Cystic Fibrosis

Cystic fibrosis (CF) is an inherited disease that affects breathing and digestion. Advances in medical treatment have improved the outlook for affected children and adults. However, there is no cure. Most affected individuals survive into their 30s, though some die in childhood and others live to age 40 or beyond (1).

The abnormal gene that causes CF was discovered in 1989 (2). This discovery led to the development of a test, called a carrier screening test, that can help determine whether a couple is at increased risk of having a baby with CF. The test is routinely offered to couples who have a family history of CF. Many health care providers also make this test available to couples who are planning pregnancy or are pregnant. Couples must decide if testing is right for them.

How is CF inherited?
Genes come in pairs. To inherit CF, a child must receive two CF genes, one from each parent who "carries" a CF gene. A carrier is a healthy individual who has one normal gene and one abnormal gene in the pair.

When both parents carry an abnormal CF gene, there is a 25 percent (1-in-4) chance that the child will have CF. There is a 50-50 (50 percent) chance that the child will be a carrier like the parents. There also is a 25 percent chance that the child will be free of the abnormal gene and neither be a carrier, nor have the disease.

The couple has the same risks with each pregnancy. If only one parent is a carrier, there is no chance that their children will have CF. However, there is a 50-50 chance of each child being a carrier.

Who should have the carrier screening test?
The American College of Obstetricians and Gynecologists (ACOG) recommends that the carrier screening test be available to all couples who are planning pregnancy or are pregnant (6). Many health care providers hand out printed material on the test for couples to read. Those who are interested in testing can then discuss it further with their provider.

Health care providers are more likely to offer the test to couples considered at increased risk of having an affected child. Such couples include those with a family history of CF and Caucasians.

Whether or not a couple chooses to take the carrier screening test is a personal decision. A couple must decide what is right for them after learning about CF and discussing the test with their health care provider.

How is the test done?
The health care provider will take a sample of blood or saliva. The sample will be sent to a lab to test for the abnormal gene.

Who is most likely to be a carrier?
One in 30 Americans—more than 10 million people—carries a gene for CF (1, 3). Someone with a family history of CF is more likely to carry a CF gene than someone from an unaffected family. Caucasians have a 1 in 29 chance of carrying the gene, compared to 1 in 46 for those of Hispanic background, 1 in 65 for African-Americans, and 1 in 90 for Asian-Americans (1, 6).

What happens if the test shows that a woman is a carrier?
The next step is to test her partner. A baby is not at risk of CF unless both parents carry the abnormal gene. Fortunately, this situation is fairly uncommon. In those of Caucasian background, the chances that both partners carry the CF gene is only about 1 in 800; the risk is less in other groups (6).

It's important to keep in mind, however, that the test is not 100 percent accurate. Scientists have discovered more than 1,300 different mutations (changes) in the CF gene, and the test looks only for some of the most common of these (7). So even when the test shows that a person is not a carrier, there's a very small chance that he or she carries an abnormal gene.

What happens if both parents are carriers?
A couple in which both partners are carriers should consider consulting a genetic counselor. A genetic counselor can discuss the risks to their future children and the option of prenatal testing (using amniocentesis or CVS) to diagnose or rule out CF in a fetus. Each child of parents who both carry an abnormal CF gene has a 25 percent chance of inheriting CF. This means that in three out of four cases, the prenatal test will show that the fetus does not have CF.

When the fetus is affected, parents can take the time before delivery to learn more about the disease and locate appropriate specialists. Prenatal testing cannot, however, tell how seriously affected the baby may be.

Adapted from the March of Dimes Website.

For references and more information, visit www.marchofdimes.com or ask your midwife.