

Canavan Disease

Positive Carrier Screening Result

You have recently learned that you are a Canavan disease (CD) carrier. Here is important information about being a CD carrier.

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

Why would you not test your partner?

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

What happens if my partner is a CD carrier?

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



Canavan is an inherited disease that affects the brain. A person with CD does not make enough of an enzyme called aspartoacylase (ASPA). This enzyme normally breaks down waste products in the brain. When there is not enough ASPA, waste products build up and damage the nerve cells in the brain. This affects movement, vision, and learning.



Babies with CD appear normal at birth but muscle weakness and delays in development start by 3 to 9 months of age. By 2 to 3 years of age, most children with CD begin to lose motor skills and mental abilities. CD usually leads to death in late childhood or teenage years.

There is no cure for Canavan disease at this time, but improvements in medical care has increased survival into the teens. Treatment can help with some symptoms but does not stop the disease. Research is being done to find new ways to treat CD.

Canavan Carrier Screening

Anyone can be a CD carrier, even if there is no history of CD in the family. The chance to be a CD carrier is highest for a person with Ashkenazi Jewish ancestry. For this reason, carrier screening for CD is routinely offered only to couples who both have Jewish ancestry. Carrier screening detects 99% of carriers in this population.



Approximate carrier rates by ancestry or ethnic background:

- Jewish: 1 in 50 (2%)
- Non-Jewish: unknown (very low)

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

Limits of carrier screening:

- A small number of carriers have changes in the CD 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 Canavan Disease



Testing can be requested soon after birth for babies who are at risk to develop CD. An early diagnosis can allow medical care to start as soon as possible. Canavan disease 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.