

CYSTIC FIBROSIS SCREENING

What is Cystic Fibrosis?

Cystic fibrosis (CF) is an inherited disorder that is usually first apparent in early childhood. It causes thick secretions in the lungs, pancreas, and intestines (bowels). It often leads to chronic and severe breathing problems and digestion problems. CF is also associated with menstrual problems and reproductive problems. It **does not** cause mental retardation or external physical deformities. The average life expectancy for women and men with CF is 28.3 years and 31.1 years respectively; however, some people with CF live much longer.

What Causes CF?

CF is an inherited disease where both parents need to be carriers for their children to be at risk (autosomal recessive). CF results when a protein called CFTR is either missing or not functioning well. The gene that codes for this protein has been mapped to chromosome 7. Over 900 mutations in the CFTR gene that cause CF have been identified.

Is There a Cure or Treatment for CF?

No cure is currently available for CF. Symptoms are managed by chest physical therapy and medicines that thin the thick secretions, treat infections, and aid in digestion.

Am I at Risk of Being a CF Carrier?

CF is carried by 1 in 29 Caucasians, 1 in 65 African Americans, 1 in 46 Hispanics, and 1 in 90 Asians. About 1 in 2,500 Caucasians actually have CF. The rate of CF in nonwhites is much lower. The American College of Obstetricians and Gynecologists (ACOG) recommends that screening for CF be offered to:

- ◆ Adults with a family history of CF
- ◆ Spouses/Partners of individuals with CF
- ◆ White couples of European and Ashkenazi Jewish heritage who are interested in having children

Is CF Testing Required? Could the Test Harm My Pregnancy? When Should Testing be Performed?

No one is required to have CF testing. If you have any questions about CF testing please ask your obstetrician or midwife at your next office visit. The test poses no risk to your developing baby as it only requires collection of blood from your arm. Ideally, screening for CF should be performed before conception or during early pregnancy.

What if I Test Positive as a CF Carrier?

If you test positive as a CF carrier your partner would also be offered testing. You would then be offered counseling. If it is determined that you and your partner are at an increased risk of having a child with CF then you would be referred to a genetics expert for further counseling and possible testing.

How Reliable is the Test for CF Carrier Status?

Testing will pick up about 85% of carriers of CF. It is very uncommon to have a test that is falsely positive.