



Rhode Island Newborn Screening Program

Family Fact Sheet: My baby had an abnormal SMA newborn screening test

What is SMA Newborn Screening? “SMA newborn screening” means that doctors first test newborn infants for Spinal Muscular Atrophy (SMA), a genetic (inherited) condition. “Screenings” are first tests to decide if additional tests are needed. Knowing if your baby has SMA as soon as possible is important so that treatment can begin right away to help reduce health risks and improve overall health. SMA is a serious condition that progresses over time. Early detection and treatment can improve health outcomes.

How can SMA affect a person’s health? SMA affects the nerves located in the spinal cord. These nerves control how muscles throughout the body work. Without treatment, muscles can become very weak; people with SMA may have difficulty walking, eating, and breathing.

What does my baby’s positive SMA newborn screening test result mean? A positive newborn screening test result means that an infant is at higher risk of having SMA. To confirm or know for certain if your baby has SMA, doctors will order a special blood test and provide a physical evaluation.

Where do I get the special testing for my baby? Your baby’s healthcare provider has a list of specialists who can order this test for your baby.

Do I need to do anything to prepare my baby for the special testing? No.

How will I know the results of the special testing? Your baby’s healthcare provider or the specialist will contact you with the results.

What do I do while I wait for the test results? There is nothing special that you need to do. Just follow instructions from your baby’s healthcare provider. Just as you would do for any newborn, you will want to keep your newborn baby from being in contact with people who are known to have a contagious illness. Also, keep your newborn out of crowded environments.

When I have the results of my baby’s special testing, will I know if my baby has SMA? Yes.

Where can I get more information? The best source of information about your baby’s results and your baby’s condition is your baby’s healthcare provider. Information from your baby’s healthcare provider will be more helpful to you than general information. There is a website with reliable information about SMA that may be useful for families if they have a child who has been diagnosed with SMA. If you choose to look at this website, please keep in mind that your baby does not yet (and may never) carry a diagnosis of SMA. In that case, the SMA information may not apply to your baby. To learn more, visit: <https://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy>

This fact sheet provides general guidelines. Please consult with your baby’s doctor to discuss your baby’s clinical treatment plan and options.