PATIENT EDUCATION

The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

Cystic Fibrosis: Prenatal Screening and Diagnosis

CF is a disease that affects a person's long-term health and lifespan. CF is a ***genetic disorder*** caused by a ***gene*** that is passed from parent to child. Carrier screening allows parents-to-be to find out their chances of having a child with CF. If you are already pregnant, a prenatal ***diagnostic test*** allows you to find out if your ***fetus*** actually has CF or is a carrier.

You Need to Know

- the cause, risk factors, and symptoms of ***cystic fibrosis (CF)***
- how ***carrier screening*** is done
- how prenatal diagnostic testing is done
- what options ***carriers*** may have for future pregnancies

About Cystic Fibrosis

CF is a lifelong illness that can affect all of the organs of the body. It often causes problems with digestion and breathing. It does not cause intellectual disability or change a person's appearance. There is no cure for CF, but it can be treated.

What are the symptoms of cystic fibrosis?

The symptoms of CF can vary in type and severity. Many people with CF produce a thick, sticky mucus in their bodies. This mucus builds up and clogs the lungs. This makes it hard to breathe and can lead to infection. CF also can make it hard for the body to break down and absorb food.

How does cystic fibrosis affect a person's health?

About 1 in 6 people with CF have a mild form. But in most people, CF seriously affects a person's health. Treatments are available, but the disease gets worse the longer a person has it. The average lifespan of a person with CF is 37 years. Those with a milder form can live into their 50s. Many people with CF attend school, have careers, and lead full lives.

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Is there a cure for cystic fibrosis?

Not yet, but new drugs and treatments have improved the outlook for people with CF. To treat lung problems, most children with CF need to have daily physical therapy. This therapy helps clear mucus from the lungs. It is easy to do and can be done by parents or other family members.

What does it mean if someone is a cystic fibrosis carrier?

It takes two genes—one from the mother and one from the father—for a person to have CF. If a person has only one copy of a gene for CF, he or she is a carrier. Carriers often do not know that they have a gene for CF. They usually do not have symptoms or may have only mild symptoms.

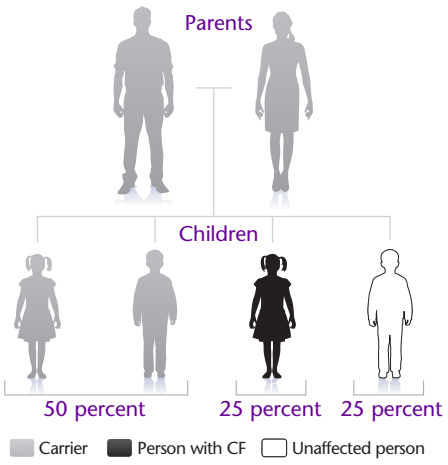
Are some groups more at risk of being carriers?

The risk of being a CF carrier is higher in families with a history of CF. The risk also is higher for certain racial and ethnic groups. CF occurs more often in non-Hispanic white people than in other racial groups.

Carrier Screening

Carrier screening for genetic disorders is voluntary. You can choose to have carrier screening or not to have carrier screening. You can choose to be tested for many genetic conditions or just a few. You also may think about the timing of when to have carrier screening.

What Are the Chances of Having a Baby With Cystic Fibrosis?



If both partners are CF carriers, there are three possible outcomes:

1. There is a 1-in-2 (50 percent) chance the baby will be a carrier, like you and your partner. Being a carrier usually will not affect the health of the baby, but he or she could have a child with CF in the future.
2. There is a 1-in-4 (25 percent) chance the baby will have CF.
3. There is a 1-in-4 (25 percent) chance that the baby will not have CF and will not pass on the disease to future children.

What are the options for carrier screening?

- Testing based on your ethnicity if your ethnic group is known to be at higher risk (***ethnic-based screening***)
- Testing for many disorders at once (***expanded carrier screening***)
- Testing for just a few specific disorders

When should I have carrier screening?

Carrier screening can be done before pregnancy or during pregnancy.

- If you have carrier screening before you get pregnant and both you and your partner are carriers, you have more options (see "Future Pregnancies").
- If you have carrier screening while you are pregnant, you have fewer options.

Who should be screened for cystic fibrosis?

Carrier screening for CF is offered to all women who are thinking about getting pregnant or who are already pregnant. It is your choice whether to have this screening.

What does carrier screening for cystic fibrosis involve?

You will be asked to give a sample of blood, saliva, or tissue from the inside of your cheek.

Who should be tested first, me or my partner?

- You usually are tested first. If results show that you are a carrier, your partner is tested.
- If your partner has a family history of CF, he may be tested first.
- If you already are pregnant, you and your partner can be tested at the same time.

How do I make decisions about carrier screening?

Your ***obstetrician–gynecologist (ob-gyn)***, other ***obstetric care provider***, or a ***genetic counselor*** can help you choose the approach that addresses your concerns and also meets current recommendations for carrier screening.

What do the CF carrier screening test results mean?

- Negative result—A negative result means that your chance of being a CF carrier is small. But no screening test checks for every known CF ***mutation***. For this reason, if your test result is negative, there still is a very small chance that you could be a carrier of a mutated gene that was not detected by the test. If you have a negative test result but a family history of CF, you also may be tested for the specific mutation in your family if that information is available.
- Positive result—If your test result is positive, it means that you are a CF carrier. The next step is to test your partner. Both partners must be CF carriers for a fetus to have CF. If your partner has a negative test result, the chance that the fetus will have CF is small.

- Both partners positive—If both partners are carriers, a couple has a 1-in-4 chance of having a child with CF (see the box "What Are the Chances of Having a Baby With Cystic Fibrosis?").

Should I tell my family if I am a carrier?

It's up to you. You may want to share this information with family members to help them plan their pregnancies. They may be at risk of being carriers themselves. But there is no law that states that you have to share information.

Prenatal Diagnostic Testing

If you are pregnant and you and your partner are CF carriers, prenatal diagnostic testing can be done to detect whether the fetus has CF. CF testing also can be done if you are having diagnostic testing for other genetic disorders and you do not know your or your partner's CF carrier status.

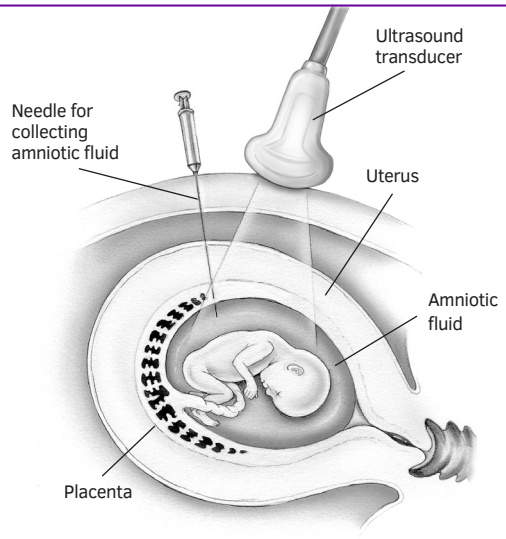
Is it possible to find out if a fetus has cystic fibrosis?

Testing can be done to learn if a fetus has CF or is a carrier. This is called prenatal diagnostic testing. This testing can be done as early as 10 weeks of pregnancy.

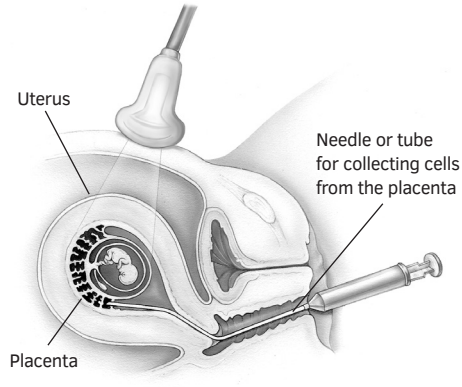
How is prenatal diagnostic testing done?

Prenatal diagnostic tests to detect CF and other disorders include ***amniocentesis*** and ***chorionic villus sampling (CVS)***.

- Amniocentesis usually is done between 15 and 20 weeks of pregnancy, but it also can be done up until you give birth. A very thin needle is used to take a small sample of ***amniotic fluid*** for testing. The cells are studied to detect the presence of the CF gene.
- CVS is done between 10 and 13 weeks of pregnancy. A small sample of tissue is taken from the ***placenta.*** The cells are then checked for the presence of the CF gene.



Amniocentesis. In this procedure, a small sample of amniotic fluid is removed with a needle to be studied.



Chorionic villus sampling. In this procedure, a small sample of cells (chorionic villi) is removed from the placenta to be studied.

Can I have diagnostic testing if I am using in vitro fertilization?

For couples using in ***vitro fertilization (IVF)*** to get pregnant, there is another testing option called ***preimplantation genetic testing***. Before an ***embryo*** is transferred to a woman's uterus, it can be tested for CF and certain other genetic disorders. The only embryos transferred are those that do not test positive for the disorders.

What do diagnostic test results mean?

The results of these prenatal tests can tell you with a high degree of certainty whether the fetus has CF or is a CF carrier. The results cannot tell you how severe the disease will be if the fetus has the disorder.

Making Decisions

There is no cure for CF. If you find out that your fetus has CF, you have options:

- You may choose to continue the pregnancy and prepare for a child with CF. Couples can use this time to learn as much as possible about the disease, current treatment options, and the experiences of other families who have a child with CF.
- Another option is to end the pregnancy. Each state has its own laws on pregnancy termination. Your ob-gyn or other obstetric care provider can answer questions you may have. You also may want to discuss the decision with your partner, counselors, and close friends.

Future Pregnancies

If tests show that both partners are carriers, it means that in each pregnancy the fetus will have a 1-in-4 chance of having CF. If you want to know whether your fetus has CF, you will need to have amniocentesis or CVS in each pregnancy.

What are my options for having children in the future?

- You can accept the level of risk and get pregnant.
- You may choose to have prenatal diagnostic testing in each pregnancy, or you may not.
- You can choose not to have children.
- You can adopt.
- You can use donor sperm or donor eggs (but the donor should be screened for CF).
- You can use IVF with your own sperm and eggs, and then use preimplantation genetic testing to see if the fertilized egg has CF or is a CF carrier.

Discuss these options with family, friends, your ob-gyn or other obstetric care provider, or genetic counselor when you are considering future pregnancies. If you are a carrier and have a new partner for another pregnancy, you should consider testing for the new partner.

Your Takeaways

1. You can find out if you are a cystic fibrosis carrier with a simple screening test.
2. If you and your partner are carriers, there is a 1-in-4 chance your child will have CF.
3. If you are pregnant and find out that you and your partner are carriers, you can have prenatal diagnostic testing to learn if the fetus has CF or is a carrier.
4. Your ob-gyn, other obstetric care provider, or genetic counselor can help guide you through these steps and help you make decisions that are right for you.

Terms You Should Know

Amniocentesis: A procedure in which amniotic fluid and cells are taken from the uterus for testing. The procedure uses a needle to withdraw fluid and cells from the sac that holds the fetus.

Amniotic Fluid: Fluid in the sac that holds the fetus.

Carriers: People who shows no signs of a disorder but could pass the gene to their children.

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Cystic Fibrosis (CF): An inherited disorder that causes problems with breathing and digestion.

Diagnostic Test: A test that looks for a disease or cause of a disease.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

Ethnic-Based Screening: Screening recommended for people who belong to an ethnic group or race that has a high rate of carriers of a specific genetic disorder.

Expanded Carrier Screening: A blood test to screen for a large number of genetic disorders.

Fetus: The stage of human development beyond 8 completed weeks after fertilization.

Gene: A segment of DNA that contains instructions for the development of a person's physical traits and control of the processes in the body. The gene is the basic unit of heredity and can be passed from parent to child.

Genetic Counselor: A health care professional with special training in genetics who can provide expert advice about genetic disorders and prenatal testing.

Genetic Disorder: A disorder caused by a change in genes or chromosomes.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Mutation: A change in a gene that can be passed from parent to child.

Obstetric Care Provider: A health care professional who cares for a woman during pregnancy, labor, and delivery. These professionals include obstetrician–gynecologists (ob-gyns), certified nurse–midwives (CNMs), maternal–fetal medicine specialists (MFMs), and family practice doctors with experience in maternal care.

Obstetrician–Gynecologist (Ob-Gyn): A doctor with special training and education in women's health.

Placenta: An organ that provides nutrients to and takes waste away from the fetus.

Preimplantation Genetic Testing: A type of genetic testing that can be done during in vitro fertilization. Tests are done on the fertilized egg before it is transferred to the uterus.

Sperm: A cell made in the male testicles that can fertilize a female egg.

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