What Does It Mean To Be A Carrier of Canavan Disease?

You’ve recently learned that you are a carrier for Canavan disease. You should know that being a carrier is not an illness and does not affect your health. In fact, you would probably not have found out that you are a carrier without having a special blood test. Being a carrier simply means that one copy of the gene that can cause Canavan disease has a change that keeps it from working properly. You have a second copy of the Canavan gene that is working normally.

What is a gene and what does it do?
Genes are our body’s “instructions” that determine our physical characteristics, such as blood type and hair color. They are located on chromosomes, which are found in almost every cell of our bodies. We inherit one set of chromosomes from each of our parents; therefore, our chromosomes, and the thousands of genes located on each of them, come in pairs.

The Canavan gene pair tells our bodies how to make a specific enzyme, called aspartoacylase (ASPA). This enzyme is required for brain development and function. If a person has one copy of the Canavan gene that does not work properly, he or she is called a carrier of Canavan disease. As long as a person has one working copy of this gene, the body has enough ASPA to do its job, which is why he/she will never develop Canavan disease.

What is Canavan disease?
When babies inherit two nonworking copies of the Canavan gene, they are unable to make enough ASPA. Without this enzyme, the body is unable to break down another substance, called NAA, which builds up and causes brain damage, mental retardation, tremors and an inability to move muscles. Babies appear normal until about three to nine months old and then begin to have problems with activities involving their muscles, like rolling over or grabbing. Developmental progress often stops by two to three years of age. Most individuals with Canavan disease die before the age of five; however, some survive longer.

How does one become a carrier?
Since all of our genes are inherited from our parents on their chromosomes, individuals who are Canavan disease carriers have inherited their non-working gene from one of their parents. This means that brothers and sisters of a carrier have a 50% chance to be a carrier.

Can anyone be a carrier of Canavan disease?
Yes. Canavan carriers can be found in any nationality and ethnic group but are much more commonly found in Jewish individuals whose ancestors came from Eastern Europe (Ashkenazi Jews). In the Eastern European Jewish population, about one in every 40 individuals carries a change in the Canavan gene.
**Inheritance of Canavan Disease**

When both parents are Canavan carriers there is:

- A 25% chance that the baby is not a Canavan carrier and does not have Canavan disease.
- A 50% chance that the baby is a Canavan carrier just like his or her parents.
- A 25% chance that the baby has Canavan disease.

**Is there a cure for Canavan disease?**
No. Although research is being conducted, there is nothing that can be done to change the course of this disease at this time. The treatment of Canavan disease is aimed at easing the problems associated with the disease.

**Can being a carrier lead to having Canavan disease?**
No. Carriers will never develop the disease because they have one working copy of the ASPA gene. Their bodies make enough of the enzyme to be healthy.

**Can my children have Canavan disease?**
Since children receive half of their genetic information, like eye and hair color, from their mother and half from their father, the answer to this question depends on whether or not your partner is a carrier.

If your partner is not a Canavan carrier, then your children will not have Canavan disease. With each pregnancy, you will have a 50% chance of having a child who is a Canavan carrier just like you and a 50% chance of having a child with two working copies of the ASPA gene. None of these children will have Canavan disease.

If your partner also is a Canavan carrier, then there are three possible pregnancy outcomes, including the possibility of a child with Canavan disease (see diagram above).

**What choices do I have if my partner is also a carrier of Canavan disease?**
There are several options available if both you and your partner are carriers. In order to find out before birth if a baby has Canavan disease, prenatal testing can be performed as early as the 10th week of pregnancy. If the results are normal, the parents can be reassured. If the results show that the baby will be affected, the parents can be better prepared, and they can make informed decisions regarding continuation or termination of the pregnancy.

**Will my child be tested for Canavan disease at birth?**
Although babies born in California have newborn screening for many different genetic disorders, babies are not routinely tested for Canavan disease. Carrier testing in the parents allows families to know if their baby may be at risk for this condition.

**Contact information for KP GENETICS DEPARTMENTS**

<table>
<thead>
<tr>
<th>City</th>
<th>Phone</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fresno</td>
<td>(559) 324-5330</td>
</tr>
<tr>
<td>Oakland</td>
<td>(510) 752-6298</td>
</tr>
<tr>
<td>Sacramento</td>
<td>(916) 614-4075</td>
</tr>
<tr>
<td>San Francisco</td>
<td>(415) 833-2998</td>
</tr>
<tr>
<td>San Jose</td>
<td>(408) 972-3300</td>
</tr>
</tbody>
</table>

---

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.

© 2008, The Permanente Medical Group, Inc. All rights reserved. Regional Genetics Department.