

## Cystic Fibrosis Carrier Screening

Cystic Fibrosis is an inherited disease affecting about 1 in 3300 Caucasians per year in the United States. The disorder causes problems with breathing and digestion, and the severity of symptoms can vary. The body produces thick mucous in these areas, leading to pneumonia, diarrhea, and poor growth. Some people are only mildly affected, but others have a more severe form of the disease that can lead to an early death. Affected individuals require lifelong medical care. Although there is no cure for CF, a great deal of research is being done on new treatments.

The American College of Obstetrics and Gynecology (ACOG) has recommended that CF carrier testing be offered to *“individuals with a family history of CF; reproductive partners of people affected with CF; couples in whom one or both partners is Caucasian and are currently planning a pregnancy or seeking pregnancy care. It is further recommended that screening be made available to couples in other racial and ethnic groups who are at a lower risk and for whom the test may be less sensitive.”*

To be affected by the disease, an individual must inherit the CF gene from **each** parent, thus carrier testing is done on adults. If **both** parents are carriers, each of their children has a 1 in 4 chance of having CF. Carrier testing is done with a blood sample, and the risk of being a carrier is much higher in Caucasians than other racial or ethnic groups. If there is no one in your family with CF, your risk of being a carrier is shown in the following table:

Racial Group:	Caucasian	Hispanic	African American	Asian American
Risk:	1 in 29	1 in 46	1 in 65	1 in 90

CF carrier testing is recommended for all women planning a pregnancy or who are currently pregnant. If she tests positive, her partner should also be tested to determine the risk to their children. If both parents test positive as CF carriers, genetic testing is advised for evaluation of the unborn child. If a woman is found to be positive and her partner negative, both may be referred for genetic counseling. The risk for having a child with CF, however, is low.

Although CF testing is recommended by ACOG and the National Institutes of Health (NIH), not all patients desire genetic testing for themselves or their children. You may wish to discuss your questions about CF carrier testing with your provider before making your decision on how to proceed. Early diagnosis of CF is available for your newborn through the **Newborn Supplemental Screening Test**, which is mandatory for all newborns before discharge from the hospital.

We urge you to contact your insurance company regarding coverage for CF carrier testing. You should also investigate whether the lab with which your insurance company participates can perform the test, and if they do not, what options are available to you. The cost of CF carrier testing varies from \$275 to \$575 per person.