

Familial Dysautonomia

Positive Carrier Screening Result

You have recently learned that you are a familial dysautonomia (FD) carrier. Here is important information about being an FD carrier.

Being a carrier does not affect your health. Everyone carries genes that do not work. Being an FD carrier means that one copy of the familial dysautonomia gene does not work properly. Carriers have a second copy of the familial dysautonomia gene that works normally. FD carriers do not develop familial dysautonomia. In fact, one of your parents is probably an FD 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 FD carriers to be at risk for a baby with FD. If one parent has a negative (normal) test result, the chance that a baby will have familial dysautonomia 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 FD.
- You want to be prepared if your baby is found to be at risk for FD.
- You might consider testing the baby for FD during pregnancy.

Why would you not test your partner?

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

What happens if my partner is an FD carrier?

When both parents are FD carriers, there is a 1 in 4 (25%) chance for a baby to develop familial dysautonomia. There is also a 3 in 4 (75%) chance that a baby will not have FD. If your partner is an FD carrier, you will be offered prenatal testing to find out if your baby could have familial dysautonomia. A procedure such as amniocentesis or CVS is able to diagnose FD 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 familial dysautonomia?



Familial dysautonomia is a rare inherited disease that affects nerves that control involuntary actions, like digestion, breathing, and blood circulation. FD also affects nerves that sense pain and control body temperature. A person with FD does not make enough of a protein called IKAP. When there is not enough IKAP, some nerves do not work normally.



FD affects many parts of the body and can shorten life expectancy. Babies with FD have weak muscles, feeding problems, and do not make tears when they cry. A person with FD is prone to wide swings in blood pressure and may have episodes of vomiting. Breathing problems, lung infections, balance problems, and eye problems are also common.

There is no cure for familial dysautonomia at this time, but treatment can help with symptoms. Research is being done to find new ways to treat FD.

Familial dysautonomia Carrier Screening

Anyone can be an FD carrier, even if there is no history of FD in the family. The chance to be an FD carrier is highest for a person with Ashkenazi (Central or Eastern European) Jewish ancestry. For this reason, carrier screening for FD is routinely offered only when someone has Jewish ancestry. Carrier screening detects 99% of carriers in this population.



Approximate carrier rates by ancestry or ethnic background:

- Jewish: 1 in 30 (3%)
- Non-Jewish: unknown (very rare)

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

Limits of carrier screening:

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

Newborn Testing for Familial dysautonomia



Testing can be requested soon after birth for babies who are at risk to develop FD. An early diagnosis can allow medical care to start as soon as possible. Familial dysautonomia is not part of routine newborn screening.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional. If you have persistent health problems, or if you have additional questions, please consult your doctor.

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