

When You're Expecting, Expect the Best.

Cystic Fibrosis Carrier Screening

PENN Ob/Gyn Care provides cystic fibrosis carrier screening, available to you on a voluntary basis. The American College of Obstetricians and Gynecologists recommends carrier screening to couples planning a pregnancy, or those already pregnant to help determine if a couple is at risk of having a child with cystic fibrosis. Screening is a personal decision based on you and your partner's level of risk, genetic history, family situation, and your religious/spiritual beliefs.

You should discuss any concerns or questions you may have about cystic fibrosis carrier screening with your obstetrician. The following information will help you better understand the condition and screening process.

What is cystic fibrosis?

Cystic fibrosis is a genetic disease that causes respiratory and digestive problems and affects about 1 in every 3,300 people in the United States. Symptoms range from mild to severe. Those with cystic fibrosis often have chronic lung problems, such as pneumonia, which worsen over time. Cystic fibrosis also affects digestion, resulting in diarrhea and poor growth. Treatments are available but in general people with cystic fibrosis have a shortened lifespan. Men with cystic fibrosis are usually infertile. Cystic fibrosis does not affect intelligence.

What is the purpose for cystic fibrosis carrier screening?

The cystic fibrosis carrier screening gives detailed information about whether or not individuals are carriers, and about their risks of having a child affected with cystic fibrosis. The test requires a simple blood sample and results are usually ready within 7 to 14 days.

If both partners are cystic fibrosis carriers, prenatal testing by chorionic villi sampling or amniocentesis can be performed to determine if the unborn baby has inherited the parents' abnormal cystic fibrosis genes. Testing the baby after birth is also an option if a couple does not want prenatal diagnosis.

Will the cystic fibrosis carrier screen detect all cystic fibrosis carriers?

No. There are extremely rare abnormalities (mutations) in the cystic fibrosis gene that the current screening cannot detect. Negative results from cystic fibrosis carrier screening significantly lowers, but does not completely eliminate the risk of being a carrier.

Could I be a carrier?

Every person has two copies of a gene, one inherited from each parent. If a person has one normal and one abnormal cystic fibrosis gene, that person is a carrier. Having one abnormal copy of the cystic fibrosis gene does not cause the disease therefore a carrier does not have any symptoms.

How is cystic fibrosis inherited?

If both parents are carriers of an abnormal cystic fibrosis gene, there is a chance that each parent will pass the abnormal gene on to their child. When both partners in a couple are carriers, any child they have has a one-in-four (25%) chance to inherit the disease. Having two copies of the abnormal cystic fibrosis gene results in cystic fibrosis.

Is cystic fibrosis screening required?

No. The decision to accept or decline screening is entirely up to the individual or couple.