FAMILIAL DYSAUTONOMIA

Everything you need to know about being a carrier

What Does It Mean To Be A Carrier of Familial Dysautonomia?

You've recently learned that you are a carrier for familial dysautonomia. 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 familial dysautonomia has a change that keeps it from working properly. You have a second copy of the familial dysautonomia 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 familial dysautonomia gene pair tells our bodies how to make a specific protein called ikB kinase complex–associated protein (IKAP). Normal IKAP protein is needed for proper development of the nervous system. If a person has one copy of the familial dysautonomia gene that does not work properly, he or she is called a **carrier** of familial dysautonomia. As long as a person has one working copy of this gene, the body has enough IKAP to do its job, which is why he/she will never develop familial dysautonomia.

What is familial dysautonomia?

When babies inherit two nonworking copies of the familial dysautonomia gene, they are unable to make enough IKAP protein in the nervous system. This causes problems in nerve cells that control involuntary actions, such as digestion, breathing, making tears, and regulating blood pressure. It also affects the nerves related to taste and pain and temperature perception.

Early signs include poor muscle tone (hypotonia), poor feeding and growth, lack of tears, frequent lung infections, and difficulty maintaining body temperature. Older individuals can have vomiting, poor balance, and blood pressure problems. The average life expectancy is now about 40 years old.

How does one become a carrier?

Since all of our genes are inherited from our parents on their chromosomes, individuals who are familial dysautonomia 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.

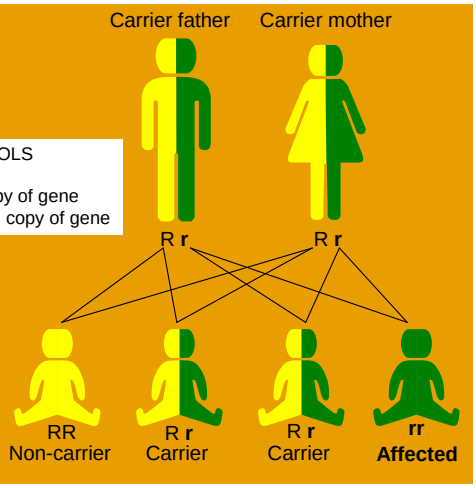
Can anyone be a carrier of familial dysautonomia?

All confirmed cases of familial dysautonomia have occurred in Jewish individuals whose ancestors came from Eastern Europe (Ashkenazi Jews). In the Eastern European Jewish population, about one in every 30 individuals carries a change in the familial dysautonomia gene. It would be extremely rare to find a person who is a carrier for this condition and does not have Eastern European Jewish ancestors.

Inheritance of familial dysautonomia

When both parents are familial dysautonomia carriers there is:

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



Is there a cure for familial dysautonomia?

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 familial dysautonomia is aimed at easing the problems associated with the disease.

Can being a carrier lead to having familial dysautonomia?

No. Carriers will never develop the disease because they have one working copy of the IKAP gene. Their bodies make enough of the protein to be healthy.

Can my children have familial dysautonomia?

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

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

To find out the chance of having a baby with familial dysautonomia, your partner should be tested.

What choices do I have if my partner is also a carrier of familial dysautonomia?

There are several options available if both you and your partner are carriers. In order to find out before birth if a baby has familial dysautonomia, 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 familial dysautonomia at birth?

Although babies born in California have newborn screening for many different genetic disorders, babies are not routinely tested for familial dysautonomia. Only carrier testing in the parents allows families to know if their baby may be at risk for this condition.

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