KAISER PERMANENTE®

INFORMATION SHEET

CYSTIC FIBROSIS

Positive Carrier Screening Result

You have recently learned that you are a cystic fibrosis (CF) carrier. Here is important information about being a CF carrier.

Being a carrier does not affect your health. Everyone carries genes that do not work. Being a CF carrier means that one copy of the cystic fibrosis gene does not work properly. Carriers have a second copy of the cystic fibrosis gene that works normally. CF carriers do not develop cystic fibrosis. In fact, one of your parents is probably a CF carrier just like you. If you have brothers or sisters, they could also be carriers.

If you are pregnant, the next step is to test your partner. Both parents must be CF carriers to be at risk for a baby with CF. If one parent has a negative (normal) test result, the chance that a baby will have CF is very small. Testing can be done at any Kaiser Permanente laboratory. A genetic counselor will let you know how to arrange testing.

Why test your partner?

- You want to know the chance that your baby could have CF.
- You want to be prepared if your baby is found to be at risk for CF.
- You might consider testing the baby for CF during pregnancy.

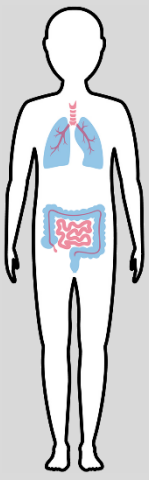
Why would you not test your partner?

- You do not want to learn about a risk for CF during pregnancy.
- The result would not change anything you do during pregnancy.

What happens if my partner is a CF carrier?

When both parents are CF carriers, there is a 1 in 4 (25%) chance for a baby to develop cystic fibrosis. There is also a 3 in 4 (75%) chance that a baby will not have CF. If your partner is a CF carrier, you will be offered prenatal testing to find out if your baby could have cystic fibrosis. A procedure, such as amniocentesis or CVS, is able to diagnose CF during pregnancy. This test allows parents to decide whether or not to continue the pregnancy. You could also choose to wait until after birth to test your baby.

What is cystic fibrosis?



Cystic fibrosis is an inherited disease that causes many health problems. Extra thick mucus in the lungs causes congestion, coughing, and frequent infections. This damages the lungs and can lead to lung failure. A person with CF can also have problems digesting food. This makes it harder to grow and gain weight. CF does not affect learning.

Signs of CF can start soon after birth. The disease gets worse over time. The average life span for people with CF is around 40 years old, though some people live longer, and others do not survive past childhood.

There is no cure for cystic fibrosis at this time, but research is being done to find ways to keep people with CF as healthy as possible. Early diagnosis and treatment can help. As treatments improve, people with CF are living longer. Treatment for CF usually includes daily chest therapy and medications, frequent doctor visits, and overnight hospital stays.

Cystic Fibrosis Carrier Screening

Anyone can be a CF carrier, even if there is no history of CF in the family. The chance to be a CF carrier depends on a person's ancestry or ethnic background. Carrier screening detects most CF carriers.

Approximate carrier rates by ancestry or ethnic background:

- Caucasian: 1 in 25 (4%)
- Jewish: 1 in 25 (4%)
- Hispanic: 1 in 60 (2%)
- African (Black): 1 in 60 (2%)
- Asian: 1 in 90 (1%)
- Mixed ancestry/other: Varies (1-4%)

The chance could be higher if there is any history of CF in the family. Before having carrier screening, it is important to let your provider know if someone in the family had CF or is a CF carrier. Special testing may be offered.

Limits of carrier screening: A small number of carriers have changes in the CF gene that cannot be found by routine testing. This can lead to an apparently normal result in someone who really is a carrier.

Newborn Screening for Cystic Fibrosis

All babies are screened for cystic fibrosis in the first few days after birth. Newborn screening helps diagnose CF as early as possible. When the screening test finds a baby at higher risk, special testing is done to check for CF.

Limits of newborn screening:

- Positive (high risk) results can happen in babies without CF.
- Newborn screening misses some babies with CF.
- Screening may identify babies who are CF carriers and do not have CF.
- Screening will not identify all CF carriers.