KAISER PERMANENTE®

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 (SMA). You should know that being a carrier **does not** mean that 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 well.

Resources

If you have more questions, ask your health care team for a referral to a genetic counselor and visit **kp.org/scal/genetics**.

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**

What is a gene and what does it do?

Genes 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 bodies. 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 spinal muscular atrophy gene pair, called SMN, tells the body how to make a protein called survival motor neuron protein that helps 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. With one working copy of this gene, the body has enough of SMN 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 nonworking copies of the SMA gene from his or her parents, the nerves in the spinal cord and the brainstem 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
- 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 because their muscles are not strong enough to help them breathe. When SMA symptoms start in adulthood, lifespan is normal.

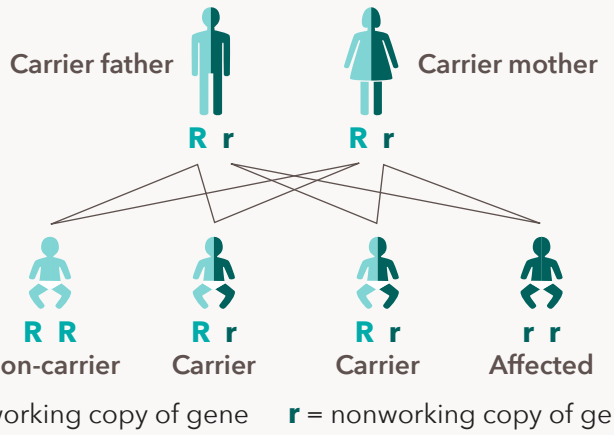
How does one become a carrier?

Since genes come from our parents, people who are carriers have received their nonworking gene from one of their parents. This means that daughters and sons of a carrier have a 50% chance of being a carrier.

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



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.

Is there a cure for spinal muscular atrophy?

Though research is ongoing, currently there is no cure for the disease. 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 SMN gene, which gives their bodies enough of the protein to be healthy.

Will 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 depends on whether your partner is a carrier.

If your partner is not an SMA carrier, the chance of your children having spinal muscular atrophy is very small, but it is not zero (because genetic tests can't detect **all** carriers). With each pregnancy, you will have a 50% chance of having a child who is an SMA carrier like you and a 50% chance of having a child who is not an SMA carrier. These children will **not** have SMA.

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

To find out the chance of having a baby with spinal muscular atrophy, your partner should be tested to see whether he or she is also a carrier for SMA. Test results may vary based on your ethnic background.

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

You have a few choices if you are both carriers. To find out before birth whether a baby has SMA, you can have a test as early as the 10th week of pregnancy. If the results are normal, you can be reassured. If the results show that the baby will be affected, you can make informed choices about keeping or ending the pregnancy. If you decide to keep the pregnancy, you can be prepared. You can learn more about SMA before the baby is born. You may want to reach out to other parents who have a baby with SMA. You can find out about current research being done to find a cure or learn about possible treatment options.

Before pregnancy, there is an option called in vitro fertilization with preimplantation 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.