KAISER PERMANENTE®

INFORMATION SHEET

Spinal Muscular Atrophy

Positive Carrier Screening Result

You have recently learned that you are a spinal muscular atrophy (SMA) carrier. Here is important information about being an SMA carrier.

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

Why would you not test your partner?

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

What happens if my partner is an SMA carrier?

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

Spinal muscular atrophy is an inherited disease that affects the nerves and muscles. Nerves that control the muscles slowly stop working. This leads to muscles that get weaker over time. A person with SMA may be unable to sit or walk and can develop trouble breathing and swallowing. Babies with SMA may have poor head control and very little body movement.

Signs of SMA can sometimes start before birth. The most common form of SMA starts in infancy and leads to death in early childhood. There are milder forms that start in teens or young adults and usually have a normal lifespan.

There is no cure for spinal muscular atrophy at this time, but research is being done to find ways to keep people with SMA as healthy as possible. Early diagnosis and treatment can help. Medications can be used to treat SMA to a limited extent.

Spinal Muscular Atrophy Carrier Screening

Anyone can be an SMA carrier, even if there is no history of SMA in the family. The chance to be an SMA carrier depends on a person's ancestry or ethnic background. Carrier screening detects most SMA carriers.

Approximate carrier rates by ancestry or ethnic background:

- Caucasian: 1 in 47 (about 2%)
- Asian: 1 in 59 (about 2%)
- Jewish: 1 in 67 (less than 2%)
- Hispanic: 1 in 68 (less than 2%)
- African (Black): 1 in 72 (less than 2%)
- Mixed ancestry/other: Varies (1-2%)

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

Limits of carrier screening:

- A small number of carriers have changes in the SMA gene that cannot be found by routine testing. This can lead to an apparently normal result in someone who really is a carrier.

- Very rarely a baby can have a new change in the SMA gene that is not inherited from either of the parents.

Newborn Testing for Spinal Muscular Atrophy

Testing can be requested soon after birth for babies who are at risk to develop SMA. An early diagnosis can allow medical care to start as soon as possible. In California, all babies are screened for SMA in the first few days after birth. When screening finds a baby with SMA, more testing will be done to confirm the diagnosis and guide medical care.

Limits of newborn screening:

- Newborn screening misses a small number of babies with SMA.
- In rare cases, a positive result can happen in a baby without SMA.