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Faces Disappearing

The Implications of Cystic Fibrosis Screening

Cystic fibrosis (CF) is a hereditary disease that affects much of the body, especially the lungs and digestive system, leading eventually to disability and death. The disease runs its course very differently in different patients; some have severe pulmonary and gastrointestinal problems starting in the first year of life, while others have relatively mild symptoms until adolescence. Lung disease is usually the primary factor in determining the quality and length of a patient's life; about 90 percent of all people with CF die from pulmonary complications. Since cystic fibrosis generally doesn't impair cognitive functioning or musculoskeletal development, it is possible for patients with mild cases to lead relatively normal lives; a 1995 survey reported that 35 percent of young adults diagnosed with CF work full-time and 90 percent had completed high school. Thanks to therapeutic advances in recent decades, the median survival age of patients has been steadily rising: According to the Cystic Fibrosis Foundation, in 1976 it was 18 years, in 1995 it was 30.1 years, and in 2005 it was 36.8 years.

About 30,000 Americans have cystic fibrosis, and around 1,000 new cases are diagnosed each year. The disease is most common among Caucasians, with an incidence of 1 in 3,500 live births (compared to roughly 1 in 12,000 in non-white populations). Our understanding of the genetic causes of the disease have improved; since 1989, over 1,300 mutations of a particular gene have been linked to CF. These discoveries have made it possible to use genetic tests to determine if expecting parents are unwary carriers of a cystic fibrosis mutation, or to determine if a person actually has the disease—so parents with afflicted newborns, for instance, can know about the disease before symptoms are manifest. A workshop convened by the Centers for Disease Control in 1997 recommended such neonatal CF screening, calling it a "paradigm for public health genetics policy development."

Two years later, in 1999, a panel put together by the National Institutes of Health (NIH) recommended that CF screening be pushed further back from neonatal care to prenatal care. The NIH panel published a statement intended to give health care providers, patients, and the general public a "responsible assessment of the optimal practices for genetic testing for cystic fibrosis" so that individuals would be able to obtain enough information to make "informed decisions." The panel recommended that health care providers offer cystic fibrosis testing to adults with a family history of CF, to partners of people with CF, to couples planning a pregnancy, and to couples seeking prenatal care. In 2001, following the lead of the NIH, the American College of Obstetricians and Gynecologists (ACOG) issued an essentially identical set of recommendations, in the form of guidelines and educational materials distributed to ACOG members.

Since the publication of the ACOG guidelines, cystic fibrosis testing has become an increasingly common part of prenatal care. About a year after the guidelines were published, *OB/GYN News* reported that leading commercial genetic and diagnostic testing laboratories performed between 300,000 and 500,000 CF carrier tests in the United States. While that figure represented a tripling, quadrupling, or in one case an elevenfold increase, in testing volumes at those labs when compared with 2001, it was still far short of ACOG's "target levels," since the number of tests was "dwarfed by the approximately 4 million live births a year in the United States."

For expecting parents, a carrier test is either a one-step (both the woman and the man are tested simultaneously) or a two-step process (the woman is tested first; if she is a carrier, the man is then tested). If both are carriers, an amniocentesis or chorionic villus sampling is performed to test the unborn child. There are no definite statistics on the number of cystic fibrosis tests now performed on fetuses in the United States, but it is clear that prenatal CF testing is quickly joining the so-called "quadruple screen," a common regimen of tests that look for chromosomal abnormalities (Down syndrome in particular) and neural tube defects. If a prenatal test shows that a fetus has inherited both parents' mutations, the unborn child is classified as having cystic fibrosis. The parents are then usually referred to genetic counseling, either from a licensed genetic counselor or, more frequently, from their OB-GYN, to receive information about the disorder, the prognosis, and the available treatment options.

In practice, however, such counseling is woefully inadequate. The test results are especially tricky to interpret: a child may have a CF genotype, but it is unclear how the disease would be phenotypically expressed. The NIH statement on this issue points out that identifying the specific CF mutation is not "highly predictive of the severity and course of pulmonary disease, which is the major factor affecting patient quality of life and longevity."

Some men with "a high frequency of CF mutations" are healthy but for sterility caused by a genital deformation; some women with CF mutations "are normal or develop chronic sinusitis or bronchitis as the extent of their morbidity," according to the NIH. "It is unclear whether such mildly affected individuals can be reliably identified by their genotype." In other words, an unborn child who tests positive for CF may have minimal if any symptoms or may ultimately have extensive life-shortening pulmonary disease. It is possible to predict the extent of pancreatic insufficiency, but not possible to predict the extent of the symptoms that would have the greatest effect on the child's life.

This inability to predict the extent of the disease is highly significant for genetic counselors trying to communicate with parents. In a 2002 review of the state of prenatal CF testing for the *Journal of Pediatrics*, Philip M. Farrell and Norman Fost conclude:

> Many couples will undoubtedly benefit, but there is reason for concern about the potential for harm, and uncertainty about how many couples will be making truly informed choices. CF is one of the most complex single-gene disorders with extraordinary genotypic and phenotypic variation coupled to an evolving, inexorably improving array of therapies. Communicating information about prognosis is a daunting challenge, especially for health care professionals with limited experience in managing patients with CF, as is the case with most obstetricians.

Farrell and Fost note that geneticists believe it would require a trained counselor an hour or more to convey the necessary information. It is unlikely that this is happening in busy obstetrical practices. Given the complexities of the diagnosis in the prenatal period, the meaning of the diagnosis once it is made, and the likelihood that insufficient information is being conveyed, there is great potential for doing more harm than good. Such genetic counseling is inadequate at best, and may be irresponsible.

More fundamentally, while the NIH and ACOG do not claim any moral standpoint on abortion, they recommend widespread testing in the interest of "helping people make informed decisions." They assume abortion is an appropriate response to physical or mental disability, and this perspective is built into genetic counseling. That has certainly proven true in the case of Down syndrome. Currently, roughly 90 percent of the fetuses that test positive for Down syndrome in the United States are aborted. "For me, it's just faces disappearing," the mother of a daughter with Down syndrome recently told the *New York Times*. The rapid mainstreaming of prenatal CF testing is poised to follow precisely the same course.

Even though early diagnosis can be immensely beneficial in providing a

couple with information to begin treatment before the child develops symptoms, newborn screening would suffice for this. The prenatal test is explicitly designed to diagnose CF in order to give the patient the option to abort the child—and by simply offering it, the clinician is, at some level, morally sanctioning abortion.

While the NIH and ACOG policy recommendations are formally neutral on the subject of abortion, some of the reasoning supporting those recommendations explicitly connects cystic fibrosis testing to the supposed economic benefits of ending pregnancies—employing the language of cost-benefit analysis to rationalize aborting fetuses diagnosed with CF. The NIH report matter-of-factly notes the amount of money that our already-strained health care system will save by aborting fetuses with cystic fibrosis: "studies showed that the cost per identified CF fetus averted ranged from $250,000 to $1,250,000 for a Caucasian population of Northern European ancestry"; the "direct and ancillary costs associated with a CF birth" are estimated at $800,000.

Although the NIH and ACOG statements stipulate that CF screening should be strictly voluntary, women often feel pressured to have prenatal genetic testing done. Frequently, doctors simply order the tests as part of the "routine standard of care." And if previous experience with other prenatal tests is any guide, as CF testing becomes more common, it will lead inevitably to more abortions. As a physician, I have personally had patients tell me they were pressured by other physicians to have prenatal tests—or even to abort their babies. One woman for whom I provided prenatal care had a child diagnosed with a severe heart anomaly by ultrasound at 20 weeks gestation. The anomaly is virtually always lethal. Both a perinatologist and a geneticist recommended to the couple that the woman have an abortion. Despite being deeply offended by the recommendation and informing the physicians that she would carry the child to term, the suggestion was repeated numerous times. Her experience is far from anomalous. One woman's account in a survey published in 1999 by the Royal Association for Disability and Rehabilitation is a case in point:

> I was pregnant last year and came under severe pressure from every medical professional I saw about my decision to have no tests. Even when I pointed out that they were talking to a disabled person about the possibility of eliminating her child if it was disabled, they could not see how offensive it was.

As the armamentarium of prenatal screening techniques grows, such intolerance will only deepen—a problem made even more vexing by the grim fact that some of the fetuses being aborted today were wrongly diagnosed. At a meeting of the President's Council on Bioethics in June 2006, Dr. Benjamin Carson, a pediatric neurosurgeon, cautioned that blanket testing isolated

from clear purpose and moral understanding is gravely irresponsible:

> The question has come up [of] what have we historically done with data that we've acquired on newborns or prenatal individuals?…Almost all women in this nation now receive ultrasounds during the course of their pregnancy and you know, a number of things can be picked up on those ultrasounds, one of which is hydrocephalus. And almost uniformly, when there is an indication of hydrocephalus a recommendation for termination is made.…A significant number of those patients who decide not to go the abortion route it turns out end up with children who are normal, who never required a shunt, never required anything and yet had come to see me for a recommendation for abortion.

What is particularly chilling in Dr. Carson's remarks is that the assumption that disabled children must not be born is apparently so powerful that many babies who would have been *completely healthy* are being aborted as well.

In 1999, embryologist and IVF pioneer Robert Edwards said, "Soon it will be a sin for parents to have a child that carries the heavy burden of genetic disease. We are entering a world where we have to consider the quality of our children." Such considerations are becoming ever more routine. Discussing nascent life in the cold language of quality control and cost analysis, pressuring mothers to inspect their unborn children for potential defects, ending pregnancies because of the mere possibility that the child might be imperfect—this is the direction we are headed, toward a new eugenics. We must resolutely decide to take another course.

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