Americans
1 in 625.	Caucasian American couples where both partners are carriers and hence at 1 in 4 risk of having a child with CF with each pregnancy
6,000,000.	Pregnancies per year that might be screened for CF status[a]
10,000,000.	American men and women who might be screened annually if routine screening in the prenatal context is practiced[b]
125,000,000. . . .	Number of Americans of reproductive age who theoretically could be involved in CF carrier screening
9,310.	OTA estimate of the number of CF carrier tests in 1991
63,000.	OTA estimate of the number of CF carrier tests in 1992

[a]Based on the annual number of births and spontaneous abortions.
[b]Twenty-four percent of pregnant women receive no prenatal care until the third trimester; this number reflects that statistic.

SOURCE: Office of Technology Assessment, 1992.

Prospects for routine CF carrier screening are viewed with mixed feelings. Invariably, discussions about CF carrier screening focus on how health insurers use, or might use, genetic information, and they become linked to the broader debate about health care access in the United States. Some also question the adequacy of quality assurance for DNA diagnostic facilities and the tests themselves. Today's tests detect 85 to 95 percent of CF carriers, depending on a person's race and ethnicity. This lack of perfection troubles those who would like more

Figure 2—Decisionmaking After Carrier Identification



SOURCE: Office of Technology Assessment, 1992, based on F. Cohen, *Clinical Genetics in Nursing Practice* (Philadelphia, PA: Lippincott, 1984).

sensitive detection rates before the tests are put into widespread use. Some also wonder whether the current number of genetic specialists can handle a swell of CF carrier screening cases, let alone cases from tests for other genetic conditions made possible by the Human Genome Project. And, fundamental to consideration of CF carrier screening is the issue of genetic counseling and abortion.

Those who advocate CF carrier screening in the general population are equally concerned about these issues. They assert, however, that all individuals should be routinely informed about the availability of CF carrier assays so they can decide for themselves whether to be voluntarily screened. Proponents of offering CF carrier tests believe that failing to inform everyone now about their availability denies people the opportunity to make personal choices about their reproductive futures (figure 2).

In the past year, the number of CF carrier tests performed in the United States is expected to increase nearly 7-fold (table 1). Some physicians inform all individuals about CF carrier assays, not only those who have a family history of the condition, but this practice is not routine as of mid-1992. Nevertheless, OTA concludes that routine CF carrier screening in the United States is a matter of when, not if. What is unclear is the timeframe for a custom of care to evolve where health care providers routinely inform patients about the availability of CF carrier tests. It could be within a year or two, but more likely will be a gradual process over several years. OTA identifies eight factors that will affect routine utilization:

- genetic services delivery and customs of care;
- public education;
- professional capacity;
- financing;
- stigmatization, classification, and discrimination issues;
- quality assurance of clinical laboratories and DNA test kits;
- automation; and
- costs and cost-effectiveness.

OTA concludes that the value of the CF carrier test is 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. Some people want and seek this information; others do not. CF carrier tests also tell them something about their own parents and siblings because genetic assays, unlike most medical tests, convey information about family members.

OTA presents options for action by Congress in six policy areas:

- genetics education and the public,
- genetics training and education of health care professionals,
- discrimination (e.g., in access to health care coverage),
- clinical laboratory and medical device regulation,
- instrumentation to automate DNA diagnostics, and
- integration of DNA assays into clinical practice.

Finally, the Report concludes that the number of widely available DNA tests for genetic disorders and predispositions unquestionably will increase rapidly over the next decade. Many of the policy issues raised in this Report extend beyond implications for CF carrier screening to other applications of DNA technologies.

Copies of the report for congressional use are available by calling 4-9241.

Copies of the report for non-congressional use can be ordered from the Superintendent of Documents, U.S. Government Printing Office, Washington, DC 20402-9325 (202) 783-3238. The GPO stock number for the OTA report, "Cystic Fibrosis and DNA Tests: Implications of Carrier Screening," is 052-003-01291-0. The price is $16.00. Summaries of reports are available at no charge from the Office of Technology Assessment.

For further information contact OTA's Publications Office. Address: OTA, U.S. Congress, Washington, DC 20510-8025 (202) 224-8996.