

Spinal muscular atrophy (SMA)

NEWBORN SCREENING FINDINGS

The state laboratory performs newborn screening on tiny samples of blood taken from a baby's heel after birth. The screening test for SMA looks for changes in the DNA of the baby's SMA gene. This baby had an abnormal newborn screen for SMA because **there was a missing piece, or deletion, in both copies of the SMA gene**. This means this baby very likely has a form of SMA.

ABOUT THE CONDITION

SMA is a treatable disorder that affects the nerves in the spinal cord and their ability to send signals to the muscles to tell them how to work. When these nerves don't work, muscles can't do their job and become very weak. SMA is caused by a missing or altered version of the gene known as SMN1. Babies with SMA usually receive two copies of this altered gene—one from each parent. About one in every 15,000 babies in the United States is born with SMA.

SIGNS/SYMPTOMS

There are four different types of SMA that are based on the age that symptoms begin and the severity of the condition. Depending on the type of SMA, signs may first appear within the first few months of life, or not until late adulthood, and may include trouble breathing or swallowing, not meeting motor milestones, or muscle weakness.

TREATMENT

The pediatric neurologist will help decide whether treatment needs to start right away, or whether the baby will be closely monitored for a time. Treatment may include a one-time gene therapy or, medicine that is delivered by injection on a regular basis; other types of treatment are also being developed. If treatment is recommended, it should be initiated right away, since babies with SMA tend to have better outcomes with early treatment.

NEXT STEPS

The newborn screening test is good at finding babies with SMA, but can't tell which babies will have a severe or mild form. The baby should be referred immediately to a pediatric neurologist with experience treating SMA. They will be able to explain SMA in more detail, order the additional testing needed, and start any necessary treatments. The baby does not need any special diet or treatments while waiting to be seen by Seattle Children's Hospital or Mary Bridge Children's Hospital.

For more information about SMA, visit:

- www.curesma.org
- www.babysfirsttest.org/newborn-screening/conditions/spinal-muscular-atrophy
- www.ghr.nlm.nih.gov/condition/spinal-muscular-atrophy