Spinal Muscular Atrophy (SMA) Test

Dear Parent,

Shortly after your baby was born, a blood sample was obtained from his/her heel to test for multiple disorders that can affect your baby’s health. One of these tests is called the SMA test.

WHAT DOES THIS TEST LOOK FOR? The SMA test looks for a serious disease called Spinal Muscular Atrophy (SMA). SMA is caused by abnormal changes in the gene called SMN1. This test detects the most common abnormal change. In patients with abnormal genetic changes in SMN1, the nerves that tell muscles to move our body do not function well and may eventually stop working. This results in worsening weakness in the arms and legs as well as breathing and swallowing muscles.

WHAT NEEDS TO BE DONE IF THE TEST IS POSITIVE? The SMA test is only a screening test done at birth, and another blood test is needed. The purpose of this second test is to confirm the results of the first test as well as test another gene called SMN2. The information about SMN2 can help estimate how severe SMA disease will be if SMA is confirmed.

WHEN WILL I KNOW THE RESULTS OF THE SECOND BLOOD TEST? Once blood has been drawn and sent out for the second test, it takes several days for the laboratory to complete and report back results to your doctor. Your baby’s doctor will contact you with the results.

After your baby’s test results return, you will be referred to an SMA doctor who will discuss these results with you and any required next steps.

CAN TEST RESULTS BE UNCERTAIN? The screening test done at birth is expected to detect about 95% of all patients with SMA. It is important to have the second test done to confirm any screening results.

WHY SHOULD WE TEST FOR SMA BEFORE MY BABY HAS SYMPTOMS? Effective treatments for SMA are now available. Earlier treatment is more likely to result in less weakness, more developmental milestones, and better health outcomes for your baby.

WHERE CAN I FIND SUPPORT AND ADDITIONAL INFORMATION ABOUT SMA?
Cure SMA – www.curesma.org
Muscular Dystrophy Association - https://www.mda.org/disease/spinal-muscular-atrophy