

SPINAL MUSCULAR ATROPHY: EVERYTHING YOU NEED TO KNOW ABOUT BEING A CARRIER

You've just learned that you are a carrier for spinal muscular atrophy or SMA. You should know that being a carrier does not mean you are sick. In fact, you may not have found out that you are a carrier without being tested for SMA. Being a carrier means that one copy of your gene that can cause SMA has a change that keeps it from working right. You have a second copy of the SMA gene that is working as usual.

HEALTH NOTES



For more information

Visit kp.org/health

- Read about Spinal Muscular Atrophy in the Health Encyclopedia

Healthy living classes

- To register call Monday-Friday, 8:30 a.m.-5 p.m., **(301) 816-6565** or **(800) 444-6696**. For TTY, dial **711**.

Call the Kaiser Permanente Genetics Department

- **(301) 702-5250**

Call Kaiser Permanente Medical Advice

- Talk to your medical advice nurse 24 hours a day, 7 days a week by calling **(703) 359-7878** or **(800) 777-7904**. For TTY, dial **711**.

What is a gene and what does it do?

Genes are our body's instructions that decide our physical traits, such as blood type and hair color. They are found in chromosomes, which are in most of the cells in our body. We get one set of chromosomes from each of our parents. Our chromosomes, and the thousands of genes found on each of them, come in pairs.

The SMA gene pair, called SMN1, tells our bodies how to make a protein called survival motor neuron protein that helps our muscles work the right way. If a person has one copy of the SMA gene that does not work right, he or she is called a carrier of SMA. As long as a person has one working copy of this gene, the body has enough of SMN1 to do its job, so that person does not have and will never have SMA.

What is spinal muscular atrophy?

When a baby gets two non-working copies of the SMA gene from their parents, the nerves in the spinal cord and the brain stem start to break down and not work. This can cause:

- poor weight gain,
- trouble sleeping,
- pneumonia,
- curved spine,
- trouble walking or not being able to walk at all, and
- joint problems.

Signs of SMA can start in unborn babies or in adults. The earlier the disease starts the worse it is. In the worst cases of SMA, children die of lung failure – their muscles are not strong enough to help them breathe. When SMA symptoms start in adulthood, lifespan is normal.

How does one become a carrier?

Since all of our genes come from our parents, people who are SMA carriers have received 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 for spinal muscular atrophy?

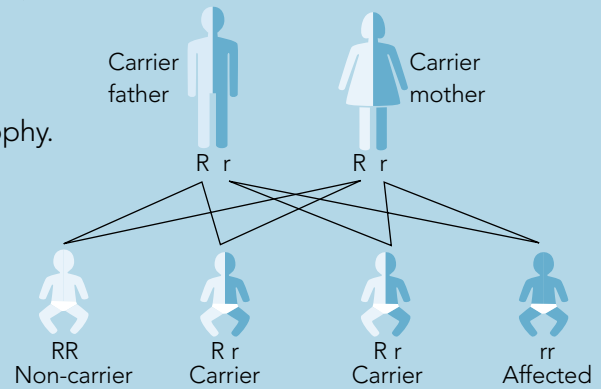
Yes. SMA carriers are found in all races and ethnic groups. Carrier rates vary from 1 in 16 people of Iranian descent to 1 in 83 people of Hispanic descent. The average chance of being a carrier for SMA is 1 in 50.

Your chance of passing down SMA

When both parents are SMA carriers there is:

- A 25% or 1 in 4 chance that the baby is not a SMA carrier and does not have spinal muscular atrophy.
- A 50% or 1 in 2 chance that the baby is a SMA carrier just like his or her parents.
- A 25% or 1 in 4 chance that the baby has spinal muscular atrophy.

R = working copy of gene
r = non-working copy of gene



Is there a cure for spinal muscular atrophy?

Though research is ongoing, there is nothing that can be done to cure the disease at this time. People with SMA can get help managing and treating their health care needs.

Can being a carrier lead to having spinal muscular atrophy?

No. Carriers of SMA will never get SMA because they have one working copy of the SMN1 gene, so their bodies make enough of the protein to be healthy.

Can my children have spinal muscular atrophy?

Since children get half of their traits, like eye and hair color, from their mother and half from their father, the answer to this question depends on whether your partner is a carrier.

If your partner is not a SMA carrier, the chance of your children having spinal muscular atrophy is very small but is not zero (because genetic tests cannot detect *all* carriers). With each pregnancy, you will have a 50% chance of having a child who is a SMA carrier just like you and a 50% chance of having a child who is not a SMA carrier. None of these children will have SMA.

If your partner is also a SMA carrier, then there are three possible pregnancy outcomes. One is the chance of having a child with spinal muscular atrophy (see diagram above).

To find out the chance of having a baby with spinal muscular atrophy, your partner should be tested to see if he or she is also a carrier for SMA. The exactness of this test result varies based on ethnic background.

The information presented here is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent medical problems, or if you have further questions, please consult your doctor or member of your health care team.

What choices do I have if my partner is also a carrier for spinal muscular atrophy?

There are a few choices you can make if both you and your partner are carriers. To find out before birth if a baby has SMA, a test can be done 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 make informed choices about keeping or ending the pregnancy. If the decision is to keep the pregnancy, parents can be prepared. They can learn more about SMA before the baby is born. They may want to reach out to other parents who have a baby with SMA. They can find out about current research being done to find a cure.

Before pregnancy, there is an option called in vitro fertilization with pre-implantation genetic diagnosis (IVF with PGD). In IVF with PGD, several of your eggs are fertilized outside the uterus. The embryos are then tested for the SMA genes. Only an embryo without the SMA gene is implanted into the uterus.

Resources

For more information on spinal muscular atrophy visit:

- Muscular Dystrophy Association
mda.org/disease/spinal-muscular-atrophy

For more information on getting involved with SMA research and support visit:

- Spinal Muscular Atrophy Foundation
smafoundation.org
- Cure SMA
curesma.org