Both the American College of Obstetricians and Gynecologists and the American College of Medical Genetics and Genomics recommend that cystic fibrosis (CF) carrier testing be offered to all women of reproductive age. This test is best done before pregnancy, but is also available in early pregnancy.

This brochure answers key questions that concern those considering CF carrier screening.

What is cystic fibrosis (CF)?

CF is a serious and potentially lethal lifelong illness characterized by chronic lung infections and difficulties digesting fat requiring most individuals to take pills with every meal to assist digestion. Daily respiratory therapy, frequent antibiotics, and pills for digestion of food constitute the main treatment for CF, a disorder with a shortened lifespan. Currently there is no cure and ultimately lung transplantation may be necessary.

What causes CF?

CF is a genetic disorder caused by inheriting two genetic changes (mutations), one gene change from each parent. An individual with only one CF gene change is a carrier who will not have any health problems.

What is the chance of you being a carrier of CF?

The chance of being a carrier depends on your ethnic background. About 1 in 25 whites, 1 in 46 Hispanics, 1 in 65 African Americans, and 1 in 90 Asian Americans are carriers for CF. For more detailed information on this subject, the book *Your Genes, Your Health: A Critical Family Guide that could Save Your Life* by Aubrey Milunsky, M.D., published by Oxford University Press, can be consulted. If you have a family history of CF, your chance of being a carrier is considerably greater than if you had no such history.

Why have CF carrier screening?

Knowing the carrier status of yourself and your partner places you in control and allows personal decision-making. If you are both carriers, prenatal diagnosis in early pregnancy is possible. This would allow you to make decisions such as making preparations to have a child with lifelong special health care needs, electing to stop a pregnancy with CF, or avoiding CF. Preimplantation embryo diagnosis is also available.

What are the limitations of carrier screening?

A large number of gene changes (mutations) have been reported in the CF gene. The current recommendation is to test the most common 23 mutations. The Center for Human Genetics provides analysis of at least 110 CF mutations. Such testing typically yields up to 93% detection for whites, with lower detection rates in other ethnicities. Full gene analysis yields detection rates greater than 98% for all ethnicities. Normal carrier screening results will reduce, but not eliminate, the chance of being a CF carrier.

What if both you and your partner are CF carriers?

If both partners are carriers, they have a 25% chance to have a child with CF. An appointment to see a genetic counselor/ clinical geneticist at the Center for Human Genetics is advised. Appointments can be made by calling 617-492-7083.

What if one partner is a CF gene mutation carrier?

If pregnancy is planned or pregnancy is less than 3 months along, and one partner carries a gene mutation, an important option is to analyze (called sequencing) the entire CF gene. Sequencing and deletion analysis will detect greater than 98% of the close to 2,000 known, mostly rare, mutations.

What if a male CF gene mutation carrier has no sperm?

Couples who experience infertility have tests including sperm counts. Too few or no sperm leads to other tests, including sequencing of the CF gene. Frequently, another mutation is found. This finding is important since pregnancy can still be achieved by aspirating sperm from the testis, and then considering prenatal or preimplantation diagnosis.

Other available carrier screening*

If your ancestors were…	Genetic disorder	Chance of being a carrier
Ashkenazi Jewish	Tay Sachs disease	1 in 31
	Canavan disease	1 in 41
	Gaucher disease	1 in 18
	Familial Dysautonomia	1 in 31
	Factor XI deficiency	1 in 8
	Glycogen storage disease type 1A	1 in 71
	Maple Syrup Urine disease, type 1B	1 in 81
	Fanconi Anemia type C	1 in 89
	Niemann Pick, type A	1 in 90
	Bloom Syndrome	1 in 107
	Mucolipidosis, type IV	1 in 127
Black	Sickle cell disease	1 in 12
Armenian, Turkish, Arabic, or Sephardic Jewish	Familial Mediterranean Fever	1 in 8

*If both partners are carriers, prenatal diagnosis is available.

There are a few thousand other genetic disorders for which DNA tests for diagnosis or carriers are available (see our website www.chginc.org).

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